# School experiences of children with rare genetic conditions associated with high risk for neurodevelopmental difficulties

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**Thesis Summary**

Neurodevelopmental copy number variants (ND CNVs) are rare genetic variants robustly associated with increased risk for neurodevelopmental conditions. The associated phenotype of ND CNVs is heterogeneous. This thesis explored an important childhood environmental experience, specifically school experiences, to elucidate whether such exposures could contribute to our understanding of phenotypic heterogeneity in this group.

Children with ND CNVs were at increased risk for peer bullying compared to their unaffected siblings. Neurodevelopmental risk factors and increased age were positively associated with bullying experiences in both groups, however those with a ND CNV were likely to be at higher risk for reasons beyond their increased propensity for neurodevelopmental challenges.

Almost 25% of parents who had a child with a ND CNV were unsatisfied with the support they received at school. Children with behavioural problems were more likely to have unsatisfied parents, whereas parents of children who had secured educational support (as measured by whether they had an educational health care plan (EHCP)) were more likely satisfied. Qualitative data provided insight into the circumstances in which parents were unsatisfied. Unhelpful learning environments, difficulties accessing higher levels of statutory support, limited understanding of children's needs, limited resources, and inadequacy regarding the amount and type of support were implicated. Children meeting criteria for indicative autism spectrum disorder, intellectual disability or who had a *de novo* ND CNV were more likely to have an EHCP.

Finally, semi-structured interviews with mothers of children with a genetic condition associated with neurodevelopmental challenges provided in-depth understanding about the impacts of receiving educational support and the impacts and experiences of navigating 'the system' in order to obtain it.

This thesis contributes to our understanding about the environmental factors associated with phenotypic heterogeneity in children with ND CNVs.

## Contents

**Figures**

**Tables**

## Glossary of Abbreviations

| | |
|---|---|
| 22q11 DS | 22q11.2 Deletion Syndrome |
| ADHD | Attention deficit hyperactivity disorder |
| A-Level | Advanced Level |
| AS | Angelman Syndrome |
| ASD | Austism spectrum disorder |
| BFQ | Bullying and Friendship Interview Schedule Questionnaire |
| BH-FDR | Benjamini–Hochberg false discovery rate |
| CAPA | Child and Adolescent Psychiatric Assessment |
| CD | Conduct disorder |
| CNV | Copy Number Variant |
| CYP | Children and Young People |
| DCD | Developmental coordination disorder |
| DCDQ | Developmental Coordination Disorder Questionnaire |
| DfE | Department for Education |
| DfH | Department for Health |
| DiGEN | Dissecting the effects of genomic variants on neuro-behavioural dimensions in CNVs enriched for neuropsychiatric disorders study |
| DNA | Deoxyribonucleic acid |
| DPMCN | Division of Psychological Medicine and Clinical Neurosciences |
| DS | Down Syndrome |
| DSM-5 | Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition |
| EBD | Emotional and behavioural disorders |
| ECHO | The Experiences of CHildren with cOpy number variants study |
| EHCP | Education, Health and Care Plan |
| EP | Emotional problems |
| FA | Framework analysis |
| FSIQ | Full-scale IQ |
| GAD | Generalised anxiety disorder |
| GCSE | General Certificate of Secondary Education |
| GLMM | Generalised linear-mixed model |
| HCEC | House of Commons Education Committee |

| | |
|---|---|
| ID | Intellectual disability |
| IEP | Individualised Education Programme |
| IMAGINE-ID | The Intellectual Disability & Mental Health: Assessing the Genomic Impact on Neurodevelopment study |
| IQ | Intelligence quotient |
| KTP | Knowledge Transfer Project |
| LA | Local authority |
| LD | Learning disability |
| NAO | National Audit Office |
| ND CNV | Neurodevelopmental CNV |
| NHS | National Health Service |
| NI | Northern Ireland |
| ODD | Oppositional defiant disorder |
| ONS | Office for National Statistics |
| PE | Physical education |
| PE | Psychotic experiences |
| PIQ | Performance IQ |
| PWS | Prader-Willi Syndrome |
| RNA | Ribonucleic acid |
| RTA | Reflexive thematic analysis |
| SCQ | Social Communication Questionnaire |
| SDH | Social Determinants of Health |
| SDQ | Strengths and Difficulties Questionnaire |
| SEMH | Social, emotional and mental health |
| SEN | Special educational need |
| SENCO | Special Education Needs Coordinator |
| SEQ | School Experiences Questionnaire |
| SES | Socioeconomic status |
| TA | Teaching assistant |
| TA | Thematic analysis |
| UN | United Nations |
| UNESCO | The United Nations Educational, Scientific and Cultural Organization |
| UNICEF | United Nation's Childrens Fund |

| | |
|---|---|
| VIQ | Verbal IQ |
| WASI | Wechsler Abbreviated Scale of Intelligence |
| WGS | Whole genome sequencing |
| WHO | World Health Organisation |

# 1 Introduction

## 1.1 Chapter Overview

*"Schooling does matter greatly. Moreover, the benefits can be surprisingly long lasting" (Rutter, 1991).*

School is a major part in all children's lives, yet to the best of my knowledge, no research to date has explored the school experiences of children with neurodevelopmental conditions of genetic aetiology, specifically known as copy number variants. I argue that such investigation is important to better our understanding about what environmental factors influence the considerable phenotypic heterogeneity observed in this group. Such research will provide a building block for further exploration into these children's educational experiences, increase educators' vigilance for potential educational problems and empower parents when advocating for the educational support needed by their child when required. It is hoped that this will enable better identification and implementation of supportive educational environments and practices, thus helping to provide a vulnerable group of children with the best opportunities for later life.

Within this chapter, I will define copy number variants and describe their associated phenotype. I will discuss some of the reasons why phenotypic heterogeneity is observed in this group, as well as the challenges posed by such variance for children and their parents, as well as clinical professionals. I argue that these challenges will become more widely felt as the UK's envisioned healthcare system, which seeks to utilise patients' genomic data to provide personalised care, develops.

Next, I explain the UK education system and then provide more detailed background information relative to the explicit topic areas of this thesis: the social experiences of children with copy number variants at school, and the accessibility of special educational provision. I argue the case for exploring these experiences within this group specifically throughout. Finally, I present the specific aims of this thesis.

Some of the research undertaken and presented in this thesis benefitted from the insight of stakeholders via a steering group for a Knowledge Transfer Partnership (KTP) project between Cardiff University and Cerebra, which I managed alongside

this PhD. Within this chapter, I will provide further information about Cerebra, the KTP project, the coordination and facilitation of the steering group, and how stakeholder views were taken into consideration.

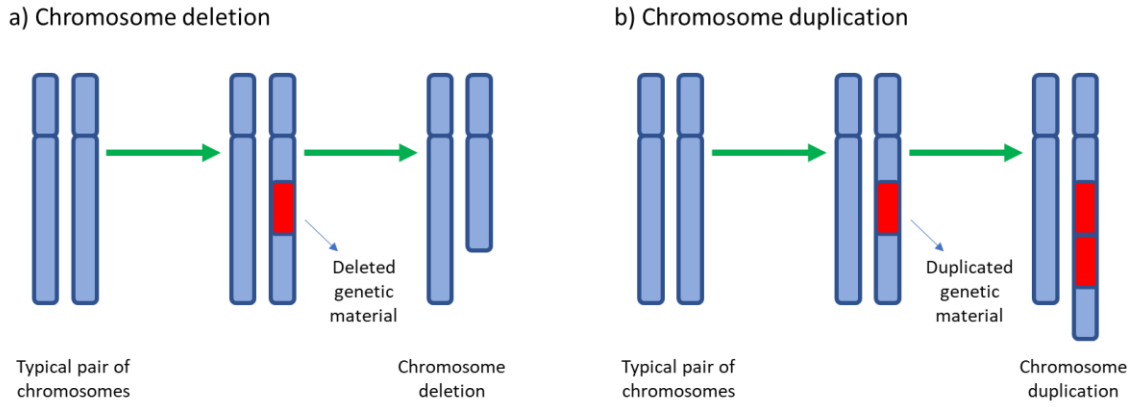
## 1.2 Genetic variation

In 2001, The Human Genome Project and Celera Genomics successfully sequenced the entire human genome (Lander et al., 2001; Venter et al., 2001), which provided scientists with the first 'reference genome' from which they could begin to ascertain the scope of genetic variation between individuals. Research indicates approximately 4 million genetic variants exist within the human genome, but that there are many more variants yet to be discovered (Marian, 2020).

### 1.2.1 Neurodevelopmental Copy Number Variants

Excluding gametes (i.e., egg and sperm cells), within each cell of a typical human genome, there are twenty-three pairs of chromosomes. Atypical chromosomes (for example, in number or in size) can lead to significant health and functional problems. A copy number variant (CNV) is a type of genomic structural variant whereby a loss (deletion) or gain (duplication) of genetic material (**Figure 1.1**), equal to or larger than 1000 base pairs (or 1 kilo base) occurs in one of the 46 chromosomes (Lee & Scherer, 2010).

*Figure 1.1 Cartoon depiction of deletion and duplication copy number variant formation.*




*a) the cartoon shows a segment of genetic material is missing (deleted) from one of the typical chromosomes within the pair b) the cartoon shows a segment of genetic material has been added (duplicated) to one of the typical chromosomes within the pair.*

CNVs were historically thought to be a definitive marker for disease, however they have since been found to occur frequently within healthy populations (Iafrate et al., 2004; Sebat et al., 2004). They make up approximately 10% of the human genome and contribute to population diversity (Zarrei et al., 2015). CNVs in specific locations within the genome can, however, confer risk for disease. Both common (those found in at least 1% of the population) and rare (those occurring in <1% of the population) CNVs increase risk for morbidity (Crawford et al., 2019; Fanciulli et al., 2010). Common CNVs have been associated with common illnesses, including autoimmune, infectious, cardiovascular and psychiatric conditions, although not all people with a particular common CNV will develop the associated illness. Rare CNVs are typically associated with greater risk of morbidity compared to common CNVs, but again, not all individuals with a rare CNV will notably exhibit any of the associated conditions. For example, 22q11.2 Deletion Syndrome (22q11 DS), also known as DiGeorge Syndrome or Velocardiofacial Syndrome is caused by a CNV whereby 0.7–3 million base pairs are missing on chromosome 22. With an estimated prevalence of 1 in every 3,000-6,000 live births, 22q11 DS is the most common chromosomal microdeletion syndrome (McDonald-McGinn et al., 2015). 22q11 DS confers risk for an array of health complications such as cardiovascular, autoimmune, palatal and psychiatric conditions (McDonald-McGinn et al., 2015),

however not all individuals will present with marked symptoms (McDonald-McGinn et al., 2001).

Children with rare CNVs which result in rare genomic disorders known to increase risk for neurodevelopmental conditions are the population group of focus in this thesis. These CNVs will hereon be referred to as 'neurodevelopmental CNVs' (ND CNVs).

## Neurodevelopmental conditions

As defined by the Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition (DSM-5) (American Psychiatric Association, 2013), neurodevelopmental conditions are a group of psychiatric conditions "characterized by developmental deficits that produce impairments of personal, social, academic, or occupational functioning". Conditions currently classed as a neurodevelopmental condition within DSM-5 include intellectual disability (ID), communication disorders, autism spectrum disorder (ASD), attention deficit hyperactivity disorder (ADHD), motor disorders and specific learning disorders. Although formally still classified as a mental health condition, schizophrenia is commonly regarded as a neurodevelopmental condition.

All neurodevelopmental conditions are associated with some form of neurocognitive deficit and atypical brain development (Thapar et al., 2017). They have an early onset, typically before children begin school. There is an overrepresentation of boys diagnosed with neurodevelopmental conditions compared to girls (May et al., 2019), indicating that girls could present with atypical symptomology, or be more adept at masking symptoms. Neurodevelopmental multimorbidity is common, meaning two or more neurodevelopmental conditions are frequently observed in the same individual (Hansen et al., 2018). Some researchers have argued that the high rates of multimorbidity suggest neurodevelopmental conditions do not represent distinct diagnostic categories, but symptoms of varying severity of one overarching condition (Kaplan et al., 2001).

### *a) Intellectual Disability*

According to DSM-5, ID is a lifelong condition, characterised by difficulties in intellectual and adaptive functioning which impact day to day life. Intellectual

functioning comprises skills like learning and problem solving, whereas adaptive functioning involves day to day functioning such as communication and living independently. ID affects roughly 1% of the global population (Patel et al., 2020), and can have genetic causes, or arise from a trauma caused by head injury, illness, or complications during birth. Environmental causes also include infections or other harmful exposures during pregnancy and early childhood. Onset must be within the early developmental period as opposed to in later life. A person's ID can be classified as mild, moderate, severe, or profound. This judgement is made by assessment by a clinician.

*b) Communication disorders*

Communication disorders represent difficulties with language and speech. In DSM-5, communication disorders fall under the following categories: language disorder, speech sound disorder, childhood-onset fluency disorder (i.e., stuttering), social communication disorder and unspecified communication disorder.

*c) Autism spectrum disorder*

Individuals with ASD experience persistent difficulties in communication and interaction with others. Such difficulties impact day-to-day functioning at school or work, as well as the person's social relationships. It is common for individuals to display repetitive behaviours, have a restricted set of interests and preference for routine. The 'spectrum' within ASD reflects the varying degree of impairment between individuals. For example, whilst some people can live independently, others will need life-long additional support and care. The incidence of ASD is estimated to be 1 in 100 children worldwide (Zeidan et al., 2022).

*d) Attention deficit hyperactivity disorder*

The average prevalence of ADHD in children is 5% (Sayal et al., 2018). ADHD is characterised by difficulties in sustaining attention and/or atypically high levels of hyperactivity-impulsivity. To satisfy diagnostic criteria for ADHD, these difficulties must be persistent and pervasive, causing impairment across multiple settings (e.g.,

in school and at home). Presentation can attenuate with age, however symptomology is still observable in adults (Mannuzza & Klein, 2000).

DSM-5 defines ADHD as a child-onset condition, with symptom onset being evident before the age of 12. However researchers (Moffitt et al., 2015) have questioned this notion after finding a proportion of adults who met criteria for ADHD in adulthood, but did not display ADHD symptoms as children. Subsequent evidence (Cooper et al., 2018) found support for what has been termed ‘genuine late-onset ADHD’.

*e) Motor disorders*

Motor disorders listed in DSM-5 include developmental coordination disorder (DCD), stereotypic movement disorder, Tourette syndrome and tic disorders. Individuals experience difficulties in controlling movements and therefore often display involuntary actions, or experience challenges when trying to perform actions in the way they intend.

*f) Specific learning disorders*

Specific learning disorders can be mild, moderate or severe. They comprise difficulties in specific areas of functioning, including reading, writing and maths which often cause difficulty at school, and sometimes with peers. Dyslexia, dysgraphia and dyscalculia are examples of specific learning disorders. Approximately 15% of children have a specific learning disorder (American Psychiatric Association, 2013).

*g) Schizophrenia*

Schizophrenia is a psychiatric condition characterised by psychotic symptoms (i.e., hallucinations and delusions), negative symptoms (e.g., apathy), and in which cognitive impairments are common. Schizophrenia affects approximately 1% of the population (McGrath et al., 2004). Diagnoses are normally given during late adolescence or early adulthood. Schizophrenia is not formally classified as a neurodevelopmental condition, but there is evidence to support a neurodevelopmental hypothesis (Murray & Lewis, 1987; Weinberger, 1986) which posits that subtle behavioural, motor and cognitive symptoms in childhood

premanifest more obvious psychotic and cognitive symptoms. For example, Reichenberg et al. (2010) found that adults who met criteria for schizophrenia had lower childhood intelligence quotient (IQ) scores compared to the childhood IQ scores of adults who met criteria for depression and controls. Furthermore, factors such as foetal exposure to infection and childhood trauma, both of which can cause atypical brain development have been linked with schizophrenia (Brown & Derkits, 2010; Varese et al., 2012).

ND CNVs confer risk for the neurodevelopmental conditions described above, namely ID, ADHD, ASD and schizophrenia (Coe et al., 2014; Levinson et al., 2011; Rees et al., 2014). Examples of ND CNVs and their associated named genetic syndromes include the already mentioned 22q11 DS, as well as 17p11.2 Deletion Syndrome (Smith-Magenis Syndrome), and 7q11.23 Deletion Syndrome (Williams-Beuren Syndrome or Williams Syndrome).

### 1.2.2 Phenotypes

An individual's phenotype relates to the characteristic manifestation of their genetic makeup, for example their appearance or the symptoms of illness they present with (Schulze & McMahon, 2004). Children with ND CNVs have complex phenotypes. ND CNVs have pleiotropic effects meaning individuals are often impacted in many domains. ND CNVs have been associated with physical problems, such as congenital heart disease, facial dysmorphology, compromised immune functioning, obesity and seizures (Chawner et al., 2021; Eaton et al., 2019). Additional developmental concerns include speech and language delay (Sahoo et al., 2011) and impaired motor functioning (Cunningham et al., 2021). Children also display increased prevalence of psychiatric and neurodevelopmental conditions such as anxiety and depression (Chawner et al., 2019; Wolstencroft et al., 2022). Poor sleep functioning is also observed in this group (Agar et al., 2021, 2022; Chawner et al., 2023; Moulding et al., 2020).

Phenotypic heterogeneity

As touched upon earlier, although ND CNVs confer risk for certain difficulties, there is a great degree of phenotypic heterogeneity between individuals (Chawner et al., 2019). Heterogeneity has been observed between individuals with the same ND CNV, and even between biological family members (Chawner et al., 2019; McDonald-Mcginn et al., 2001). Whilst some children experience substantial impacts in one or more of the domains listed above, others will remain relatively unaffected. In fact, some parents only discover they carry a ND CNV after their child who has inherited the CNV from them presents with severe symptoms which prompt clinical genetic testing for the child and their family members (McDonald-McGinn et al., 2001). Interestingly, evidence does suggest that individuals within the general population (who may or may not be aware they carry a ND CNV) may be impacted somewhat, even if the effect is not overtly noticeable. For example, 'healthy' individuals living with ND CNVs perform lower on cognitive tests, achieve lower levels of educational attainment, have a lower household income and exhibit poorer physical and mental health (Crawford et al., 2019; Kendall et al., 2017, 2019). Given that individuals can unknowingly live with these conditions, the recorded clinical rates of ND CNVs are thought to be an underestimation of their true prevalence. Reports from a population based study indicate that ND CNVs collectively occur in approximately 1.12% of the population (Kendall et al., 2019).

The 'penetrance' and 'expressivity' of ND CNVs impact phenotypic heterogeneity (Kingdom & Wright, 2022). Penetrance and expressivity sit on a continuum which depicts the degree of influence the genomic variant has on phenotype. If an associated phenotype for a ND CNV is seen in 100% of people who carry the variant, it is said to have 'complete penetrance'. Any less than 100% prevalence and the ND CNV has 'incomplete penetrance'. Expressivity is defined as the severity of the associated phenotype. ND CNVs have both incomplete penetrance and variable expressivity which in part explains why children inherit pathogenic ND CNVs from their seemingly unaffected parents and why ND CNVs are found in ostensibly healthy population samples.

The causes of phenotypic heterogeneity are not fully understood. Factors believed to impact phenotype include ND CNV specific factors, namely the specific ND CNV

(e.g., 22q11 DS vs Williams syndrome), the size of the ND CNV, the type of CNV (deletion vs duplication), and CNV inheritance status.

ND CNVs which are large and span more genes, particularly those expressed in the brain, have been associated with more severe phenotypes compared to smaller ND CNVs (Guyatt et al., 2018; Martin et al., 2017). ND CNVs which involve the loss of genetic material (deletions) may be more harmful compared to duplications (Fernandez et al., 2010). The specific ND CNV an individual has has also been implicated (Chawner et al., 2019) however the contribution of CNV type may only be minor, explaining only 5-20% of the variance in behavioural and cognitive outcomes between different ND CNVS. ND CNVs which are not inherited from a parent are known as *'de novo'* and have been suggested as more likely pathogenic (Lee & Scherer, 2010). The rationale driving this assumption is that parents carrying a ND CNV cannot be so affected that their fecundity is too compromised to pass the variant onto future generations. Individuals with *de novo* ND CNVs which have severe effects on phenotypic traits which affect their likelihood of reproduction (e.g., early mortality, ID) are less likely to reproduce and have offspring who inherit the variant. This partly explains why individuals with ND CNVs who have more severe phenotypes (e.g., 22q11 DS) more often carry a *de novo* variant compared to those with less severe phenotypes (Rees et al., 2011). Wolstencroft et al. (2022) found children with a *de novo* ND CNV displayed more impairments in intellectual functioning, however they reported higher rates of specific neurodevelopmental and mental health conditions (ADHD, ASD, oppositional defiant disorder (ODD), conduct disorder (CD) and anxiety) in those with an inherited ND CNV. Cunningham et al. (2022) also reported increased risk for emotional and behavioural problems in those with an inherited ND CNV.

Beyond ND CNV specific factors, additional genetic variation in other areas of the genome likely influences phenotype. Work by Girirajan et al. (2010) suggests that additional large CNVs within an individual's genome (a 'second hit') contribute to more severe phenotypes. Additional CNVs could also have protective effects on phenotype. For example, Beckmann et al. (2007) suggested the impacts of a deleterious ND CNV could be compensated for if the individual also carries a duplication in the same region on the homologous chromosome. Carelle-Calmels et

al. (2009) found supporting evidence for such a compensatory effect, noting differences in the clinical presentation between a biological father and daughter, although case study reports cannot be generalised. The impact of additional common genetic variants (those occurring in at least 1% of people) have also been implicated in phenotypic variation (Niemi et al., 2018).

Epigenetic research has uncovered further potential explanations for variable phenotypic presentation. Epigenetics is a broad term, which in general refers to the study of modifications to deoxyribonucleic acid (DNA) that do not alter the genetic code itself, but that may result in altered regulation of gene expression, sometimes in response to environmental exposures. Gene expression is the process whereby the information encoded in DNA is transcribed into ribonucleic acid (RNA), and the information encoded in RNA is translated into protein. Most human cells are diploid. That is, they contain maternally and paternally inherited genes. Every gene comprises a section of an individual's DNA which in turn instructs which proteins are produced by the cell housing the gene. Genes that are 'turned on' (i.e., expressed) produce the proteins instructed for, whereas genes which are 'turned off' (i.e., silenced) do not produce the proteins which would otherwise be instructed for.

'Genomic imprinting' is an epigenetic function whereby either the maternal or paternal copy of a gene within a cell is expressed only (i.e., both copies are not expressed). Genomic imprinting has lifetime effects and is thought to impact ND CNV phenotypes (Isles, 2022). For example, Angelman (AS) and Prader-Willi syndromes (PWS) are two neurodevelopmental syndromes caused by either a maternal or paternal deletion in the region of chromosome 15q11-q13 respectively (Knoll et al., 1989). This region has a complex pattern of imprinting, some genes only being expressed when they are inherited maternally, others only expressed when inherited paternally. AS is typically caused by a maternally inherited deletion which results in the loss of expression of the set of genes only expressed on the maternal chromosome. Conversely, PWS is caused by a paternally inherited deletion in the same region leading to the loss of expression of the genes that are only expressed from the paternal chromosome. Although the same chromosomal region is deleted in AS and PWS, the phenotypic presentation of the conditions differs. AS is associated with a severe neurodevelopmental phenotype, including ID, whereby children have

extremely limited verbal ability, whereas children with PWS typically show a milder form of learning disability (Buiting, 2010). This phenotypic heterogeneity is thought to be partly explained by the difference in inherited gene expression.

*Environmental impact on phenotypic expression*

The environmental experiences of children with ND CNVs and how these experiences impact their phenotype are not well understood. Currently, there is evidence implicating greater socio-economic deprivation as an environmental risk factor for behavioural difficulties in children with ND CNVs (Wolstencroft et al., 2022). Additionally, Moreno-De-Luca et al. (2015) found that cognitive and social functioning in children with 16p11.2 deletion syndrome is influenced by their parents' performance on the same measures, indicating that family background could contribute to phenotypic outcome. However, considering cognitive and social functioning are heritable traits, such findings are likely to be explained by both environmental as well as genetic influences. Our knowledge of environmental experiences and how these exposures could influence phenotypic outcomes is otherwise limited.

*Challenges posed by phenotypic heterogeneity*

Phenotypic heterogeneity presents challenges for clinicians and families of children with ND CNVs, as well as for the child themselves. Firstly, with such variability between children, clinicians are challenged in anticipating the best ways to manage each child's clinical care. This challenge is compounded by the rarity of ND CNVs which results in a general limited awareness of these conditions and their associated difficulties (Rizzo et al., 2020). With incomplete understanding about what the presence of a ND CNV will mean for a child and their family, clinicians are limited in the information they can provide parents of newly diagnosed children about what the future will bring. On occasion, certain information about a child's condition is not shared with parents, and this is particularly true of the child's risk for psychiatric conditions (van den Bree et al., 2013). Reasons for non-disclosure as reported by genetic counsellors include not wanting to overwhelm parents with information deemed to be more relevant in later life and parents' emotional state at the time of genetic diagnosis (Martin et al., 2012), both of which are important considerations.

However, additional considerations included counsellors' own discomfort in discussing psychiatric manifestations of the condition, which in part stemmed from their limited knowledge about how to identify the early signs of these conditions, treatment and prevention, and the limited literature addressing these issues (Martin et al., 2012).

Access to expert information is undoubtedly helpful to those who see a genetic counsellor after diagnosis (Macleod et al., 2002) and receiving a genetic diagnosis can validate parents' concerns about their child's challenges (Makela et al., 2009). However, parents can also feel great levels of uncertainty and a lack of control after receiving a genetic diagnosis for their child and after genetic counselling (Goodwin et al., 2017; Lipinski et al., 2006). Uncertainty is associated with poor mental health in mothers of children with chronic conditions (Holm et al., 2008) and mothers of children with ND CNVs are indeed at increased risk for psychological problems (Baker et al., 2021; Fitzgerald & Gallagher, 2022; Griffith et al., 2011; Niarchou et al., 2022). However, I am not aware of any study to date to have researched the link between uncertainty and psychological distress in this group specifically.

The lack of knowledge and confidence amongst clinicians and other professionals about how to best predict and manage children's support needs can leave children with ND CNVs and their family unsupported (Čagalj et al., 2018). Primary caregivers often adopt the role of care-coordinator for individuals with rare genetic conditions which can affect their own psychosocial health and functioning (Simpson et al., 2021). With parents often acting as care-coordinator, children are likely dependent not only on their parents' wherewithal to detect the early signs of health problems, but also their ability to access services accordingly. This is a great challenge for parents, and leaves children vulnerable to missing out on the support they need.

As we move deeper into the 'genomic era' of medicine (Guttmacher & Collins, 2003) and aspirations to increase the utility of whole genome sequencing (WGS) in routine healthcare become reality (discussed below), it is likely that the detection rates of ND CNVs will rise and such challenges may become more widely experienced.

## 1.3 The Future of Genomic Medicine

Our increasing knowledge of the human genome has presented great opportunities for medicine by providing better understanding about the biological mechanisms by which diseases occur. Researchers and clinicians increasingly utilise such information to inform targeted treatments and examples of successful gene therapies for certain illnesses already exist (Daley, 2021). Knowledge is also used to advise patients about treatment options, for example for patients diagnosed with genetic forms of breast cancer via genetic counselling.

Nations worldwide are seizing the opportunities genomics can bring to healthcare (Manolio et al., 2015). The UK Government has recently outlined its ambitions for the future of genomic medicine in the UK in its report, 'Genome UK: The Future of Healthcare' (HM Government, 2020), in which they state the UK will have 'the most advanced genomic healthcare system in the world'. WGS will be offered as part of routine care for certain groups, including critically ill children with the aim to improve diagnostic rates, shortening patients' often lengthy 'diagnostic odyssey'. Furthermore, WGS will facilitate individualised and preventative support to improve the population's mental and physical health. Other UK nations have echoed these ambitions within their own devolved strategies. For example, the Welsh Government will create an 'internationally competitive environment for genetics and genomics to improve health and healthcare' (Welsh Government, 2017).

The implementation of these strategies will likely increase the number of children diagnosed with a ND CNV in the UK and globally. Whilst this has great benefits for the potential to inform and facilitate access to support and treatment, this potential can only be recognised fully if we have a comprehensive understanding of what a diagnosis of a given ND CNV means for the child and their family. Our ability to diagnose must go hand in hand with our knowledge of the implications of that diagnosis, otherwise we will continue to see parents experiencing uncertainty, psychological distress, and lack of support for them and their child.

Understanding the implications of a diagnosis comes with better understanding of the similarities and differences in the health outcomes of children with ND CNVs and the factors which contribute to those outcomes. Currently, there are still major gaps

in our knowledge of the factors contributing to phenotypic heterogeneity. Governments recognise that our health is not just determined by our genome, but that environmental factors also play a crucial role, and that the evolution of healthcare systems must incorporate such influences holistically to mitigate against morbidity (HM Government, 2020). Guttmacher and Collins (2003) emphasise that so long as it is technically difficult and ethically questionable to alter human genomes, the focus of our efforts to translate genomic knowledge into health improvements should be the modification of our environment. Thus, attention should be given to the potential environmental compounders of adverse outcomes in children with ND CNVs and their families, as well as strategies to prevent the impact of these environmental experiences. Such an approach will not only improve the health and quality of life of this group, but preventative approaches such as this will go some way in lightening the burden on the National Health Service (NHS), which is already struggling to keep up with increasing demand (Ham, 2020; Pencheon, 2015).

### 1.4 School Experiences

Experiences at school are an important environmental exposure in children's lives which 'greatly matter' for their long-term outcomes (Rutter, 1991). School is likely to have such a considerable impact on later life given the large amount of time young people spend there. In the UK, children are typically in full-time education from 5-16 years old. Furthermore, children and young people go through substantial developmental change during these years, especially during adolescence, and are particularly vulnerable to their environment at these key developmental stages (Sisk & Gee, 2022) .

Despite this, to my knowledge the only study to have investigated the school experiences of children with a ND CNV specifically is that of Whittington et al (2004). They found children with PWS and those with alternative causes for learning disability (LD) showed underachievement compared to their predicted level of achievement, but those with LD showed greater levels of underachievement in measures of reading and spelling compared to the PWS group. Underachievement was similar between the groups in mathematics. Poorer social functioning predicted underachievement in the PWS group but not in the LD group, highlighting the importance of social skills for educational attainment in those with PWS. Time spent

in a special school also predicted underachievement in the PWS group only. The authors suggested that children with PWS in special schools may be less likely to be taught certain skills compared to in mainstream schools and hypothesised that their needs may be less severe than other children in their special class and therefore they may get less academic attention. Alternatively, their presentation on other measures (e.g., behaviour), or the special school placement itself, could bias teachers against children's academic capability.

Further research which explored parents' concerns about their child's genetic condition indicate that parents have worries about their child at school. Concerns included how their genetic condition might impact their academic performance, their psycho-social wellbeing (e.g., feeling different from their peers and bullying), the ability of their child's school to manage the health implications of their condition and whether the school had the resources to support their child (Gallo et al., 2008). This study did not include parents of children with an ND CNV specifically so direct inferences cannot be made to the parents of this group, however it is not unreasonable to assume that such concerns are shared amongst parents of children who have any additional need that might make school life more challenging. Indeed, parents of children with ADHD and ASD have expressed similar concerns (Lee et al., 2008). Furthermore, Lee et al. (2005) found that teachers struggled to identify the cognitive and behavioural features of children with 22q11.2 DS compared to those associated with Down syndrome (DS) and fragile X syndrome, suggesting that teachers of children with ND CNVs may not have always have the knowledge to appropriately manage the implications of the child's condition. The findings of Whittington et al. (2004) also imply that children with ND CNVs may not always meet their academic potential. Thus, the concerns of parents taking part in the study of Gallo et al. (2008) could be warranted in parents of children with a ND CNV.

### 1.4.1 Defining 'school experiences'

'School experiences' is a broad term used within this thesis to encapsulate the individual experiences children have during their time at school. Studies reporting on school experiences have explored an array of topics including children's academic achievement, school climate and their social relationships.

With such an expanse of possible experiences to explore, coupled with the dearth of research into those of children with ND CNVs specifically, the experiences researched and presented in this thesis were informed partly by my own interest into children's peer relationships, as well as the literature suggesting the pertinence of such relationships to children's later outcomes (discussed below). Secondly, my research focus was informed by the issues deemed of particular interest to parents of children with ND CNVs. This evaluation was made following my own discussions with parents via the Cerebra Steering Group (see below), as well as discussions between field team members of the Rare Genetic Research Group at Cardiff University and parents taking part in the group's research, which often focus on the level of support received by children at school. Thus, the experiences I set out to explore were children's experiences with their peers at school, namely their bullying experiences, and children's access to special educational needs (SEN) provision. During my conversations with parents, their wish for medical, educational, and other support services to see their child's needs holistically within the context of the family became apparent. This thesis therefore also partly explores the impact of the children's school experiences on their parents and family.

I will now present some further information about the KTP project I completed with Cardiff University and Cerebra and the associated steering group. Then I will provide an overview of the UK education system and background information related to the main topics of this thesis: access to SEN provision and bullying experiences at school.

## 1.5 Cardiff University and Cerebra KTP Project

From January 2019 to January 2022, I completed a KTP project with Cardiff University and Cerebra, which was joint funded by Cerebra, Welsh Government and Innovate UK. Professor Marianne van den Bree was the Lead Academic for the project.

KTP projects link a business or third sector organisation with a research institution to deliver innovation projects. Cerebra are a charity supporting children with brain conditions and their families. They fund research and then translate findings into accessible information resources to empower families with improved knowledge

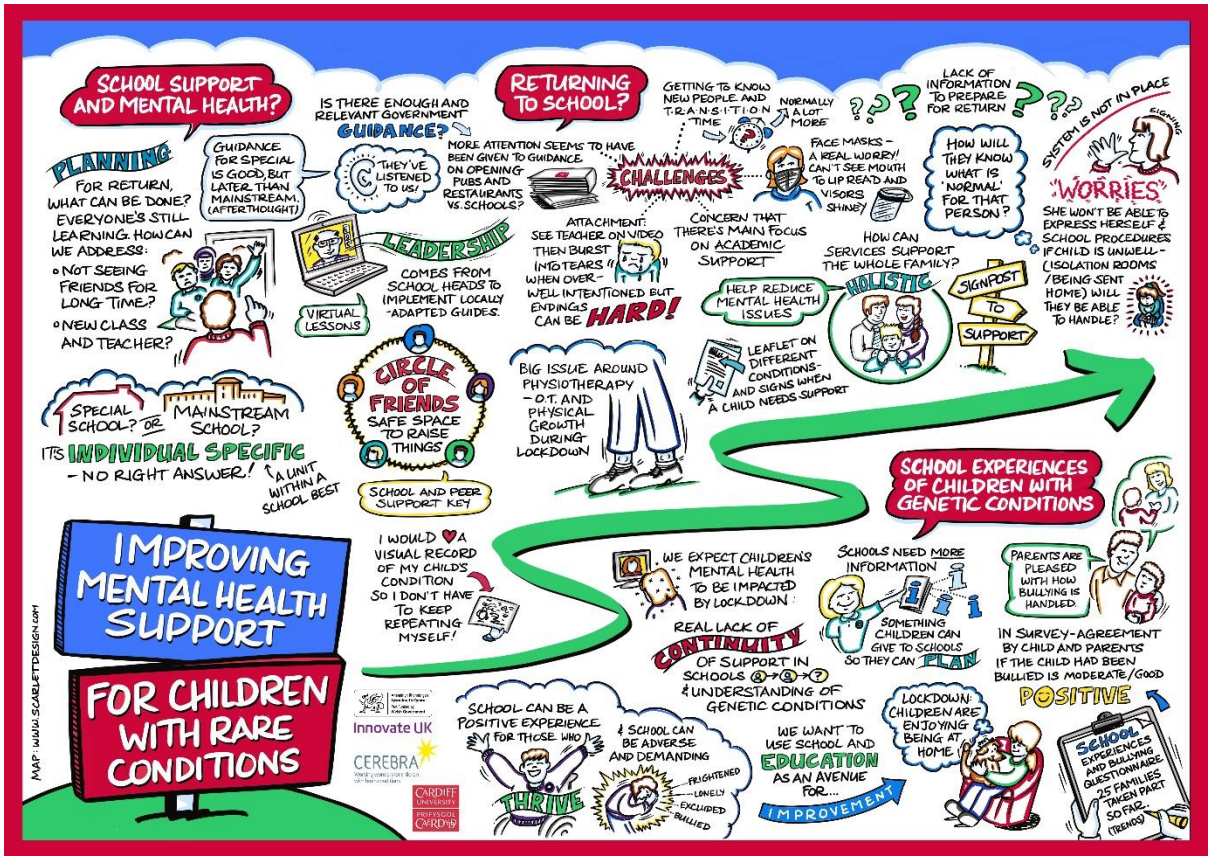
about their child's condition and how to access support. Cerebra also create resources for professionals, as well as individualised support equipment to help children engage in everyday activities.

The aim of the KTP project was to improve access to information and mental health support for children with rare genetic conditions. After interviewing parents, as well as clinical professionals and third sector staff about the challenges currently facing families and support providers in this regard, a steering group (the 'Cerebra Steering Group') was established to advise on strategies to address the barriers that had been identified. The Cerebra Steering Group was made up of parents, clinicians, charity workers and researchers. Information leaflets (https://cerebra.org.uk/download/information-and-support-for-parents-of-a-child-recently-diagnosed-with-a-genetic-condition/), and a parent toolkit (https://cerebra.org.uk/download/mental-health-in-children-with-rare-genetic-conditions/) were produced to provide families with information identified as unavailable at the time.

Following COVID-19 guidelines, most steering group meetings were held online, and each meeting had a specific topic of focus for discussion. For example, see **Figure 1.2** for an infographic which presents the visual minutes of one of the meetings in which the group discussed children's educational experiences over the COVID-19 pandemic.

Whilst the aim of the group was to advise on the KTP project and not my PhD project specifically, elements of this thesis have benefitted from the insights of the members. I will provide further clarification throughout the thesis as to which aspects of the work presented here had their input.

*Figure 1.2 Infographic depicting 'visual minutes' taken during a Cerebra Steering Group meeting about the educational experiences of children with genetic conditions over the COVID-19 pandemic.*




*Infographic produced by Fran O'Hara at Scarlett Design.*

## 1.6 The UK Education System

The education system in the UK has seen many reforms over the last century aimed to make educational attainment less elitist and more accessible to all. Some of the most major reforms include the abolition of school fees in 1918, the raising of compulsory school leaving age from 14 to 15 years old in 1947 (and later to 16 in 1973), and the phasing out of grammar and secondary modern schools in favour of comprehensive schools in 1965 (although some grammar schools still exist in England today). Education has been a devolved matter in the UK since 1999, meaning each of the UK nations govern education policy for their respective nation.

All children and young people in the UK are entitled to free education between the ages 5-16 (5-18 in England and 4-16 in Northern Ireland (NI)), when they are of

compulsory school age. Most schools are ‘community’ or ‘maintained’ state-funded schools (i.e., funded by local authorities (LAs) or government) and follow the national curriculum set out by the respective government, however there are differences between nations regarding the type of school children can attend. For example, England offers community, foundation, voluntary, academy, faith and grammar schools. In comparison, children in Wales can attend either community schools, voluntary controlled and aided schools, or foundation schools. Children in Wales also have the option to attend English or Welsh medium schools. Community schools include both ‘mainstream’ and ‘special’ schools. Special schools are designed to provide specialist provision to children with SEN, whereas mainstream schools do not have such specialised support. Ninety-eight percent of children in the UK who attended a community primary, middle or secondary school attended a mainstream school in the academic year 2021/22 (Office for National Statistics (ONS), 2022). Parents can also choose to enrol their child in an independent school. Independent schools are not funded by the state, but via tuition fees, and they do not have to abide by the national curriculum. Home-schooling is also possible in the UK.

All UK schools are subject to inspections, which are conducted by the nation’s educational review body, for example, Estyn in Wales and Ofsted in England. Inspections aim to ensure schools are meeting the standards outlined by government, that there is appropriate consistency between schools, and to increase their accountability on these measures. Inspection results are publicly available, and some parents will consider this information when choosing their child’s school.

Children and young people sit national public assessments and examinations at different educational stages. Again, systems differ between nations, however all pupils undertake examinations in the year they reach ‘school leaving age’ which is typically 16 years old (18 years old in England). In England, Wales and NI, pupils will complete General Certificate of Secondary Education (GCSE) examinations and in Scotland, National 4 and 5 examinations. Many young people complete higher education to gain Advanced Level (A-Level) qualifications (England, Wales and NI), or Highers and Advanced Highers qualifications (Scotland), and then university. Others undertake vocational training such as apprenticeships or will enter the

workforce. Like school inspection reports, public examination results are publicly available via what are often called ‘league tables’.

Despite the noted educational reforms, education in the UK remains subject to socioeconomic inequality. Children from the least deprived neighbourhoods in England perform better in their examinations at age 16 compared to those from the most deprived areas (Crawford & Greaves, 2015). Furthermore, young people from socially advantaged areas are more likely to continue to higher education (Hunter Blackburn et al., 2016), with 49.4% of young people from mid to high-income households entering higher education compared to 29.2% of young people from low-income households (Department for Education (DfE), 2023).

### 1.6.1 Access to SEN Provision

Until recently, the term SEN described ‘a learning difficulty or disability which calls for special educational provision to be made’ (DfE, Department for Health (DfH), 2015) throughout the UK. The learning difficulty or disability can be permanent or temporary but should cause ‘significantly greater difficulty in learning’ for the young person compared to their peers, or difficulty in using ‘facilities of a kind generally provided for others of the same age’ (DfE, DfH, 2015). SEN is the term still used in England and NI, however ‘additional learning need’ is now used in Wales, and ‘additional support need’ in Scotland. For the purposes of consistency, the term SEN will be used throughout this thesis.

All children with SEN are entitled to SEN provision by which they should receive additional support compared to their peers to help them access the school’s curriculum and achieve goals set by their school and parents. Children whose needs cannot be met with SEN provision should be eligible for an Education, Health and Care Plan (EHCP) (‘Co-ordinated Support Plans’ in Scotland, or a ‘Statement of Special Educational Needs’ in NI). Wales are currently in the process of transitioning to a new system of educational support. Under the new system all children with SEN will be entitled to an ‘Independent Development Plan’ in Wales, not just those who present with the most severe needs. Educational plans are collectively referred to as EHCPs in this thesis and are statutory documents which outline a child’s needs, the support that should be provided to meet those needs, as well as the child’s school

placement. They do not just focus on educational requirements but are designed to provide more substantive support to meet the pupil's health and social care needs too. To receive an EHCP, children must first undergo a needs assessment. This is performed by the child's LA ('education authority' in NI), which is then under legal obligation to provide the relevant support as concluded by the assessment. Schools also have a statutory duty to 'endeavour' to meet the pupil's needs to the best of their ability. Children and young people are entitled an EHCP up until the age of 25 in England and Wales, 16 in Scotland and 19 in NI.

## SEN provision in the UK

### *Pupil characteristics*

The following information reports on school consensus data for England only. In July 2023, 13% of pupils in England received SEN support and 4.3% had an EHCP, which reflected a marginal increase from 12.6% and 4.0% respectively from the previous year (ONS, 2023). The most common type of need recorded for children with an EHCP was ASD, whereas speech, language and communication needs was the most common need for children with standard SEN support. There was an overrepresentation of boys who received SEN support (62.8%) and who had an EHCP (72.4%). The percentage of children with SEN was largest at ages 9 and 10 at which age 15.7% of pupils were recorded as having SEN. The percentage of pupils with such support thereafter decreased. Children with SEN were more likely to receive free school meals and the number of children with SEN for whom English was their first language was disproportionately high compared to the general school population. Children with SEN also make up a disproportionate percentage of children who were permanently excluded from school (ONS, 2023).

### *The Inclusion Debate*

Historically, children with SEN were excluded from education, or segregated from their peers. However, in 1979, the Warnock Report advocated for what was then termed 'integration', now known as 'inclusion' (Warnock, 1979). Later, in 1994, the Salamanca Statement on Special Need Education was published (The United Nations Educational, Scientific and Cultural Organization (UNESCO), 1994) and called on governments to facilitate inclusive education to improve the quality of

education and society. In 2006, the United Nation's (UN) Convention on the Rights of Persons with Disabilities (UN, 2006) was published which went further and legally bound nations, including the UK, to implement inclusive education.

The underlying principle of inclusive education is 'all children in the same classrooms, in the same schools' (United Nation's Childrens Fund (UNICEF), n.d.). Inclusivity is now one of the key principles listed in the SEND Code of Practice (DfE, DoH, 2015), under which the UK government outlines its commitment to inclusive education and the 'progressive removal of barriers to learning and participation in mainstream education'. Such sentiments can be found in the corresponding codes of practice for Wales, Scotland and Northern Ireland (Additional Learning Needs Code for Wales (2021).; Supporting Children's Learning Code of Practice (2017); and the Code of Practice on the Identification and Assessment of SEN (1998) (Northern Ireland)).

However, despite the backing of government at national and international levels, inclusion has not been met without resistance, and disapproval for such systems has been noted by some (Imray & Colley, 2017). What remains under debate is whether full inclusion for all children is possible or appropriate. Some argue against this approach (Imray & Colley, 2017), and others contend inclusion only fails when conditions are such that inclusive practices cannot be implemented properly (Slee & Tomlinson, 2018; Thomas & Loxley, 2022). National policy and guidance, demonstrative school leadership, flexibility in the school curriculum and appropriate levels of staff expertise and support are reported to impact the success of inclusivity (Schuelka, 2018; Shaw, 2017; Thomas & Loxley, 2022).

*Does SEN provision meet need?*

The findings of a recent review into the UK SEN system were published in 2022 (DfE, Department of Health and Social Care (DHSC), 2022). Prior to this, the House of Commons Education Committee (HCEC) (2019) had already concluded that LAs and schools were unsuccessfully meeting the needs of children with SEN and that many children were being failed by the system. The SEN review findings (DfE, DHSC, 2022) were largely a reflection of those reported by the HCEC.

Firstly, despite the noted commitments to provide inclusive education, exclusionary practices were reported (HCEC, 2019). This included schools refusing places to children with SEN and instances of 'off-rolling', which is the removal of a pupil without formal school exclusion processes being followed for reasons which are more beneficial to the school versus the child. As already noted, children with SEN are also overrepresented amongst children who are formally permanently excluded from education within the UK (ONS, 2023). The pressures on schools from government to deliver high standards as judged by publicly available examination results and school inspection results, have been criticised as contributing to these exclusionary practices (Shaw, 2017).

Secondly, the level of support provided was not consistent across schools or geographical areas, which was partly influenced by local practices and the level of SEN expertise within a school. This contributed to a 'postcode lottery' of provision.

Unviable budgets were also reported to be an issue and had previously been highlighted by the National Audit Office (NAO, 2020). The UK Government has recognised the financial unsustainability of the system and has announced 'record investment' in children with SEN to address this issue (DfE, DHSC, 2023).

Overall, the SEN review highlighted 'a vicious cycle of late intervention, low confidence and inefficient resource allocation' (DfE, DHSC, 2022). Frustration felt by parents and carers was the noted consequence of having to navigate a system which is underfunded, riddled with processing delays and deemed to be complex and combative. The marked rise in the number of appeals made by parents against LAs in response to their refusal to grant their child an EHCP, which rose from 298 in 2013/14 to 526 in 2017/18 (NAO, 2019), provides some evidence for parent dissatisfaction with the current support available.

### Access to SEN provision for children with ND CNVs

Resources have been created to help educators support children with different ND CNVs at schools, for example, guidelines and information for those working with children with Williams syndrome, PWS and 22q11 DS (Chedd et al., 2006; Reilly & Stedman, 2013; Udwin et al., 2007; Ward & Farrar, 2022). An online resource has recently been developed by researchers at the University of Surrey and the Cerebra

Network for Neurodevelopmental Disorders to support educators working with children who have genetic syndromes (https://www.findteacherresources.co.uk/). Whilst these resources are valuable sources of information for teachers and parents, the ability of parents to actually access educational support or the implications of doing so have not been investigated.

Given their phenotypic heterogeneity, it is possible that not all children with a ND CNV will require SEN support or an EHCP, but it is likely that many will. Considering the purpose of SEN provision is to facilitate access to the school's curriculum, children with a ND CNV who receive such support when it is needed should display an improved ability to successfully access education. Conversely, those not in receipt of such support when needed could be hindered in this regard.

The importance of accessing education has long been recognised. The right to education is listed under the 1998 Human Rights Act and 'Quality Education' is the fourth goal of the UN's Sustainable Goals, aiming to 'ensure inclusive and equitable quality education and promote lifelong learning opportunities for all'. In 2015, the UK Minister of State for Schools, Nick Gibb, stated the importance of education in preparing individuals for adult life, as well as for the country's culture and economy (DfE, 2015).

At the individual level, access to education can help to level the playing field between the most and least disadvantaged in society. According to UNESCO (2019), education is 'one of the most powerful tools by which economically and socially marginalized children and adults can lift themselves out of poverty and participate fully in society'. Furthermore, the Social Determinants of Health (SDH) approach to health (World Health Organisation (WHO), 2008), which describes the influence that non-medical factors have on a person's health outcomes, recognises access to education as a key determinant of individual health and health equity. Education is linked to health via economic mediating factors like employment status and income, as well as social and psychological mediators such as access to social support, problem solving, cognitive ability and recognition of health symptoms (Raghupathi & Raghupathi, 2020). Additionally, the health benefits of education can span generations. Children of more highly educated parents are less likely to be overweight (McCrory et al., 2019) and are more likely to be more highly educated

themselves (Akresh et al., 2023), and are thus expected to experience the health benefits associated with education. Beyond implications for the individual, education is linked with a reduction in poverty, greater productivity, national economic growth, lower crime rates, less demand on public health services, and reduced need for public welfare (Mitra, 2011).

Although the ability to access education via SEN provision for children with ND CNVs has not been investigated previously, studies exploring related aims can give us an indication of such experiences. In a survey of 562 parents and carers of children with LD and disabilities in the UK (Parsons et al., 2009), parents were on average satisfied with the support their child's school was providing. However, the most dissatisfied parents were those whose children displayed psychosocial difficulties, which included emotional and behavioural disorders (EBD) and mental health difficulties. Whilst there is no formal definition for EBD, Place et al. (2000) reported that children in specialist EBD schools had ADHD, CD, ODD, depression and anxiety, all of which have been found at increased rates in children with ND CNVs, indicating that the parents of children with ND CNVs could be particularly dissatisfied with their child's educational support. Conversely, parents were most satisfied if their child had an EHCP. Since this study, there has been a change in government in the UK and several consequent reforms to the education and SEN provision systems. Therefore, these results may no longer present an accurate picture of parent satisfaction. Indeed, the SEN review findings indicate considerable failures in meeting need within the current SEN system (DfE, DHSC, 2022).

More recently, parents of children with a newly diagnosed developmental condition (ADHD, ASD or foetal alcohol spectrum disorder) reported barriers to access which included failure to recognise and support need, especially at school (McCarthy, 2022). Parents felt 'patronised' and 'belittled' by school professionals and reported there was a need for children to have a diagnosis before support could be provided. Again, whilst the findings of this study cannot be directly applied to children with ND CNVs, they support the notion that this group could face similar challenges given higher incidence rates of ADHD and ASD in those with ND CNVs.

Russell et al. (2023) reported on the qualitative educational experiences of children with ADHD. One of the main themes identified after speaking with parents (N = 28)

and young people with ADHD (N = 64) was the ‘problematic provision loop’ which described the difficulties arising from differences between children with ADHD and their peers. Children and their schools struggled to adapt to one another resulting in the reallocation of children to multiple schools, harsh disciplinary measures, bullying and negative relationships with teachers. Whilst there were examples of successful support strategies, negative impacts of children’s school experience included the labelling of parents as ‘bad parents’ and children as ‘naughty’ or ‘lazy’, exclusion and expulsion from school, and poorer school attainment. Positive experiences were noted when children’s support needs were met.

Whilst these studies do not describe the experiences of children with ND CNVs, they provide insight into the level of support received by children with similar neurodevelopmental conditions, as well as the experience of parents when trying to obtain support. Findings could be similar in children with ND CNVs.

*Investigating the experiences of children with ND CNVs specifically*

The ability of children with an ND CNV to access quality education has not been determined. Based on the research discussed above, it could reasonably be assumed that the needs of this group are not always fully supported at school, and that parents face challenges in securing support. Should this be the case, children with ND CNVs are at risk of lifetime disadvantages compared to their peers regarding their and their current and future family’s health and future income and employment opportunities. Neglecting this group also has negative economic, health and productivity implications for wider society.

There is no knowledge at present whether there are potential barriers or facilitators in accessing support which are unique to this group of children, or whether they are experienced by all children with ND CNVs. Should different families experience different barriers and facilitators, such experiences could go some way to elucidating factors which might influence the phenotypic heterogeneity of children. This knowledge is important to aid our understanding of the potential changes that could be made within school and the SEN support system to benefit this group’s learning and the subsequent personal and societal long-term outcomes. Furthermore, this knowledge will help evaluate whether measures proposed by the DfE and DHSC

(2023) will satisfy the needs of children with ND CNVs as well as children with other SEN.

As noted earlier, parents of children with ND CNVs are at increased risk of psychological problems. The pressures of care-coordination on carers of individuals with rare conditions can lead to increased stress and compromised mental health. By investigating the experiences of parents of children with ND CNVs when trying to access educational support for their children, we can better understand how such experiences impact them and whether they compound the level of stress and psychological adversity experienced by parents. This knowledge will help us to better understand what measures could be taken to help relieve parents and improve their well-being.

## 1.7 Peer relationships at school

Attending school goes beyond the purposes of just academic development and plays an important role in fostering children's social skills too (Faulkner & Miell, 1993). For most young people, attending school provides them with the greatest opportunity to build relationships with children beyond their family's social network. Starting school is the first time for many children whereby they will spend long periods of time away from home without the reassuring presence of a parent or other carer. The absence of such attachment figures whom children can rely on for 'back up' in stressful situations forces them to build alternative supportive relationships whom they can rely on within the school environment.

### 1.7.1 Friendships

A friend is 'a person with whom one enjoys mutual affection and regard' (Oxford English Dictionary, 1962). Positive early peer relationships predict healthy adult adjustment (Bagwell et al., 1998) and are important for psychological health at all stages of life (Feeney & Collins, 2015). Positive social relationships also have long-term impacts on later physical health (Cundiff & Matthews, 2018) and the SDH approach to health (WHO, 2008) also recognises the contribution of social inclusion for health equity.

In the academic year 2021/22, 63% of young people taking part in the Student Health and Wellbeing in Wales survey (Page et al., 2023) reported that they could 'count on their friends when things go wrong'. Whilst it is encouraging that most young people agreed they had such supportive relationships, a large proportion (37%) of children did not, indicating that some children may have trouble in forming close friendships.

Friendships in children with ND CNVs have not been studied, however it would not be surprising if they were overrepresented amongst children who struggled to develop friendships. As noted, children with ND CNVs exhibit a range of health and developmental conditions and such challenges have been associated with increased difficulty in making friends. For example, children with ASD face higher levels of social exclusion, and report higher levels of loneliness compared to those without ASD (Bauminger & Kasari, 2000). Children with ADHD are more likely to be rated by other children as either very shy or aggressive, and are less well-liked by their peers compared to children without ADHD (Hodgens et al., 2000). Distinctive congenital facial features are characteristic of children with ND CNVs and children with facial differences are judged less favourably and less likely to be befriended compared to other children (Masnari et al., 2013). Furthermore, parents report their child with an ND CNV to exhibit difficulties which indicate social impairment in this group (Cunningham et al., 2022). Social problems have been associated with lower levels of social inclusion especially for children with additional behavioural problems (Frostad & Pijl, 2007).

### 1.7.2 Bullying

Given the importance of friendships, it is unsurprising that experiences of bullying in childhood and adolescence can have significant consequences for a child's outcomes. Bullying is not a new phenomenon and was historically considered an inevitable experience whilst growing up. However, in 1982, three students aged 10-14 in Norway took their own lives and mass media coverage attributed the cause of suicide to bullying the children had been victims of. This triggered national concern about the impacts of such experiences and bullying has since become a common topic of research. Our knowledge about the impacts of bullying behaviour has thus

evolved, as has public opinion regarding its acceptability. Bullying is now considered a major public health issue (WHO, 2012).

Professor Dan Olweus, who is regarded a pioneer of bullying research, defined bullying as repeated and targeted aggression toward another person who is unable to defend themselves (Olweus, 1993). Such behaviours can take a traditional form, for example physical and verbal abuse, or a more modern form known as cyberbullying. Traditional forms of bullying can further be divided into direct and indirect bullying. The former comprises actions which are more outwardly obvious, for example physical abuse. Indirect methods are more subtle and therefore are harder to detect, for example spreading harmful rumours. When children are directly involved in bullying behaviour, they assume one of two positions: the bully or the victim. Bullies are the perpetrators, whereas victims are those who are on the receiving end of such behaviours. A minority of children fall into the category of bully-victim. These children both perpetrate and are victims of bullying. Additional roles, 'bystander' and 'defender' were defined by Lagerspetz et al. (1982). Bystanders are children who do not get actively involved when they witness the bullying of others, and defenders are those who intervene to help victims.

The prevalence and type of bullying behaviours exhibited by children varies across regions, but bullying is a common experience. Utilising global and regional data, UNESCO (2019) reported that nearly one third of pupils had been bullied in the last month (25% of children in Europe). Physical bullying was the most common type of bullying reported in most countries, with Europe and North America the exceptions. In these regions, psychological bullying was the most common. Trends over time also varied by region. Some countries observed a decrease in prevalence, whereas others saw an increase or no change at all. These differences could represent differing national responses to tackle bullying (Ananiadou & Smith, 2002). Girls and boys experienced similar levels of bullying, however boys were more likely to bully others. Boys were also more likely to use and experience direct, physical methods of bullying whereas girls more likely utilised and experienced indirect methods. Bullying declined with age and children from disadvantaged backgrounds and schools were more likely to be bullied. Schools with poor school climate had higher rates of bullying. School climate incorporates various aspects of a child's subjective school

experience including their relationships, learning, environment and perceived safety (Zullig & Matthews, 2014).

Bullying is associated with increased rates of suicidality, lower educational attainment, increased school absence and criminality in childhood (UNESCO, 2019). Those who are bullied also report a poorer sense of belonging at school and are less likely to continue in education after compulsory school age. Additional immediate impacts include poor sleep, feelings of loneliness and reduced quality of life. The impacts of bullying can extend into adulthood. For example, poorer social relationships and greater economic hardship is observed in adults previously involved in childhood bullying, even at 50 years old (Takizawa et al., 2014). The long-term impacts of bullying are so great it has been claimed that bullying could have even worse long-term impacts than parental maltreatment (Lereya et al., 2015). The negative impacts of bullying are particularly marked in bully-victims (Copeland et al., 2013). Evidence indicates that the impacts of bullying are dose-respondent, with worse outcomes reported for those who are more frequently bullied (Klomek et al., 2015). Protective factors like having a supportive family (Bowes et al., 2010) can help shield individuals from the harmful effects of bullying. Friendships may also help ameliorate the effects of bullying, however findings are mixed (Schacter et al., 2021).

More recently, there has been concern about the impacts of cyberbullying. Cyberbullying has been defined as ‘an aggressive act or behaviour that is carried out using electronic means by a group or an individual repeatedly and over time against a victim who cannot easily defend him or herself’ (Smith et al., 2008). The concern surrounding cyberbullying has increased as social media and access to it has grown (in the UK, 95% of 15 year olds use social media (Ofcom, 2021)). Fourteen percent of children aged 9-16 years across 7 European countries reported they had been bullied online in the last 12 months (Mascheroni & Ólafsson, 2014). However, findings also indicate there is a strong correlation between cyberbullying experience and more traditional forms of bullying. That is, children who are the victims of cyberbullying are likely to be victims of traditional forms of bullying too (Kowalski & Limber, 2013). This presents difficulties when trying to unpack the impacts of cyberbullying, but it has been suggested that cyberbullying poses little additional harm to children who are already victims of traditional forms (Wolke et al., 2017).

Nonetheless, the increased accessibility to online platforms means victims have limited opportunity for respite compared to those deemed available to older generations.

### Bullying experiences of children with ND CNVs

The bullying experiences of children with ND CNVs have not been explored beyond a pilot study conducted by Mayo and Neidham (2017), who found individuals with 22q11 DS experienced more victimisation compared to controls. However, this study was very small and collected data from only 12 participants (10 children with 22q11 DS and 2 controls) and therefore we cannot conclude there is a robust relationship between having 22q11 DS and increased bullying.

Studies that have investigated the bullying experiences of children who face similar challenges to those experienced by children with ND CNVs supports the hypothesis that children with an ND CNV are an at-risk group. Children considered 'different' in any way are more likely to be bullied, however physical appearance is reportedly the most common reason for young people to be bullied, especially for girls (UNSESCO, 2019). Indeed, children with physical characteristics associated with ND CNVs are at higher risk of being bullied. Children with congenital facial differences, including those with cleft lip report being teased because of their facial appearance (Strauss et al., 2007). Furthermore, children with neurodevelopmental conditions and emotional problems are more likely to be bullied compared to their peers (Blake et al., 2016; Symes & Humphrey, 2010). Adolescents with congenital heart disease are victims of bullying and discrimination due to lack of tolerance and empathy from peers, as well as parents' over-protectiveness (McMurray et al., 2001). Children with impaired motor skills are at increased likelihood of being name-called, purposefully socially excluded, and verbally threatened (Campbell et al., 2012).

Further investigation into the bullying experiences of children with ND CNVs is supported by Mayo et al. (2019) who argue that bullying involvement could play a role in the development of psychotic conditions in children with 22q11 DS. They suggest the vulnerability-stress model for psychosis (Zubin & Spring, 1977) as an explanatory mechanism which posits that environmental stressors interact with a person's genetic vulnerability for psychosis, and in turn elicits psychotic symptoms.

Beaton and Simon (2011) support that such a relationship could exist between chronic stress and psychosis in children with 22q11 DS. Adverse childhood experiences, including bullying have been linked to abnormal physiological stress responses (Arseneault, 2018; Elzinga et al., 2008), and are commonly experienced by children who are at clinical high risk for developing psychotic disorders (Mayo et al., 2017). Thus, Mayo et al. (2019) hypothesise that bullying involvement could cause chronic stress in children with 22q11 DS, which compounds their heightened genetic risk for psychosis (Williams & Owen, 2004) and triggers such experiences. Swearer and Hymel (2015) endorse the mediating role of chronic stress to help explain the link between bullying and psychological adversity.

Although Mayo et al. (2019) focus their argument on children with 22q11 DS, additional ND CNVs have been robustly linked with psychotic conditions, including 1q21.1 deletion and 16p11.2 duplication (Levinson et al., 2011; Rees et al., 2014). Furthermore, the stress-vulnerability model has been applied to psychiatric conditions beyond psychotic conditions (Goh & Agius, 2010). Thus, the proposed argument could apply to ND CNVs beyond 22q11 DS and to conditions beyond psychosis, however, research is still needed to elucidate the relationship between stress and psychosis in these other ND CNVs and whether children with ND CNVs are in fact at increased risk for bullying. Should bullying experiences be more common in children with ND CNVs, limiting the level of stress experienced by this group via anti-bullying measures could prove fruitful in protecting them against adverse psychological impacts. Furthermore, noting the far-reaching impacts of bullying mentioned earlier, anti-bullying measures could also help to improve children's later socioeconomic and physical health outcomes.

## 1.8 Summary, aims and implications

As indicated above, there is a dearth of knowledge regarding the environmental experiences of children with ND CNVs, and how these experiences impact children's outcomes and phenotypic variability. Both social and academic school experiences have significant and lasting effects on a person's life outcomes, and therefore represent an important environmental experience in all children's lives. At present, we have an extremely limited understanding of how children with ND CNVs experience school and most of the inferences we can make are based on

extrapolations based on studies of children with comparable physical and psychiatric challenges.

Furthermore, given the challenges and frustrations associated with the SEN provision system for parents (HCEC, 2019; DfE, DHSC, 2022), parents of children with a ND CNV who are in need of SEN support are likely also challenged in obtaining such support for them given they often take a lead role in coordinating their child's care (Simpson et al., 2021). These responsibilities have a noted harmful impact on carers' mental wellbeing and thus, better understanding the experiences of parents when accessing educational support for their children could provide context to the high rates of psychiatric conditions observed in mothers of a child with a ND CNV.

To address these knowledge gaps, I aimed to explore the peer relationships of, and the support received at school by, children with a ND CNV, as well as how such experiences can impact both children and their family.

The specific aims of this thesis were:

1. Cross-sectional investigation of bullying experiences in children with ND CNVs compared to controls and the individual and contextual factors associated with such experiences.
2. Cross-sectional investigation of access to SEN support for children with a ND CNV, and parent satisfaction with their child's educational support.
3. Explore the qualitative experiences of parents when obtaining educational support for their child, and the impacts of these experiences and the subsequent level of educational support on children with a ND CNV and their family.

Research presented in this thesis will provide insight into the school experiences of children with a ND CNV, which is a currently understudied topic area. Better understanding of these experiences could help to elucidate whether such experiences may impact children's outcomes and thus help identify a likely important environmental contribution to phenotypic heterogeneity. Importantly, this knowledge will provide a basis for future investigations into potential environmental modifications to be made at school to help decrease the likelihood of harmful

exposures and to help children's outcomes. Furthermore, this research could begin to provide professionals and parents with a preliminary basis of understanding about associated outcomes of ND CNVs given certain educational circumstances. This could provide clinicians with more confidence when first discussing genetic diagnoses with parents and empower parents with knowledge about the type of support mechanisms potentially beneficial for their child in future. Similar information could be communicated to educational professionals via charities such as Cerebra to help raise awareness of ND CNVs and the support strategies children could benefit from at school in the hope that this will increase the likelihood that the right support is implemented. Better knowledge amongst professionals on how to best support children could help to lighten the burden on parents who currently take on the role of care coordinator for their child, a role which negatively impacts their psychological health. These implications will likely be more widely felt as WGS becomes more routinely embedded in healthcare practices.

# 2 The individual and contextual risk factors for bullying in children with neurodevelopmental copy number variants

## 2.1 Chapter Overview

The phenotypic profile of children with ND CNVs is highly variable, but knowledge about which environmental factors might contribute to such variability, and which children with a ND CNV are more likely to encounter harmful environmental exposures is limited. Bullying in childhood and adolescence is associated with adverse immediate and long-term outcomes but understanding about these experiences in children with ND CNVs is sparse.

The aim of this chapter was to explore the bullying experiences of children with ND CNVs and compare experiences to a control sample of unaffected siblings. I also explored individual and contextual risk factors for bullying with the aim to begin characterising children with a ND CNV who are at greater risk for bullying. In this chapter, I define bullying, discuss methodological issues quantifying the prevalence of such experiences, and some of the risk factors for, and impacts of bullying. I introduce the ECHO, IMAGINE-ID and DiGEN studies, which provide the cohorts in which bullying is investigated.

Children with a ND CNV were more likely to experience bullying compared to controls (52.3% vs 27.4% respectively). Children's neurodevelopmental profile and age significantly influenced their victimisation experiences, however additional factors not explored in this chapter seem to have a notable impact and should be investigated.

These findings could indicate the risk of bullying is heightened in children with ND CNVs and that current anti-bullying measures may not be effectively protecting all children against such experiences. Steps to increase the awareness of risk factors for bullying in this group amongst parents and school professionals should be taken to help protect them from harmful peer interactions in childhood. Findings also emphasise the importance of investigating the environmental experiences of children with ND CNVs as bullying experiences differed between children and thus could have an important role in influencing their varied phenotype.

## 2.2 Introduction

Dan Olweus's (1993) definition of bullying is often used by researchers when characterising the behaviour. According to Olweus (1993), ‘a person is bullied when he or she is exposed, repeatedly and over time, to negative actions on the part of one or more other persons’ and ‘has difficulty defending himself or herself’. Necessary components of the behaviour include its targeted and repetitive nature, and the intention to cause harm. There must be a power imbalance between the bully (or bullies) and the victim(s), with the victim(s) being the less powerful party (e.g., in physical strength, number, popularity). Bullies can utilise different techniques to victimise their targets including physical, verbal and online means, but methods are categorised into two overarching types of bullying behaviour: direct and relational. Direct methods tend to be those which are more overt, such a physical aggression, whereas relational methods tend to be less obvious, for example, spreading rumours or purposefully excluding an individual to harm their peer relationships.

Bullying in childhood and adolescence is relatively common. In 2018, the DfE reported that 17% of children in England aged 10 to 15 had been bullied in the last year, a figure which was consistent with the rate reported for the preceding 4 years (DfE, 2018). UNESCO (2019) reported that one in three children globally were bullied by their peers on one or more days in the last month whilst at school. However, inferring the true prevalence of school-based bullying in young people is challenging due to methodological inconsistencies between studies. Researchers have used many different measures and techniques (e.g., questionnaires versus observation) to measure bullying, and the chosen informant also varies between studies (children themselves, or their peers, parents, or teachers). The way in which bullying is defined also varies between studies, as well as the period explored (e.g., the last year versus the last 6 months) and the frequency scales applied to the chosen measure (e.g., once a week/once a month versus sometimes/often/very often).

Similar to studies reporting on bullying in the general population, the reported rates of bullying in children and young people (CYP) with ID vary. This is likely due to differences in the way bullying and ID are measured, and whether researchers

include children with ID and comorbid conditions within their sample or restrict to children with ID only.

Christensen et al. (2012) investigated the rate of mother and self-reported bullying in CYP with ID compared to their typically developing peers. Fifty-two percent of mothers reported their child with ID had been bullied compared to 42% of mothers of children who were classified within the typically developing group. This finding was non-significant, however, CYP were significantly more likely to self-report they had been bullied compared to the typically developing group (62% vs 41% respectively). In both groups, bullying decreased with age. Conversely, Zeedyk et al. (2014) found children with ID self-reported lower rates of bullying (49%) compared to their mothers (57%). Again, these figures were higher than the self and mother reported bullying rates for typically developing children (42% and 36% respectively), however were lower than those reported for children with ASD (75% and 80% respectively).

A systematic review conducted by Maïano et al. (2016) concluded the rate of bullying in school aged individuals with ID to be 36.6% and the most common type of bullying experience to be verbal in nature. Self-reported bullying was typically higher than parent reported rates when data were collected via questionnaires and when bullying was measured using dichotomous measures versus severity cut off scales. Whilst some of the included studies reported higher prevalence in those with ID compared to typically developing CYP, others reported no difference. Thus, the review emphasised the disparate findings often observed between studies.

Early experiences of bullying by peers are linked to immediate adverse psychological effects, such as unhappiness at school, academic difficulties, social isolation, and self-harm and suicidal ideation (UNESCO, 2019). Persistent long-term consequences also include increased rates of anxiety, depression, suicide attempts and suicide completions (Copeland et al., 2013; Klomek et al., 2009; Sourander et al., 2007). Childhood bullying has even been linked to increased psychological distress at age 50 (Takizawa et al., 2014). Additional long-term impacts beyond mental health include higher likelihood for physical health conditions including inflammation and obesity (Takizawa et al., 2015), as well as higher rates of unemployment (Takizawa et al., 2014; Wolke et al., 2013). Childhood bullying has been linked with later criminal offending behaviour (Gibb et al., 2011) and poorer

social relationships and quality of life (Takizawa et al., 2014). Adverse consequences are particularly notable in victims of chronic bullying (Arseneault, 2018).

Bullying was traditionally considered a rite of passage for children and important for building social skills and character, but since research has uncovered the harmful individual and societal consequences associated with these experiences, many governments now have strategies in place to tackle bullying and violence in schools (Ananiadou & Smith, 2002). In the UK, all schools are legally obliged to establish an anti-bullying policy which must be known to all staff, as well as pupils and their parents (Smith et al., 2012). Many school-based bullying prevention programmes have been implemented and evaluated, some of which appear to have successfully reduced bullying incidences at school (Gaffney et al., 2019).

Despite government and school policies, some children are sadly unable to escape the taunts of their peers. Certain individual and contextual factors have been found to put children at greater risk for bullying compared to their peers. Contextual characteristics are those deemed less individualistic and are more group level based. For example, school climate has been extensively linked to pupils' bullying involvement (Thapa et al., 2013). Bullying has been found to decline in secondary school compared to primary (Pellegrini & Long, 2002), and school placement (mainstream versus special school) seems to have an impact depending on the characteristics of the child. To illustrate, Rowley et al. (2012) found that children with ASD who had more social difficulties were no more likely to be victimised in one school placement versus the other, however children with ASD with fewer social difficulties were more victimised in mainstream settings.

Individual characteristics include factors such as age, sex and personality. Findings are mixed regarding the difference in prevalence of bullying between age (DfE, 2018; UNESCO, 2019) and genders, but it is generally accepted that boys are more often victims of physical bullying, and girls use and experience more relational methods (Smith & Gross, 2006; UNESCO, 2019). Additionally, low self-esteem (O'Moore & Kirkham, 2001), social communication difficulties (Cappadocia et al., 2012) and hyperactive traits (Wolke et al., 2000) all increase children's risk of bullying. Low socio-economic status may slightly increase the odds of being bullied (Tippett & Wolke, 2014) but findings on the bullying implications associated with ethnicity are

unclear (Tippett et al., 2013; UNESCO, 2019). Children with SEN are also at increased risk of peer victimisation. In the UK, Green et al. (2010) investigated the type of bullying experienced by children who took part in the Longitudinal Study of Young People in England and found that children with SEN were more likely to be the victim of both direct and relational bullying at every age of assessment compared to their peers. Blake et al. (2012) found that children in America with a disability were up to 1.5 times more likely to be bullied compared to the national average rate for typically developing children.

Given than children with SEN and disability are at increased risk of bullying and that children with ND CNVs experience SEN related difficulties (Chawner et al., 2021), it seems likely those with a ND CNV could be at high risk of being bullied. To my knowledge, only one pilot study has investigated the bullying experiences of this group specifically. Mayo and Niendam (2017) found children with 22q11 DS (N = 10) reported higher rates bullying compared to controls (N = 2). The researchers note their small sample size limits the conclusions that can be drawn, however advocate the importance of investigating bullying experiences in this group within a larger sample (Mayo et al., 2019). They argue bullying could play a role in the development of psychosis in children with 22q11 DS via mechanisms underlying the vulnerability-stress model for psychosis (Zubin & Spring, 1977). This model proposes that environmental stressors (e.g., bullying) interact with a person's genetic predisposition for psychosis, which subsequently elicits psychotic symptoms. However, the stress vulnerability model can be applied to psychiatric conditions more generally (Goh & Agius, 2010) and could also be applied to ND CNVs beyond 22q11 DS.

Many studies have demonstrated increased risk of exposure to bullying in children with challenges associated with ND CNVs. For example, ASD, ADHD, ID, psychotic experiences (PE), congenital heart disease, facial dysmorphology and impaired motor skills are all known risk factors for victimisation (Blake et al., 2012; Campbell et al., 2012; Catone et al., 2015; Christensen et al., 2012; McMurray et al., 2001; Strauss et al., 2007), and are associated with ND CNVs (Chawner et al., 2021).

The phenotypic heterogeneity of ND CNVs (Chawner et al., 2019; McDonald-Mcginn et al., 2001) poses difficulty for professionals in knowing the clinical significance of

genetic diagnoses (Lee & Scherer, 2010) and children with ND CNVs are vulnerable to being unwittingly neglected by services (Tang et al., 2014; Young et al., 2011).

Several genetic factors are implicated in phenotypic expression (discussed in Chapter 1). Environmental factors likely also impact phenotype (Wolstencroft et al., 2022), however research which explores the potential influence of environmental experiences on the outcomes of children with ND CNVs is sparse. Given that measures can, to a certain extent, be implemented to shape our environment, effort should be dedicated to exploring whether environmental exposures, like bullying, contribute to the variability in presentation we observe. This knowledge could help to improve the health and social outcomes of children with ND CNVs by ensuring that anti-bullying interventions and programmes are inclusive to the needs of this often-neglected group. Furthermore, given the host of negative consequences associated with bullying, awareness that a child has been victimised may provide a helpful marker to guide professionals as to what kind of support that child might benefit from.

The ECHO, the IMAGINE-ID and the DiGEN study cohorts (discussed below) presented an opportunity to investigate bullying experiences in a large cohort of children with ND CNVs. Participants in these studies included the siblings of the affected children who did not have a ND CNV and whose own experiences were assessed with the same measures. This allowed me to investigate whether children with a ND CNV were more vulnerable to bullying compared to an unaffected sibling group thus controlling for family factors.

It would also be advantageous to know which children with a ND CNV are particularly vulnerable to bullying. With this in mind, I investigated if there were certain individual and contextual risk factors seen more often in children with a ND CNV who are bullied compared to those who are not bullied and to what extent these factors explained their risk for bullying.

## 2.3 Aims

The specific aims of this chapter were to answer the following questions:

**Aim 1: Are children with a ND CNV more likely to experience bullying compared to unaffected sibling controls?**

Given the challenges associated with ND CNVs (Chawner et al., 2021) and the literature reporting higher risk for victimisation in children with these challenges (Blake et al., 2012; Campbell et al., 2012; Catone et al., 2015; Christensen et al., 2012; McMurray et al., 2001; Strauss et al., 2007), I hypothesised a higher proportion of children with an ND CNV to have experienced bullying compared to their siblings.

**Aim 2: What are the individual and contextual risk factors associated with bullying in children with an ND CNV?**

Individual risk factors:

*a) Neurodevelopmental risk factors*

I predicted that children with a ND CNV who met diagnostic criteria for neurodevelopmental conditions and traits robustly associated with ND CNVs (Coe et al., 2012; Levinson et al., 2011; Rees et al., 2014) (i.e., ADHD, ASD, ID, and PE) would be at greater risk for bullying. Risk was also expected to increase with the number of neurodevelopmental 'conditions' a child met criteria for (i.e., the greater their neurodevelopmental burden). Note 'conditions' is written in inverted commas like this because PE in this study comprises psychotic-like symptoms, not a diagnosable psychotic condition per se. Children who displayed higher symptom count scores for these 'conditions' were predicted to also be at greater risk compared to children with lower symptom count scores.

*b) ND CNV inheritance status: inherited or* de novo*?*

I investigated the impact of inheritance status on children's bullying experience because this was of interest to the parents within the Cerebra Steering Group. Children with an inherited ND CNV were predicted to be at higher risk for bullying compared to children with a *de novo* ND CNV given that children with inherited ND CNVs more often experience social, emotional and behavioural problems (Cunningham et al., 2021; Wolstencroft et al., 2022).

Contextual risk factors:

*c) School placement: mainstream or special?*

Exploring school placement as a risk factor for bullying was suggested by the Cerebra Steering Group. Parents reported that children in special schools can display aggressive acts to others when they have communication and behavioural difficulties. These physical incidences might be interpreted by children as bullying, however parents did not believe there was intent behind these actions. My measure of bullying (see Methods section) utilised parent reported data and therefore I predicted that children attending a mainstream school would more often be reported to have experienced bullying compared to those in special school placements because parents would more likely regard the incidences that took place in mainstream school as intentional.

**Aim 3: To what extent do neurodevelopmental conditions and traits explain the risk of bullying in children with a ND CNV?**

I hypothesised that both children with a ND CNV and their unaffected siblings would be at greater risk for bullying if they met criteria for any of the neurodevelopmental 'conditions' mentioned above. Children in both groups who scored positive for a greater number of symptoms associated with these 'conditions', and who had a higher overall neurodevelopmental burden would also be at greater risk. However, children with a ND CNV would remain at greater risk compared to their unaffected siblings despite the presence of these neurodevelopmental factors because they would be more likely to experience additional challenges also associated with increased risk for bullying (e.g., physical).

## 2.4 Methods

### 2.4.1 The ECHO, IMAGINE-ID and DiGEN Studies

The study sample comprised children who had taken part in the ECHO, IMAGINE-ID and DiGEN studies. The latter two of these studies are still ongoing. Each has built a large cohort of children, providing a wealth of detailed data, including demographic,

psychiatric and cognitive data to explore the impact of CNVs known to be robustly associated with neurodevelopmental conditions.

### ECHO: The Experiences of CHildren with cOpy number variants study

The ECHO study was established by Professor Marianne van den Bree and Professor Sir Michael Owen (both Cardiff University) in 2010 to study the nature and prevalence of psychopathological, behavioural and neurocognitive problems in children with ND CNVs over time. Detailed information about participants was collected via self-report questionnaires, and cognitive and psychiatric assessments which were administered by trained Psychology Research Assistants.

Children taking part in ECHO were those with an ND CNV aged 6-17 years old. Children's unaffected siblings who were within the same age range were also invited to take part and provided a comparison (control) sample. Children were recruited into ECHO via NHS medical genetics clinics, charities (such as Unique, Max Appeal! and 22crew), the ECHO study website, social media, and word of mouth. Participants were re-visited up to 4 times, approximately every 2.5 years, establishing a longitudinal cohort of children to study the impacts of CNVs over time. Not all children took part at every time point due to retention issues which are common in studies of this kind (Schaffer, 1996).

### IMAGINE-ID: The Intellectual Disability & Mental Health: Assessing the Genomic Impact on Neurodevelopment study

IMAGINE-ID ('IMAGINE') is a collaborative study between University College London, Cardiff University and University of Cambridge. Its Principal Investigators are Professor David Skuse, Professor Marianne van den Bree, Professor Jeremy Hall, Professor Sir Michael Owen and Professor Lucy Raymond.

Like ECHO, IMAGINE was established to better understand the implications of rare pathogenic CNVs in children. The study began in 2014, and in 2020, additional funding was secured to undertake a second phase of the study. This second phase

(IMAGINE-2) is ongoing and builds on the first phase (IMAGINE-1) by revisiting families who took part in IMAGINE-1 to build a longitudinal cohort and recruiting new families into the study. This chapter utilises data collected from children who took part either IMAGINE-1 or IMAGINE-2.

Like ECHO, children are recruited via NHS medical genetics clinics, charities, social media and the IMAGINE-ID website. To take part, families first complete an online questionnaire to provide behavioural, developmental, and medical information about their child, aged 4 years or older. This first stage of data collection is coordinated by UCL (Workstream 1). The Cardiff team then recontact parents of children aged 6-19 who have a medically confirmed ND CNV of interest and offer them the opportunity to take part in a follow up assessment (Workstream 2). If families agree, they then complete the same detailed assessments and questionnaires used in ECHO. As in ECHO, siblings without the ND CNV were also invited to take part.

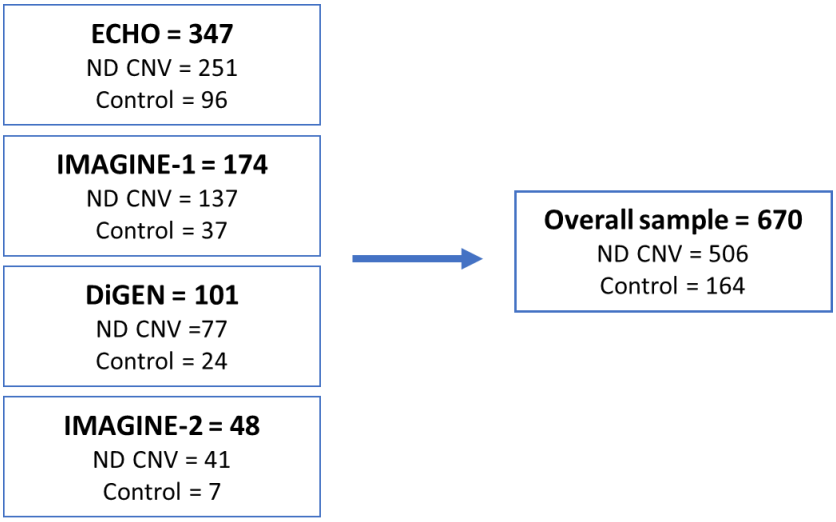
DiGEN: Dissecting the effects of genomic variants on neuro-behavioural dimensions in CNVs enriched for neuropsychiatric disorders study

DiGEN is an ongoing study, established in 2019 and led by Principal Investigators at Cardiff University and the University of Pennsylvania. It focuses on recruiting children and young people aged 7-17 with 22q11.2 and 16p11.2 deletion and duplication syndromes. Again, unaffected siblings are also invited to take part. Recruitment methods are like those outlined above and participants complete the same assessments administered in the ECHO and IMAGINE studies, meaning researchers can combine the datasets and explore their topics of interest in a larger sample than any one of the cohorts can provide alone.

### 2.4.2 Sample

A total of 670 children were included (506 children with an ND CNV, and 164 unaffected siblings). **Figure 2.1** provides a flow diagram which illustrates the number of participants included from each of the above studies.

*Figure 2.1 The respective number of participants from each study who comprised the overall sample.*




All children needed to have bullying data available and had to be attending either a primary or secondary school at the time of assessment.

Additional criteria for children with a ND CNV was confirmation of a ND CNV known to be associated with neurodevelopmental and psychiatric conditions (Kendall et al., 2019). **Table 2.1** presents a breakdown the ND CNVs included in this analysis. The presence of a ND CNV was confirmed by in-house genotyping of DNA samples (blood or saliva) provided by participants. The genotyping was completed at Cardiff University's Division of Psychological Medicine and Clinical Neurosciences (DPMCN). If the child failed to provide a genetic sample of sufficient quality or failed to provide one at all, the presence of the ND CNV was confirmed by a medical genetics report.

*Table 2.1 Breakdown of ND CNV group by ND CNV type.*

| ND CNV | *n* | *%* |
|---|---|---|
| 1q21.1 dup | 20 | 3.9 |
| 1q21.1 del | 21 | 4.1 |
| 3q29 del | 5 | 1.0 |
| 15q11.2 del | 46 | 9.1 |
| 15q13.3 dup | 12 | 2.4 |
| 15q13.3 del | 18 | 3.6 |
| 16p11.2 dup | 34 | 6.7 |
| 16p11.2 del | 77 | 15.2 |
| 16p11.2 distal del | 12 | 2.4 |
| 16p13.11 del | 5 | 1.0 |
| 22q11.2 dup | 40 | 2.9 |
| 22q11.2 del | 150 | 29.6 |
| 22q11.2 distal del | 7 | 1.4 |
| Kleefstra | 10 | 2.0 |
| NRXN1 | 19 | 3.7 |
| TAR dup | 12 | 2.4 |
| More than one priority ND CNV | 18 | 3.6 |
| **Total** | **506** | **100** |

### 2.4.3 Measures

Demographics

Demographic data were collected for all participants via a self-report questionnaire completed by the child's care giver and included children's age and biological sex, their mother's educational qualifications and family income. The latter two variables provided measures of socioeconomic status. School data collected included the type of school children attended: mainstream, special or 'other'. Those who were coded as 'other' were children who attended fee-paying schools. For children with a genetic condition, there was information for a proportion of them regarding whether their condition was inherited or appeared *de novo*.

The Strengths and Difficulties Questionnaire (SDQ)

The SDQ (Goodman, 1997) is a 25-item screening questionnaire used in clinical and research settings to measure the behavioural, emotional and social functioning of children and young people between 4-16 years old "over the last six months or this

school year". It is quick to complete, and focuses on both a child's strengths and weakness, not just the latter. Parallel versions of the questionnaire exist for parents, teachers and young people. The SDQ is a very well established measure and has high levels of validity and inter-rater reliability (Goodman, 1997).

The 25 items within the questionnaire are divided equally across five subscales: emotional problems, conduct problems, hyperactivity, peer problems and prosocial subscales. Respondents answer each item on a Likert Scale, choosing one of three options: "not true", "somewhat true" or "certainly true". Each response option has a corresponding numerical score and item scores within each subscale are added together to give a total score for that subscale, with higher scores indicating more difficulties. The sum of all the items from the emotional problems, conduct problems, hyperactivity and peer problems subscales provide the overall SDQ score for the child.

Within the peer relationships subscale, parents are asked to report how true it is that their child is "Picked on or bullied by other children". To measure bullying by peers, I used parent's responses to this item. This item from the SDQ was used by Rowley et al. (2012) to measure the bullying experiences of children with ASD. Children reported as 'not true' by their parent were coded as 'not bullied' and children reported as either 'somewhat true' or 'certainly true' were coded as 'bullied'. By recoding in this way, the sub-sample of participants within each response level was better populated, which limited the skewness of my results, particularly for later Aim 2 and Aim 3 analysis, in which participants were further categorised by whether they met criteria for the neurodevelopmental risk factors.

Prior to the COVID-19 pandemic, I had planned to use data collected by a questionnaire I had designed to capture children's school experiences to measure bullying (termed the 'School Experiences Questionnaire' (SEQ)). Child and parent versions of the questionnaire were designed and included questions about bullying experienced and perpetrated by the child. These were administered to families participating in the ongoing IMAGINE-2 and DiGEN studies.

Items within the SEQ used to measure bullying experiences included items from the Child and Adolescent Psychiatric Assessment (CAPA) (Angold et al., 1995) (see

below) and the Bullying and Friendship Interview Schedule Questionnaire (BFQ) (Wolke et al., 2001). **Table 2.2** presents the questions presented to participants in the child and parent versions of the SEQ which would have been used to measure children's bullying experiences had the SDQ not been used. However, due to government guidance in response to the pandemic to social distance, as well as nationwide lockdowns, there was a period in which data collection by the research team ceased. Thus, I decided to use the SDQ item, which had been completed by families who had taken part in the ECHO, IMAGINE (1 & 2) and DiGEN studies, to investigate bullying in a larger sample.

*Table 2.2 Bullying experienced questions in the School Experiences Questionnaire*

**Definition of bullying provided in the questionnaires:**
We say a young person is being bullied when they are made fun of, physically hurt, or scared by someone on purpose.
We say that a young person is bullying when they upset someone on purpose or try to make someone do something they don't want to do by scaring or hurting them.
We will ask you whether you have bullied or been bullied by other young people.
By 'other young people', we mean other children/adolescents, who you see either inside or outside of school.
We do **not** count bullying of siblings or being bullied by siblings in this questionnaire.

| | **Child Questionnaire** | **Parent Questionnaire** |
|---|---|---|
| CAPA | 1. Have you ever been teased or bullied at all by other young people?<br>2. Have you been teased or bullied in the last 3 months?<br>3. Have you been teased or bullied more than other children?<br>4. Have other young people ever been mean to you?<br>5. Have other young people been mean to you in the last 3 months? | 1. Has your child ever been teased or bullied at all by other young people?<br>2. Has your child been teased or bullied in the last 3 months?<br>3. Has your child been teased or bullied more than other children?<br>4. Have other young people ever been mean to your son/daughter?<br>5. Have other young people been mean to your child in the last 3 months? |
| School Relationships Questionnaire | 6. Have other young people ever taken any of your things (personal belongings) from you without you telling them that they could?<br>7. Have you ever been threatened or pressured into doing something (blackmailed) you didn't want to do by other young people?<br>8. Have you ever been hit or beaten up by other young people?<br>9. Have other young people ever called you nasty names?<br>10. Have other young people ever not wanted to hang around with you to make you upset?<br>11. Have other young people ever said they would not be friends with you anymore?<br>12. Have other young people said they would tell-tale on you?<br>13. Have other young people ever told lies, said nasty things, or told stories about you that were not true? | 6. Have other young people ever taken personal belongings from your child?<br>7. Has your child ever been threatened or blackmailed by other young people?<br>8. Has your child ever been hit or beaten up by other young people?<br>9. Have other young people ever called your child nasty names?<br>10. Have other young people ever not wanted to hang around with your child to make them upset?<br>11. Have other young people ever said they would not be friends with your child anymore?<br>12. Have other young people ever said they would tell-tale on your child?<br>13. Have other young people ever told lies, said nasty things, or told stories about your child that were not true? |

| | | |
|---|---|---|
| | 14. Have other young people ever spoilt activities e.g. sports games or class activities on purpose to make you upset? | 14. Have other young people ever spoilt activities e.g. sports games or class activities on purpose to make your child upset? |
| Additional questions | 15. If you have experienced any of the things we have just discussed, where were you when these events have taken place?<br>16. In what ways have other young people bullied you?<br>17. Does your school know that you have been bullied?<br>18. Have you ever refused to go to school because you have been bullied? | 15. If your child has experienced any of the above, where was your child when these events have taken place?<br>16. If your child has been bullied, in what ways have other young people bullied your child?<br>17. When other young people have bullied your child, what have you tended to do about it?<br>18. When other young people have bullied your child, what has your child tended to do about it?<br>19. Why do other young people bully your child?<br>20. Is your child's school aware that your child has been bullied?<br>21. If your child has been bullied, have you been happy with how your child's school addressed this problem? Please briefly explain your answer. |

The items from the CAPA and the BFQ were chosen because both are widely used measures and therefore would enable comparison of experiences within this group to community samples. For example, the Great Smoky Mountain Study (Costello et al., 1996), a large longitudinal cohort study, also used the CAPA to measure bullying (Copeland et al., 2013). The CAPA was also chosen partly for consistency purposes as instruments used by the ECHO, IMAGINE and DiGEN studies already collected data from participants using other sections of the CAPA. The BFQ was chosen to supplement information collected with the CAPA because the data collected with the former would provide a more detailed picture as to the relational and direct bullying children experienced at school compared to what the CAPA could provide alone. The BFQ was not specifically designed to be used with populations with

neurodevelopmental conditions (although previous studies have reported its use in children with cognitive impairment (Liu et al., 2021)), or parents, and so it should not be assumed as a valid and reliable measure of bullying within children with a ND CNV and their parents without piloting it within this population first. At present, the BFQ has not been formally piloted and therefore future research should seek to do so.

### Child and Adolescent Psychiatric Assessment

The CAPA was used to measure the presence of ADHD and PE. The CAPA is a gold standard interviewer-based research-diagnostic psychiatric assessment. Interviewers ask a series of questions to gather a detailed picture of the child's lifetime experiences, as well as those occurring 3 months prior to interview. Following completion of the interview, the respondent's answers are coded to determine whether the child meets the threshold for various psychiatric conditions according to DSM-IV-TR (American Psychological Association, 2000) diagnostic criteria.

Separate versions of the CAPA exist: 1) The CAPA: Parent Interview Omnibus Version ('Parent CAPA') which is to be completed with a primary caregiver about their child and 2) The CAPA: Child Interview Omnibus Version ('Child CAPA') which is to be completed with the child. Both versions are largely similar in terms of organisation and the items covered, with a few exceptions. The child CAPA does not include a measure of ADHD because child self-reports of ADHD are typically unreliable (Angold & Costello, 2000). Additionally, the parent version includes a truncated version of the substance use and psychosis sections compared to the child CAPA.

Psychology Research Assistants were trained extensively in administering the CAPA. The team also had input from a consultant child and adolescent psychiatrist when coding interviews. Interviews were audiotaped with participants' consent for the research team to hold consensus meetings to ensure inter-coder reliability.

Children were coded 'yes' or 'no' for meeting diagnostic criteria for ADHD based on their parent's responses to the ADHD items of the Parent CAPA.

Children were coded ‘yes’ or ‘no’ for PE based on parent and child responses to the Psychotic Disorders section of their relevant version of the CAPA. As noted already, the Psychotic Disorders section of the parent CAPA is less extensive compared to that of the child CAPA (see **Table 2.3**). The psychotic symptoms measured by the child CAPA fall under 3 screener questions to shorten the length of time taken to complete the interview with children who do not report PE**.** The child CAPA screener questions were only coded as positive if the presence of any of the respective symptoms under the screener were confirmed during consensus reliability meetings. To score positive for PE, children had to score positive for any of the parent reported items or any of the child reported screener items for psychosis.

*Table 2.3 Items within the psychosis sections of the parent and child versions of the CAPA*

| **Parent CAPA** | **Child CAPA** |
|---|---|
| - **Delusions and delusional interpretations**<br>- **“Psychotic” abnormalities of thought at speech**<br>- **Idiosyncratic behaviour**<br>- **Sensory changes and hallucinations**<br>- **Temporal co-occurrence delusions or hallucinations with mood disorder** | - **Perceptual disorders and hallucinations**<br>  - Changes in perception (déjà vu/jamais vu/ derealisation/depersonalisation)<br>  - Changed perception<br>  - Changed perception of time<br>  - Delusional mood<br>  - Hallucinations (auditory/visual/occurring as part of seizure/other)<br>- **Psychotic abnormalities in thought processes**<br>  - Thought intrusion/insertion<br>  - Thought broadcast or thought sharing<br>  - Thought echo or commentary<br>  - Delusions of thoughts being read<br>- **Delusions**<br>  - Delusions of control<br>  - Delusions of reference<br>  - Delusions of persecution<br>  - Delusions of assistance<br>  - Delusions of guilt<br>  - Delusions of depersonalisation or nihilism<br>  - Hypochondrial delusions<br>  - Simple delusions concerning appearance<br>  - Delusions of grandiose ability or identity<br>  - Primary delusions |

*Items in **bold** are the items used in my analysis to determine whether children met criteria for psychotic experiences. Child CAPA items in bold represent the child CAPA screener questions.*

Symptom count measures of ADHD and PE were also derived. The symptom count measure for ADHD comprised the total number of positive ADHD symptoms children met criteria for as reported by parents in the CAPA interview (i.e., overall ADHD symptom count). The maximum number of symptoms children could score was 18. Several participants (N = 198/670, 29.6%) had missing data for at least one of the ADHD CAPA items. For those with missing data for ≤10% of the items (N = 20/198, 10.1%), I calculated the mean total symptom count for the items they did have data for and pro-rated this value for each missing item. Participants with missing data for >10% (178/198, 89.9%) of the items were excluded from this analysis.

To derive the symptom count measure of PE (i.e., children's overall PE symptom count), I added the total number of parent reported positive PE symptoms to the total number of child reported positive PE screener items. Children could score positive for a maximum of 8 PEs (5 parent reported and 3 child reported). Children with any missing items were excluded from the analysis (N = 136/670, 20.4%) because children with even just one item missing had >10% of their data missing.

Social Communication Questionnaire (SCQ)

I used information provided by parents via the SCQ (Rutter, 2003) to derive a measure of ASD traits. The SCQ is not a diagnostic tool, but a widely used screening measure for the presence of ASD, based on the Autism Diagnostic Interview-Revised (Rutter et al., 1994), and has been proven effective at discriminating between children with and without probable ASD (Berument et al., 1999).

Responses to the questionnaire are scored, with total scores ranging from 0 to 39. A total score of 15 or more is indicative of ASD, however further clinical evaluation would be needed to conclude whether a formal ASD diagnosis should be given. Children who scored <15 were coded 'no' for indicative ASD. Children who scored ≥15 were coded 'yes' for indicative ASD.

The symptom count measure of indicative ASD was derived by calculating the total SCQ score (i.e., the number of items children scored positively for). Missing data were handled in the same way as for the symptom count measure of ADHD. A total of 117/670 (17.5%) of children had missing data for the indicative ASD items. For those with missing data for ≤10% of the items (N = 93/117, 79.5%), I calculated the

mean of the total symptom count for the items they did have data for and pro-rated this mean value for each missing item. Participants with missing data for >10% (n = 24/117 = 20.5%) of the items were excluded from this analysis.

Wechsler Abbreviated Scale of Intelligence (WASI)

The WASI (Wechsler, 1999) was used to derive children's IQ. The WASI can be administered to individuals aged 6-89 and consists of four subtests. Two of the subtests measure Verbal IQ (VIQ) and the remaining two measure Performance IQ (PIQ). The raw performance scores for each of the subtests are standardised and converted into age-adjusted t-scores as per the normative sample tables within the WASI manual (Wechsler, 1999). The total of the two VIQ subtest t-scores generates the overall VIQ score, providing a measure of crystallised intelligence. The total of the two PIQ subtest t-scores generates the PIQ score and provides a measure of fluid intelligence.

Full-scale IQ (FSIQ) is then derived by either the standardised combined total of one VIQ subtest score and one PIQ subtest score (FSIQ-2) or the combined total of both VIQ subtest scores and both PIQ subtest scores (FSIQ-4). Children who were seen face-to-face by a researcher completed all four tests. However, over the COVID-19 lockdown period, children took part virtually, via online Zoom calls. In these circumstances, children completed the two VIQ subtests, but only one of the two PIQ subtests because the remaining subtest required in-person interaction between the researcher and participant. Therefore, these children's FSIQ score represented their FSIQ-2 score, whereas the FSIQ score of children who were seen in person represented their FSIQ-4 score. This may raise concerns about the comparability of FSIQ-2 and FSIQ-4 scores however, 'very small differences are noted on the subtest level' (Wolraich et al., 2008). FSIQ-2 scores were used for a total of 77 children (11.5%).

Lower VIQ, PIQ and FSIQ scores are indicative of poorer cognitive functioning and a FSIQ score <70 signifies the presence of intellectual disability. Thus, children with an FSIQ score <70 were coded positively for ID. Those with a score of ≥70 were coded as not having ID. I used children's FSIQ score for my 'symptom count' measure of ID.

Neurodevelopmental burden

To derive a measure of neurodevelopmental burden I calculated the total number of the neurodevelopmental 'conditions' children met criteria for (ADHD, PE, indicative ASD and ID). Thus, children's neurodevelopmental burden score could range from 0-4.

### 2.4.4 Analysis

Data were cleaned in SPSS version 27 and then loaded into RStudio to perform statistical analyses. I conducted chi-square tests to determine statistical group differences between the ND CNV group and the control group regarding categorical characteristic variables (e.g., gender distribution). Differences in continuous characteristics (e.g., age, neurodevelopmental symptom counts) were statistically compared using Mann-Whitney U test because Shapiro-Wilk test revealed that none of the continuous variables followed a normal distribution.

**Aim 1: Are children with a ND CNV more likely to experience bullying compared to unaffected sibling controls?**

I conducted chi-square analysis to compare the proportion of children with a ND CNV reported as bullied to the proportion of unaffected sibling controls.

Then I ran a generalised linear-mixed model (GLMM) to establish if the ND CNV group were more likely to be bullied whilst controlling for child's age and biological sex, mother's education (educated to university undergraduate or postgraduate degree level: yes or no) and family relatedness between the ND CNV and sibling groups. Affected status (ND CNV carrier vs control), age, sex and mother's education were all entered into the model as fixed effects and family ID (i.e., the family a child belonged to) was entered into the model as a random effect to control for relatedness between siblings.

As there was only data available for 90.1% of the sample regarding their family income (90.3% of the ND CNV group, 89.6% of the control group), I conducted a follow up sensitivity analysis in which family income was added as an additional fixed effect (approximate family income ≤£19,999: yes or no).

GLMMs were chosen as the appropriate statistical test given the ability to control for family relatedness when running these models. This would not have been possible if I had conducted a standard logistic regression. GLMMs have been used in other studies for the same reason (Chawner et al., 2019).

**Aim 2: What are the individual and contextual risk factors associated with bullying in children with an ND CNV?**

To investigate the impact of individual and contextual risk factors on bullying experience, I ran a series of logistic regressions in children with ND CNVs only. A given diagnostic neurodevelopmental measure (the presence of ADHD, indicative ASD, PE or ID), neurodevelopmental symptom count measure (ADHD, indicative ASD and PE symptom counts and FSIQ) or neurodevelopmental burden were included in a respective model as the independent variable (i.e., a separate regression was performed for each neurodevelopmental measure). Child's age and sex, and mother's education were all added into the models as covariates. Then I reran the model with inheritance status entered as the independent variable (i.e., did children have an inherited or *de novo* ND CNV), and then once more with school placement entered as the independent variable (i.e., did children attend a mainstream or special school). Again, a sensitivity analysis was performed after each of the regressions to consider the impact of family income.

Most logistic regression assumptions were met for all of the models. Firstly, the dependent variable (bullied: yes or no) was binary in nature. Secondly, no outliers were detected within any of the independent variables. I checked for the presence of multicollinearity between the independent variables by using variance inflation factor (VIF). VIF values above 5 indicate that a correlation exists between two or more variables which could impact the reliability of the regression's output (Menard, 2001). VIF for all the predictors within the model did not surpass 5. However, the assumption of linearity between child's age and bullying was violated in the following models: ADHD model, ADHD symptom count model, indicative ASD model, indicative ASD symptom count model and the inheritance status model. To ensure this violation did not impact the results, I conducted an additional regression into which I added the log transformation of age into the model and compared the findings of the regressions.

Next, to ensure the findings of the regressions were not due to collider bias, I ran a series of GLMMs to consider each of the neurodevelopmental risk factors when both the ND CNV and unaffected sibling group were included in the analysis. In each of the models, the respective neurodevelopmental risk factor was included as a fixed effect, along with affected status (ND CNV carrier vs unaffected sibling), the child's age and sex, and their mother's education. Family ID was entered into the model as a random effect. Parent reported bullying was entered into the model as the dependent variable. Again, each GLMM was followed up with a sensitivity analysis whereby family income was included as an additional fixed effect.

None of the siblings met criteria for ID or attended a special school so I could not perform the associated GLMMs. Additionally, given that unaffected siblings did not have an ND CNV, I could not perform the GLMM for inheritance status.

**Aim 3: To what extent do neurodevelopmental conditions and traits explain the risk of bullying in children with a ND CNV?**

I reran the GLMMs as described under Aim 2 but added an interaction effect between the neurodevelopmental risk factor in the model and participant affected status to investigate whether children with a ND CNV and a given neurodevelopmental risk factor were at increased risk for bullying compared to unaffected siblings who had the same neurodevelopmental risk factor.

Post-hoc Power Analysis

I conducted post-hoc power analysis to determine the power of the study to detect associations between the independent and dependent variables in each regression model presented in this chapter. To do this I used the 'pwr' package in R and I assumed a significance level of 0.05 throughout. Odds ratios and coefficients selected by significance are subject to inflation due to the "winner's curse" phenomenon, thus, I used the lower bound of the 95% confidence interval in my calculations to be conservative.

Multiple Testing Correction

All statistical analyses conducted for this chapter were corrected for multiple testing using the Benjamini–Hochberg false discovery rate (BH-FDR) method. An alpha of 0.05 was used throughout. BH-FDR has been used in other studies investigating populations with ND CNVs (Chawner et al., 2023).

## 2.5 Results

### Sample characteristics

A breakdown of the sample's characteristics and between group differences can be seen in **Table 2.4**. The age range of the sample was 6-16 years.

*Table 2.4 Sample characteristics*

| | Total Sample | | | ND CNV Group | | | Control Group | | | Statistics | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N (670) | Mean (SD) | N (%) | N (506) | Mean (SD) | N (%) | N (164) | Mean (SD) | N (%) | z | $X^2$ *(df)* | p-value |
| Child's age | 670 | 9.71 (2.74) | | 506 | 9.47 (2.77) | | | 10.45 (2.50) | | -4.32 | | **<.001** |
| Sex: | 670 | | | 506 | | | 164 | | | | 6.23 *(1)* | **0.01** |
| Male | 379 | | 56.6% | 300 | | 59.3% | 79 | | 48.2% | | | |
| Female | 291 | | 43.4% | 206 | | 40.7% | 85 | | 51.80% | | | |
| Mother's education: | 611 | | | 468 | | | 143 | | | | 0.77 *(1)* | 0.41 |
| University degree | 187 | | 30.6% | 139 | | 29.7% | 48 | | 33.6% | | | |
| No university degree | 424 | | 69.4% | 329 | | 70.3% | 95 | | 66.4% | | | |
| Family income: | 604 | | | 457 | | | 147 | | | | 5.20 *(1)* | **0.02** |
| ≤£19,999 | 163 | | 27.0% | 134 | | 29.3% | 29 | | 19.7% | | | |
| ≥£19,999 | 441 | | 73.0% | 323 | | 70.7% | 118 | | 80.3% | | | |
| Inherited status: | 317 | | | 317 | | | - | | | | - | - |
| De novo | 139 | | 43.8% | 139 | | 43.8% | - | | - | | | |
| Inherited | 178 | | 56.2% | 178 | | 56.2% | - | | - | | | |
| | | | | | | | | | | | | |
| Neurodevelopmental characteristics: | | | | | | | | | | | | |
| ADHD | 640 | | | 490 | | | 150 | | | | 75.15 *(1)* | **<.001** |
| Yes | 223 | | 34.8% | 215 | | 43.9% | 8 | | 5.3% | | | |
| No | 417 | | 65.2% | 275 | | 56.1% | 142 | | 94.7% | | | |
| Indicative ASD | 644 | | | 485 | | | 159 | | | | 71.52 *(1)* | **<.001** |
| Yes | 317 | | 49.2% | 285 | | 58.8% | 32 | | 20.1% | | | |
| No | 327 | | 50.8% | 200 | | 41.2% | 127 | | 79.9% | | | |
| Psychotic experiences | 548 | | | 419 | | | 129 | | | | 4.62 *(1)* | **0.03** |
| Yes | 118 | | 21.5% | 99 | | 23.6% | 19 | | 14.7% | | | |
| No | 430 | | 78.5% | 320 | | 76.4% | 110 | | 85.3% | | | |
| ID | 560 | | | 434 | | | 126 | | | | | |
| Yes | 105 | | 18.8% | 105 | | 24.2% | 0 | | 0.0% | | 37.52 *(1)* | **<.001** |
| No | 455 | | 81.3% | 329 | | 75.8% | 126 | | 100.0% | | | |
| Neurodevelopmental burden | 471 | 1.21 (1.07) | | 357 | 1.48 (1.04) | | 114 | 0.39 (0.66) | | -9.84 | | **<.001** |
| ADHD symptom count | 493 | 6.39 (5.54) | | 370 | 7.95 (5.19) | | 123 | 1.71 (3.58) | | -12.03 | | **<.001** |
| Indicative ASD symptom count | 646 | 15.23 (9.28) | | 486 | 17.63 (8.26) | | 160 | 7.95 (8.38) | | -11.75 | | **<.001** |
| PE symptom count | 534 | 0.26 (0.61) | | 405 | 0.29 (0.66) | | 129 | 0.16 (0.41) | | -1.70 | | 0.90 |
| FSIQ | 582 | 84.72 (17.13) | | 434 | 79.02 (13.96) | | 148 | 101.44 (14.47) | | -13.40 | | **<.001** |

*Table 2.4 continued*

| | Total Sample | | | ND CNV Group | | | Control Group | | | Statistics | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N (670) | Mean (SD) | N (%) | N (506) | Mean (SD) | N (%) | N (164) | Mean (SD) | N (%) | z | $X^2$ *(df)* | p-value |
| School placement: | 444 | | | 329 | | | 115 | | | | 31.54 *(1)* | **<.001** |
| Mainstream | 369 | | 83.1% | 254 | | 77.2% | 115 | | 100.0% | | | |
| Special | 75 | | 16.9% | 75 | | 22.8% | 0 | | 0.0% | | | |
| | | | | | | | | | | | | |
| Bullied: | 670 | | | 506 | | | 164 | | | | 30.97 *(1)* | **<.001** |
| Yes | 310 | | 46.30% | 265 | | 52.4% | 45 | | 27.4% | | | |
| No | 360 | | 53.70% | 241 | | 47.6% | 119 | | 72.6% | | | |

**Aim 1: Are children with a ND CNV more likely to experience peer bullying compared to unaffected sibling controls?**

A larger proportion of children with an ND CNV were reported as bullied compared to children in the unaffected sibling group (52.4% vs 27.4% respectively; $X^2$ = 30.97, p = <.001). **Table 2.5** shows the number and percentage of children with a ND CNV who were reported as bullied broken down by ND CNV type.

*Table 2.5 Number and percentage of children with an ND CNV reported as bullied broken down by ND CNV type.*

| ND CNV | *Bullied (n)* | *%* |
|---|---|---|
| 1q21.1 dup | 14/20 | 70.0 |
| 1q21.1 del | 15/21 | 71.4 |
| 3q29 del | 2/5 | 40.0 |
| 15q11.2 del | 23/46 | 50.0 |
| 15q13.3 dup | 8/12 | 66.7 |
| 15q13.3 del | 10/18 | 55.6 |
| 16p11.2 dup | 21/34 | 61.8 |
| 16p11.2 del | 35/77 | 45.5 |
| 16p11.2 distal del | 8/12 | 66.7 |
| 16p13.11 del | 1/5 | 20.0 |
| 22q11.2 dup | 26/40 | 65.0 |
| 22q11.2 del | 73/150 | 48.7 |
| 22q11.2 distal del | 3/7 | 42.9 |
| Kleefstra | 3/10 | 30.0 |
| NRXN1 | 7/19 | 36.8 |
| TAR dup | 5/12 | 41.7 |
| More than one priority ND CNV | 11/18 | 61.1 |

Children with a ND CNV were more likely to be bullied compared to siblings after controlling for children's sex and age, their mother's level of education and family relatedness (OR = 4.87, 95% CI 2.74 – 8.64, p = <.001, power = 1.00). Age was the only other significant predictor of bullying. Children's odds of being bullied increased as they increased in age by one year (OR = 1.17, 95% CI 1.07 – 1.27, p = <.001). Children's sex, nor their mother's level of education predicted bullying (sex: OR = 0.76, 95% CI 0.51 – 1.14, p = .18; mother's education: OR = 0.85, 95% CI 0.55 – 1.31, p = .46). Results were unchanged after sensitivity analysis.

**Aim 2: What are the individual and contextual risk factors associated with bullying in children with an ND CNV?**

I ran logistic regression models to investigate whether neurodevelopmental risk factors, inheritance status, school placement were associated with bullying experiences. Then, I ran a series of GLMMs including both the ND CNV group and the sibling group to check that findings were not the result of collider bias.

**Table 2.6** presents the findings of the logistic regressions.

Children were at greater odds for bullying if they met diagnostic criteria for ADHD (OR = 2.08, 95% CI 1.41 - 3.10, p = <.001, power = .44), indicative ASD (OR = 1.81, 95% CI 1.22 - 2.70, p = <.05, power = .41) and PE (OR = 3.55, 95% CI 2.07 – 6.33, p = <.001, power = .85) respectively. Higher symptom counts for these conditions were also positively associated with bullying experience (ADHD symptom count: OR = 1.08, 95% CI 1.03 – 1.13, p = <.05; power = .06; indicative ASD symptom count: OR = 1.05, 95% CI 1.02 – 1.07, p = <.001, power = .06; PE symptom count: OR = 2.31, 95% CI 1.52 – 3.70, p = <.001, power = .98). Finally, higher neurodevelopmental burden was positively associated with bullying (OR = 1.66, 95% CI 1.32 – 2.11, p = <.001, power = .68). Neither meeting criteria for ID, or IQ predicted bullying (ID: OR = .64, 95% CI .39 – 1.03, p = .09, power = .98; IQ: OR = 1.01, 95% CI 1.00 - 1.03, p = .14, power = .05).

Age was a significant predictor of bullying in each of the models (ADHD model: OR = 1.17, 95% CI 1.09 - 1.26, p = <.001; ADHD symptom count model: OR = 1.19, 95% CI 1.10 – 1.30, p = <.001; indicative ASD model: OR = 1.16, 95% CI 1.08 - 1.25, p = <.001; ASD symptom count model: OR = 1.17, 95% CI 1.09 – 1.26, p = <.001; PE model: OR = 1.14, 95% CI 1.06 - 1.24, p = <.001; PE symptom count model: OR = 1.16, 95% CI 1.07 – 1.26, p = <.001; ID model: OR = 1.20, 95% CI 1.11 - 1.31, p = <.001; FSIQ model: OR = 1.19, 95% CI 1.10 – 1.29, p = <.001; neurodevelopmental burden model: OR = 1.17, 95% CI 1.08 – 2.11, p = <.001).

In the inheritance status model, age was the only significant predictor of bullying (OR = 1.13, 95% CI 1.04 - 1.23, p = <.05). Inheritance status was not significantly associated with bullying (OR = 1.47, 95% CI 0.92 - 2.37, p = .11, power = .84).

School placement was not a significant predictor of bullying (OR = 0.61, 95% CI 0.35 - 1.06, p = .08, power = 1.00), however age remained significant (OR = 1.12, 95% CI 1.03 - 1.23, p = <.05).

Sensitivity analyses did not change the results of any of the models, nor did substituting age with the log transformation of age into the relevant models.

*Table 2.6 Results of logistic regression analyses on the effect of neurodevelopmental characteristics, inheritance status and school type on bullying experience whilst controlling for demographic factors in children with a ND CNV only*

| | | Bullied | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Bullied | Not bullied | OR | 95% conf.int | SE | z-score | p-value | $R^2$ |
| A) | 455 | 242 | 213 | | | | | | 0.05 |
| **Meets criteria for ADHD** | | | | 2.08 | 1.41 - 3.10 | 0.20 | 3.64 | **<.001** | |
| Age | | | | 1.17 | 1.09 - 1.26 | 0.04 | 4.34 | **<.001** | |
| Female | | | | 0.81 | 0.55 -1.20 | 0.20 | -1.05 | 0.30 | |
| Mother has a university degree | | | | 1.05 | 0.69 - 1.60 | 0.22 | 0.21 | 0.83 | |
| | | | | | | | | | |
| B) | 348 | 185 | 163 | | | | | | 0.05 |
| **ADHD symptom count** | | | | 1.08 | 1.03 - 1.13 | 0.02 | 3.25 | **<.05** | |
| Age | | | | 1.19 | 1.10 - 1.30 | 0.04 | 4.06 | **<.001** | |
| Female | | | | 0.98 | 0.62 - 1.54 | 0.23 | -0.10 | 0.92 | |
| Mother has a university degree | | | | 1.29 | 0.76 - 2.19 | 0.27 | 0.95 | 0.34 | |
| | | | | | | | | | |
| C) | 448 | 236 | 212 | | | | | | 0.04 |
| **Meets criteria for indicative ASD** | | | | 1.81 | 1.22 - 2.70 | 0.20 | 2.94 | **<.05** | |
| Age | | | | 1.16 | 1.08 - 1.25 | 0.04 | 4.13 | **<.001** | |
| Female | | | | 0.91 | 0.61 - 1.34 | 0.20 | -0.48 | 0.63 | |
| Mother has a university degree | | | | 1.11 | 0.72 - 1.70 | 0.22 | 0.46 | 0.64 | |
| | | | | | | | | | |
| D) | 449 | 235 | 214 | | | | | | 0.05 |
| **Indicative ASD symptom count** | | | | 1.05 | 1.02 - 1.07 | 0.01 | 3.59 | **<.001** | |
| Age | | | | 1.17 | 1.09 - 1.26 | 0.04 | 4.30 | **<.001** | |
| Female | | | | 0.93 | 0.63 -1.38 | 0.20 | -0.35 | 0.73 | |
| Mother has a university degree | | | | 1.13 | 0.74 - 1.74 | 0.22 | 0.56 | 0.58 | |

*Table 2.6 continued*

| | | Bullied | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Bullied | Not bullied | OR | 95% conf.int | SE | z-score | p-value | $R^2$ |
| E) | 386 | 212 | 174 | | | | | | 0.08 |
| **Meets criteria for PE** | | | | 3.55 | 2.07 - 6.33 | 0.28 | 4.46 | **<.001** | |
| Age | | | | 1.14 | 1.06 - 1.24 | 0.04 | 3.37 | **<.001** | |
| Female | | | | 0.69 | 0.45 - 1.07 | 0.22 | -1.67 | 0.09 | |
| Mother has a university degree | | | | 1.14 | 0.72 - 1.83 | 0.24 | 0.57 | 0.57 | |
| | | | | | | | | | |
| F) | 374 | 203 | 171 | | | | | | 0.07 |
| **PE symptom count** | | | | 2.31 | 1.52 - 3.70 | 0.23 | 3.70 | **<.001** | |
| Age | | | | 1.16 | 1.07 - 1.26 | 0.04 | 3.69 | **<.001** | |
| Female | | | | 0.68 | 0.43 - 1.05 | 0.23 | -1.73 | 0.08 | |
| Mother has a university degree | | | | 1.14 | 0.71 - 1.84 | 0.24 | 0.56 | 0.58 | |
| | | | | | | | | | |
| G) | 400 | 219 | 181 | | | | | | 0.04 |
| **Meets criteria for ID** | | | | 0.64 | 0.39 - 1.05 | 0.25 | -1.76 | 0.09 | |
| Age | | | | 1.20 | 1.11 - 1.31 | 0.04 | 4.44 | **<.001** | |
| Female | | | | 0.86 | 0.57 - 1.30 | 0.21 | -0.70 | 0.48 | |
| Mother has a university degree | | | | 1.03 | 0.66 - 1.61 | 0.23 | 0.11 | 0.91 | |
| | | | | | | | | | |
| H) | 400 | 219 | 181 | | | | | | 0.04 |
| **FSIQ** | | | | 1.01 | 1.00 - 1.03 | 0.01 | 1.46 | 0.14 | |
| Age | | | | 1.19 | 1.10 - 1.29 | 0.04 | 4.35 | **<.001** | |
| Female | | | | 0.86 | 0.57 - 1.30 | 0.21 | -0.71 | 0.48 | |
| Mother has a university degree | | | | 1.03 | 0.66 - 1.62 | 0.23 | 0.15 | 0.88 | |

*Table 2.6 continued*

| | | Bullied | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Bullied | Not bullied | OR | 95% conf.int | SE | z-score | p-value | $R^2$ |
| I) | 329 | 181 | 148 | | | | | | 0.09 |
| **Neurodevelopmental burden** | | | | 1.66 | 1.32 - 2.11 | 0.12 | 4.21 | **<.001** | |
| Age | | | | 1.17 | 1.08 - 1.28 | 0.04 | 3.59 | **<.001** | |
| Female | | | | 0.76 | 0.47 - 1.22 | 0.24 | -1.14 | 0.25 | |
| Mother has a university degree | | | | 1.58 | 0.95 - 2.69 | 0.27 | 1.73 | 0.08 | |
| | | | | | | | | | |
| J) | 294 | 152 | 142 | | | | | | 0.03 |
| **Inheritance status** | | | | 1.47 | 0.92 - 2.37 | 0.24 | 1.61 | 0.11 | |
| Age | | | | 1.13 | 1.04 - 1.23 | 0.43 | 2.78 | **<.05** | |
| Female | | | | 0.94 | 0.58 - 1.52 | 0.24 | -0.25 | 0.80 | |
| Mother has a university degree | | | | 1.06 | 0.63 - 1.79 | 0.26 | 0.23 | 0.82 | |
| | | | | | | | | | |
| K) | 311 | 161 | 150 | | | | | | 0.02 |
| **Child attends special school** | | | | 0.61 | 0.35 - 1.06 | 0.29 | -1.73 | 0.08 | |
| Age | | | | 1.12 | 1.03 - 1.23 | 0.04 | 2.61 | **<.05** | |
| Female | | | | 0.86 | 0.54 - 1.36 | 0.24 | -0.66 | 0.51 | |
| Mother has university degree | | | | 0.84 | 0.52 - 1.36 | 0.25 | -0.70 | 0.48 | |

**Table 2.7** shows the number and percentage of children within the ND CNV and control group who were reported as bullied and not bullied relative to whether they met criteria for ADHD, indicative ASD, PE and ID. Results of the GLMMs are presented in **Table 2.8**.

Meeting criteria for ADHD, indicative ASD and PE respectively was associated with bullying experience (ADHD: OR = 2.23, 95% CI 1.43 – 3.49, p = <.001, power = .54; indicative ASD: OR = 2.03, 95% CI 1.30 – 3.17, p = <.05, power = .34; PE: OR = 3.52, 95% CI 1.83 – 6.79, p = <.001, power = .79). Children who scored positively for a higher number of symptoms for the same conditions were also more likely to be bullied (ADHD symptom count: OR = 1.09, 95% CI 1.04 – 1.14, p = <.001, power = .07; indicative ASD symptom count: OR = 1.06, 95% CI 1.03 – 1.09, p = <.001, power = .07; PE symptom count: OR = 2.34, 95% CI 1.41 – 3.87, p = <.001, power = .99). None of the unaffected siblings met criteria for ID so I could not run a GLMM with respect to ID, however IQ did not predict bullying (OR = 1.01, 95% CI 0.99 - 1.02, p = .70, power = .05). Higher neurodevelopmental burden was also positively associated with bullying (OR = 1.82, 95% CI 1.34 – 2.47, p = <.001, power = .91).

Children with a ND CNV were more likely to be victimised in each of the models (ADHD model: OR = 3.37, 95% CI 1.89 – 6.02, p = <.05, power = .85; ADHD symptom count model: OR = 2.52, 95% CI 1.32 – 4.80, p = <.05, power = .21; indicative ASD model: OR = 3.93, 95% CI 2.17 – 7.13, p = <.001, power = .96; indicative ASD symptom count mode: OR = 3.25, 95% CI 1.77 – 5.98, p = <.001, power = .77; PE model: OR = 4.91, 95% CI 2.47 – 9.76, p = <.001, power = .98; PE symptom count model: OR = 5.00, 95% CI 2.48 – 10.10, p = <.001, power = .98; FSIQ model: OR = 7.91, 95% CI 3.34 – 18.76, p = <.001, power = 1.00; neurodevelopmental burden model: OR = 3.24, 95% CI 1.48 – 7.10, p = <.05, power = .34).

Increased age was also positively associated with being bullied in each of the models (ADHD model: OR = 1.18, 95% CI 1.09 – 1.29, p = <.001; ADHD symptom count model: OR = 1.18, 95% CI 1.08 – 1.29, p = <.001; indicative ASD model: OR = 1.19, 95% CI 1.08 – 1.30, p = <.001; indicative ASD symptom count model: OR = 1.20, 95% CI 1.09 – 1.33, p = <.001; PE model: OR = 1.19, 95% CI 1.07 – 1.31, p = <.05; PE symptom count model: OR = 1.21, 95% CI 1.08 – 1.35, p = <.001; FSIQ

model: OR = 1.23, 95% CI 1.10 – 1.37, p = <.001; neurodevelopmental burden model: OR = 1.23, 95% CI 1.08 – 1.39, p = <.05).

Females were significantly less likely to be bullied in the PE and PE symptom count models (PE model: OR = 0.61, 95% CI 0.37 – 0.99, p = <.05; PE symptom count model: OR = 0.60, 95% CI 0.36 – 0.99, p = <.05).

Sensitivity analyses only revealed a change in findings within the PE and PE symptom count models whereby sex became non-significant in predicting bullying experience (PE model: OR = 0.64, 95% CI 0.39 - 1.05, p = .07; PE symptom count model: OR = 2.06, 95% CI 1.25 - 3.37, p = .08).

*Table 2.7 Number and percentage of 'bullied' and 'not bullied' children within the ND CNV and control groups relative to whether they meet criteria for ADHD, indicative ASD, PE and ID*

| | ND CNV Group | | Control Group | |
|---|---|---|---|---|
| | Bullied | Not bullied | Bullied | Not bullied |
| Meets criteria for: | | | | |
| ADHD | 133 (61.9%) | 82 (38.1%) | 5 (62.5%) | 3 (73.9%) |
| Indicative ASD | 164 (57.5%) | 121 (42.5%) | 13 (40.6%) | 19 (59.4%) |
| PE | 76 (76.8%) | 23 (23.2%) | 5 (26.3%) | 14 (73.7%) |
| ID | 54 (51.4%) | 51 (48.6%) | - | - |

*Table 2.8 Results of GLMM analyses exploring the effect of neurodevelopmental characteristics and having a ND CNV on bullying experience whilst controlling for demographic factors*

| | | Bullied | | | | | | |
|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Bullied | Not bullied | OR | 95% conf.int | SE | z-score | p-value |
| A) | 587 | 276 | 311 | | | | | |
| **Meets criteria for ADHD** | | | | 2.23 | 1.43 - 3.49 | 0.23 | 3.51 | **<.001** |
| Child has a ND CNV | | | | 3.37 | 1.89 - 6.02 | 0.30 | 4.11 | **<.001** |
| Age | | | | 1.18 | 1.09 - 1.29 | 0.04 | 3.90 | **<.001** |
| Female | | | | 0.71 | 0.48 - 1.07 | 0.21 | -1.63 | 0.10 |
| Mother has a university degree | | | | 0.97 | 0.62 - 1.50 | 0.22 | -0.14 | 0.89 |
| | | | | | | | | |
| B) | 460 | 215 | 245 | | | | | |
| **ADHD symptom count** | | | | 1.09 | 1.04 - 1.14 | 0.02 | 3.66 | **<.001** |
| Child has a ND CNV | | | | 2.52 | 1.32 - 4.80 | 0.33 | 2.81 | **<.05** |
| Age | | | | 1.18 | 1.08 - 1.29 | 0.05 | 3.59 | **<.001** |
| Female | | | | 0.87 | 0.57 - 1.34 | 0.22 | -0.62 | 0.53 |
| Mother has a university degree | | | | 1.24 | 0.76 - 2.02 | 0.25 | 0.86 | 0.39 |
| | | | | | | | | |
| C) | 587 | 271 | 316 | | | | | |
| **Meets criteria for indicative ASD** | | | | 2.03 | 1.30 - 3.17 | 0.23 | 3.11 | **<.05** |
| Child has a ND CNV | | | | 3.93 | 2.17 - 7.13 | 0.30 | 4.51 | **<.001** |
| Age | | | | 1.19 | 1.08 - 1.30 | 0.05 | 3.73 | **<.001** |
| Female | | | | 0.81 | 0.54 - 1.23 | 0.21 | -0.97 | 0.33 |
| Mother has a university degree | | | | 1.00 | 0.63 - 1.58 | 0.23 | -0.01 | 0.99 |
| | | | | | | | | |
| D) | 589 | 270 | 319 | | | | | |
| **Indicative ASD symptom count** | | | | 1.06 | 1.03 - 1.09 | 0.01 | 3.82 | **<.001** |
| Child has a ND CNV | | | | 3.25 | 1.77 - 5.98 | 0.31 | 3.79 | **<.001** |
| Age | | | | 1.20 | 1.09 - 1.32 | 0.05 | 3.80 | **<.001** |
| Female | | | | 0.84 | 0.55 - 1.29 | 0.22 | -0.78 | 0.44 |
| Mother has a university degree | | | | 0.99 | 0.62 - 1.59 | 0.24 | -0.02 | 0.98 |

*Table 2.8 continued*

| | | Bullied | | | | | | |
|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Bullied | Not bullied | OR | 95% conf.int | SE | z-score | p-value |
| E) | 498 | 241 | 257 | | | | | |
| **Meets criteria for PE** | | | | 3.52 | 1.83 - 6.79 | 0.34 | 3.75 | **<.001** |
| Child has a ND CNV | | | | 4.91 | 2.47 - 9.76 | 0.35 | 4.54 | **<.001** |
| Age | | | | 1.19 | 1.07- 1.31 | 0.05 | 3.26 | **<.05** |
| Female | | | | 0.61 | 0.37 - 0.99 | 0.25 | -2.01 | **<.05** |
| Mother has a university degree | | | | 1.02 | 0.60 - 1.72 | 0.27 | 0.07 | 0.94 |
| | | | | | | | | |
| F) | 486 | 232 | 254 | | | | | |
| **PE symptom count** | | | | 2.34 | 1.41 - 3.87 | 0.26 | 3.29 | **<.001** |
| Child has a ND CNV | | | | 5.00 | 2.48 - 10.10 | 0.36 | 4.49 | **<.001** |
| Age | | | | 1.21 | 1.08 - 1.35 | 0.05 | 3.44 | **<.001** |
| Female | | | | 0.60 | 0.36 - 0.99 | 0.26 | -2.01 | **<.05** |
| Mother has a university degree | | | | 1.01 | 0.59 - 1.74 | 0.27 | 0.05 | 0.96 |
| | | | | | | | | |
| G) | 529 | 250 | 279 | | | | | |
| **FSIQ** | | | | 1.01 | 0.99 - 1.02 | 0.01 | 0.70 | 0.49 |
| Child has a ND CNV | | | | 7.91 | 3.34 - 18.76 | 0.44 | 4.69 | **<.001** |
| Age | | | | 1.23 | 1.10 - 1.37 | 0.05 | 3.78 | **<.001** |
| Female | | | | 0.76 | 0.47 - 1.21 | 0.24 | -1.16 | 0.24 |
| Mother has a university degree | | | | 0.99 | 0.58 - 1.68 | 0.27 | -0.04 | 0.97 |
| | | | | | | | | |
| H) | 426 | 206 | 220 | | | | | |
| **Neurodevelopmental burden** | | | | 1.82 | 1.34 - 2.47 | 0.16 | 3.81 | **<.001** |
| Child has a ND CNV | | | | 3.24 | 1.48 - 7.10 | 0.40 | 2.94 | **<.05** |
| Age | | | | 1.23 | 1.08 - 1.39 | 0.06 | 3.26 | **<.05** |
| Female | | | | 0.68 | 0.40 - 1.19 | 0.28 | -1.35 | 0.18 |
| Mother has a university degree | | | | 1.41 | 0.76 - 2.60 | 0.31 | 1.09 | 0.28 |

**Aim 3: To what extent do neurodevelopmental conditions and traits explain the risk of bullying in children with ND CNV?**

I repeated the GLMMs as described under **Aim 2** but added an interaction effect between the relevant neurodevelopmental factor within the models and affected status. **Table 2.9** presents the results of the GLMMs with the added interaction effect. I did not conduct a follow up analysis with an interaction effect for IQ because IQ had not proven to be significantly predictive of bullying in the previous analysis.

When the interaction term was added, ADHD, indicative ASD and PE all became non-significant predictors of bullying experience (ADHD: OR = 3.85, 95% CI 0.58 – 25.55, p = .16, power = 1.00; indicative ASD: OR = 2.65, 95% CI 0.90 – 7.85, p = .08, power = .86; PE: OR = 0.96, 95% CI 0.22 – 4.32, p = .96, power = 1.00). Increased symptom count continued to increase children's odds of being bullied in the ADHD (OR = 1.23, 95% CI 1.06 - 1.42, p = <.05, power = .12), and the indicative ASD (OR = 1.07, 95% CI 1.01 – 1.13, p = <.05, power = .05) symptom count models, but not the PE symptom count model or neurodevelopmental burden model (PE symptom count: OR = 0.86, 95% CI 0.22 – 3.33, p = .83, power = 1.00; neurodevelopmental burden model: OR = 1.67, 95% CI 0.73 – 3.83, p = .23, power = .94).

The interaction effect was non-significant in each model (ADHD model: OR = 0.56, 95% CI 0.08 – 3.91, p = .56; ADHD symptom count model: OR = 0.87, 95% CI 0.75 – 1.02, p = .09; indicative ASD model: OR = 0.73, 95% CI 0.22 – 2.36, p = .59; indicative ASD symptom count model: OR = 0.99, 95% CI 0.93 – 1.05, p = .64; PE: OR = 4.80, 95% CI 0.90 – 25.66, p = .07; PE symptom count model: OR = 3.13, 95% CI 0.73 – 13.38, p = .12; neurodevelopmental burden model: OR = 1.10, 95% CI 0.45 – 2.66, p = .83).

Children with a ND CNV still remained more likely to experience bullying in each of the models (ADHD model: OR = 3.51, 95% CI 1.93 – 6.38, p = <.001, power = .38; ADHD symptom count model: OR = 3.41, 95% CI 1.61 – 7.21, p = <.05, power = .10; indicative ASD model: OR = 4.32, 95% CI 2.15 – 8.69, p = <.001, power = .61; indicative ASD symptom count model: OR = 3.80, 95% CI 1.53 – 9.49, p = <.05, power = .10; PE model: OR = 3.80, 95% CI 1.88 – 7.67, p = <.001, power = .29; PE

symptom count model: OR = 4.02, 95% CI 1.96 – 8.25, p = <.001, power = .35; neurodevelopmental burden model: OR = 3.09, 95% CI 1.26 –7.60, p = <.05, power = .06).

Age also remained positively associated with bullying in each of the models (ADHD model:  OR = 1.18, 95% CI 1.09 – 1.28, p = <.001; ADHD symptom count model: OR = 1.17, 95% CI 1.08 – 1.28, p = <.001; indicative ASD model: age: OR = 1.19, 95% CI 1.08 – 1.30, p = <.001; Indicative symptom count model: OR = 1.20, 95% CI 1.09 – 1.32, p = <.001; PE model: OR = 1.18, 95% CI 1.07 – 1.31, p = <.05; PE symptom count model: OR = 1.20, 95% CI 1.08 – 1.34, p = <.001, neurodevelopmental burden model: OR = 1.23, 95% CI 1.08 – 1.39, p = <.05).

Sex no longer significantly predicted bullying in the PE model nor PE symptom count models.

The only change that occurred when conducting the sensitivity analysis was that indicative ASD symptom count became non-significant (OR = 1.04, 95% CI 0.99 – 1.10, p = .15).

*Table 2.9 Results of GLMMs with added interaction effect*

| | | Bullied | | | | | | |
|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Bullied | Not bullied | OR | 95% conf.int | SE | z-score | p-value |
| A) | 587 | | | | | | | |
| Child has a ND CNV*__Meets criteria for ADHD__ | | | | 0.56 | 0.08 - 3.91 | 0.99 | -0.58 | 0.56 |
| Meets criteria for ADHD | | | | 3.85 | 0.58 - 25.55 | 0.97 | 1.40 | 0.16 |
| Child has a ND CNV | | | | 3.51 | 1.93 - 6.38 | 0.31 | 4.12 | **<.001** |
| Age | | | | 1.18 | 1.09 - 1.28 | 0.04 | 3.91 | **<.001** |
| Female | | | | 0.72 | 0.48 - 1.07 | 0.21 | -1.61 | 0.11 |
| Mother has a university degree | | | | 0.96 | 0.62 - 1.49 | 0.22 | -0.18 | 0.86 |
| | | | | | | | | |
| B) | 460 | | | | | | | |
| Child has a ND CNV*__ADHD symptom count__ | | | | 0.87 | 0.75 - 1.02 | 0.08 | -1.72 | 0.09 |
| ADHD symptom count | | | | 1.23 | 1.06 - 1.42 | 0.08 | 2.73 | **<.05** |
| Child has a ND CNV | | | | 3.41 | 1.61 - 7.21 | 0.38 | 3.21 | **<.05** |
| Age | | | | 1.17 | 1.08 - 1.28 | 0.05 | 3.57 | **<.001** |
| Female | | | | 0.88 | 0.58 - 1.35 | 0.22 | -0.57 | 0.57 |
| Mother has a university degree | | | | 1.17 | 0.72 - 1.91 | 0.25 | 0.62 | 0.54 |
| | | | | | | | | |
| C) | 587 | | | | | | | |
| Child has a ND CNV*__Meets criteria for indicative ASD__ | | | | 0.73 | 0.22 - 2.36 | 0.60 | -0.53 | 0.59 |
| Meets criteria for indicative ASD | | | | 2.65 | 0.90 - 7.85 | 0.55 | 1.76 | 0.08 |
| Child has a ND CNV | | | | 4.32 | 2.15 - 8.69 | 0.36 | 4.10 | **<.001** |
| Age | | | | 1.19 | 1.08 - 1.30 | 0.05 | 3.71 | **<.001** |
| Female | | | | 0.81 | 0.54 -1.23 | 0.21 | -0.98 | 0.33 |
| Mother has a university degree | | | | 0.97 | 0.61 - 1.56 | 0.24 | -0.11 | 0.91 |

*Table 2.9 continued*

| | | Bullied | | | | | | |
|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Bullied | Not bullied | OR | 95% conf.int | SE | z-score | p-value |
| D) | 589 | | | | | | | |
| Child has a ND CNV*__Indicative ASD symptom count__ | | | | 0.99 | 0.93 - 1.05 | 0.03 | -0.46 | 0.64 |
| Indicative ASD symptom count | | | | 1.07 | 1.01 - 1.13 | 0.03 | 2.36 | **<.05** |
| Child has a ND CNV | | | | 3.80 | 1.53 - 9.49 | 0.47 | 2.87 | **<.05** |
| Age | | | | 1.20 | 1.09 - 1.32 | 0.05 | 3.78 | **<.001** |
| Female | | | | 0.84 | 0.55 - 1.29 | 0.22 | -0.79 | 0.43 |
| Mother has a university degree | | | | 0.97 | 0.60 - 1.57 | 0.25 | -0.12 | 0.91 |
| | | | | | | | | |
| E) | 498 | | | | | | | |
| Child has a ND CNV*__Meets criteria for PE__ | | | | 4.80 | 0.90 - 25.66 | 0.86 | 1.83 | 0.07 |
| Meets criteria for PE | | | | 0.96 | 0.22 - 4.32 | 0.77 | -0.05 | 0.96 |
| Child has a ND CNV | | | | 3.80 | 1.88 - 7.67 | 0.36 | 3.72 | **<.001** |
| Age | | | | 1.18 | 1.07 - 1.31 | 0.05 | 3.22 | **<.05** |
| Female | | | | 0.61 | 0.38 - 1.00 | 0.25 | -1.97 | 0.05 |
| Mother has a university degree | | | | 1.04 | 0.61 - 1.75 | 0.27 | 0.13 | 0.89 |
| | | | | | | | | |
| F) | 486 | | | | | | | |
| Child has a ND CNV*__PE symptom count__ | | | | 3.13 | 0.73 - 13.38 | 0.74 | 1.54 | 0.12 |
| PE symptom count | | | | 0.86 | 0.22 - 3.33 | 0.69 | -0.21 | 0.83 |
| Child has a ND CNV | | | | 4.02 | 1.96 - 8.25 | 0.37 | 3.79 | **<.001** |
| Age | | | | 1.20 | 1.08 - 1.34 | 0.05 | 3.41 | **<.001** |
| Female | | | | 0.60 | 0.37 - 0.99 | 0.25 | -2.00 | 0.05 |
| Mother has a university degree | | | | 1.03 | 0.60 - 1.75 | 0.27 | 0.10 | 0.92 |

*Table 2.9 continued*

| | | Bullied | | | | | | |
|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Bullied | Not bullied | OR | 95% conf.int | SE | z-score | p-value |
| H) | 426 | | | | | | | |
| Child has a ND CNV***Neurodevelopmental burden** | | | | 1.10 | 0.45 - 2.66 | 0.45 | 0.21 | 0.83 |
| Neurodevelopmental burden | | | | 1.67 | 0.73 - 3.83 | 0.42 | 1.21 | 0.23 |
| Child has a ND CNV | | | | 3.09 | 1.26 - 7.60 | 0.46 | 2.46 | **<.05** |
| Age | | | | 1.23 | 1.08 - 1.39 | 0.06 | 3.26 | **<.05** |
| Female | | | | 0.68 | 0.40 - 1.19 | 0.28 | -1.35 | 0.18 |
| Mother has a university degree | | | | 1.42 | 0.76 - 2.65 | 0.32 | 1.11 | 0.27 |

Multiple Testing Correction

All results presented in this chapter survived BH-FDR correction.

## 2.6 Discussion

As the largest investigation into the bullying experiences of children with ND CNVs, this study helps to address our limited knowledge of the environmental experiences of children with these conditions, namely peer bullying at school. It is also the only study to explore the link between bullying experiences and individual and contextual risk factors, including neurodevelopmental profile, inheritance status and school placement.

Furthermore, the bullying experiences of this group had never previously been studied collectively (i.e., the study was not confined to the experiences of children with a specific ND CNV, for example, 22q11 DS). This approach helped to increase the sample size of this study, which is large compared to many studies researching ND CNVs. A collective approach was supported by members of the Cerebra Steering Group who noted similarities regarding day-to-day experiences and children's difficulties in spite of the specific ND CNV carried by the child. This observation that has been supported by Chawner et al. (2019). Although high rates of victimisation were seen in all ND CNVs (**Table 2.7**), this study did not explore the extent to which individual ND CNVs contributed to bullying risk. Therefore, although this study suggests children with ND CNVs are collectively at higher risk, it is still important to recognise the difference in experiences between children with different ND CNVs and explore these differences in future.

### 2.6.1 Findings and implications

As predicted, according to parents, a higher proportion of children with a ND CNV were picked on or bullied by their peers compared to sibling controls, indicating this group could be at high-risk for bullying. A strength of this study is that the experiences of both participant groups were measured in the same way, providing confidence that this finding is not the result of methodological differences in measurement.

The percentage of children who had a ND CNV who were reported as bullied was much higher than the national average reported in schools across England and Wales (52% vs 17%) (DfE, 2018), however this difference *could* be due to disparate methodologies. The DfE's results are based on children between the ages of 10-15,

on children's reports of their own experiences and on their experiences over the past 12 months, including cyber-bullying. Cyberbullying was not explicitly measured in this study. Furthermore, parents were the sole informants of bullying, but parent and child reported bullying experiences in children do not always agree (Shakoor et al., 2011). Therefore, rates of bullying within children with a ND CNV could actually be higher than those reported here if the DfE's measure was used.

The percentage of siblings reported as victimised was also higher than the national average (27% vs 17% respectively). Whilst the noted methodological differences between these studies should be kept in mind, findings could suggest that siblings of children with a ND CNV could represent a high-risk group for victimisation compared to the general population. Siblings may come to their brother or sister's defence if they are aware they are being bullied and may in turn become a target themselves (Huitsing et al., 2014). Alternatively, they could be at greater risk simply by association. The literature on the bullying experiences of siblings of children with SEN in general is limited and warrants future investigation.

The presence of ADHD, indicative ASD and PE were associated with increased odds for being bullied, corroborating previous findings that children with these neurodevelopmental conditions and traits are at increased risk for bullying (Cappadocia et al., 2012; Catone et al., 2015; Wolke et al., 2000). Furthermore, children who scored for an increased number of ADHD, indicative ASD and PE symptoms were also at greater odds for being bullied, indicating that children do not have to present with symptoms of the required severity or number for a research diagnosis, to be bullied by peers. The more neurodevelopmental conditions children met criteria for, the more likely they were to be victimised which could suggest a dose-response type relationship between neurodevelopmental challenges and bullying. These findings were replicated when the unaffected sibling group were also considered, minimising the risk that results are subject to collider bias.

IQ and ID did not predict bullying experience. This finding was out of keeping with other studies (Zeedyk et al., 2014), and could be the result of methodological differences. For example, Zeedyk et al. (2014) classified children with an IQ in the borderline range (71-84 IQ points) as having ID in their study. Furthermore, the WASI was used in the current study, whereas Zeedyk et al. (2014) measured IQ

using the Wechsler Intelligence Scale for Children (WISC-IV) (Wechsler, 2003). Individuals tend to score a few points higher when IQ is measured using the WASI compared to the WISC-IV, however scores are still comparable when using both FSIQ-4 and FSIQ-2 scores (Wolraich et al., 2008). Additionally, children with an ND CNV who were not coded as having ID in the current study may still have met criteria for one of the other neurodevelopmental conditions, increasing their odds for bullying despite not meeting criteria for ID.

In the GLMMs (without the added interaction effect), all the previous neurodevelopmental factors found to predict bullying remained significant; but the presence of a ND CNV also consistently predicted bullying experience, indicating that whilst the presence of neurodevelopmental conditions and traits may be important risk factors for bullying in children with ND CNVs, they do not explain the full picture. Additional traits commonly observed in children with ND CNVs are likely to also contribute to their risk of being targeted by peers.

When the interaction effect between a given neurodevelopmental risk factor and participant group was added into the GLMMs, only ADHD symptom count and indicative ASD symptom count remained significant predictors of bullying experience. The interaction effect itself was not significant in any of the models, however the presence of a ND CNV consistently heightened children's odds for being bullied. This finding strengthens the theory that children with a ND CNV are not at increased risk for bullying over and above other children with neurodevelopmental challenges for reasons explained by the neurodevelopmental challenge itself, but for other reasons related to different traits commonly seen in children with ND CNVs, for example facial dysmorphology. Indeed, physical appearance is reported to be the most common reason for young people to be bullied (UNESCO, 2019). It is important to better understand which additional factors impact bullying risk to improve the inclusivity of anti-bullying measures for this group. Interventions which target neurodevelopmental challenges are important, but additional challenges should also be addressed.

The literature on the relationship between age and victimisation is mixed (DfE, 2018; UNESCO, 2019). The results of this study indicate that as children with a ND CNV get older, their odds for being bullied increase. The developmental differences

between children with ND CNVs and their peers could become more notable as they get older and make them more vulnerable to being picked on. There is also some evidence of cognitive deterioration in children with ND CNVs (Vorstman et al., 2015). The current study did not explore experiences of children at different ages (i.e., chronic bullying), however this could be an important area of focus for future studies given that adverse outcomes associated with chronic bullying are particularly marked (Wolke et al., 2013). Investigating the impacts of chronic bullying would require longitudinal analysis.

Other demographic factors did not seem to impact bullying experiences. Similarly, neither inheritance status or school placement seemed to have an effect. We might expect there to be little predictive value in these factors given that approximately half of children with an ND CNV in this study were reported as bullied. Instead, bullying in this group seems to be common across the board.

Studying bullying experiences in children with ND CNVs can not only inform schools' approach to intervention, but it could also help to elucidate environmental contributions to the variation we see in the outcomes of this group, which so far have been relatively understudied. This study suggests that bullying in children with ND CNVs is influenced by neurodevelopmental profile, as well as other factors which remain unexplored. Thus, children with these risk factors could be particularly at risk for the poorer health and socioeconomic outcomes known to be associated with childhood and adolescent bullying experiences (Copeland et al., 2013; Takizawa et al., 2014, 2015). Parents and teachers should be informed about these at-risk children to increase their vigilance and ability to detect the signs of potential bullying so that they are better enabled to step in and support children in need.

The value of studying bullying in children with ND CNVs could go beyond the needs of this group. Exploring the experiences of children with ND CNVs who often have a broad range of difficulties in different functional domains could help us to deduce how different risk factors interplay with each other in the same individual, and which of these factors may increase risk over and above others. This would provide important insights for anti-bullying efforts at school, making them more impactful and beneficial to all children.

### 2.6.2 Limitations

Although this study addresses a gap in the literature and has the additional strengths outlined above, there are limitations. Firstly, I utilised one of the items within the SDQ to measure bullying experience. The SDQ does not provide a definition of bullying and therefore parents will have answered the question with their own understanding of what it means to be 'picked on or bullied'. This includes whether parents considered bullying at home, online or in other areas, or whether they only considered the school environment when responding. Similarly, I could not be confident as to whether relational and direct methods of bullying were both considered. All self-report measures suffer from participant subjectivity to some degree, however with no definition provided in this study to guide reporting, this could be particularly problematic here. As mentioned earlier, I had intended analysing responses to the SEQ, a questionnaire I created for the purposes of this chapter which provided a definition of bullying, to better understand bullying experiences in this group. The Rare Genetics Research Group at Cardiff University continue to collect data using the SEQ, which can be used to investigate whether the results of the current study are replicated when a clear definition of bullying is provided.

By using the SDQ, I opted for a single item measure of bullying experience. There are advantages in using single-item measures, for example, parsimony and ease for participants (Allen et al., 2022). Furthermore, Wanous (1997) argued that single items may be the preferred option when they are measuring unambiguous concepts. Bullying as measured by the SDQ is arguably an unambiguous concept, however, there remain issues with single-item measures, such as questionable reliability of results and limited ability to investigate phenomena in depth. Again, continued data collection with the SEQ could help to address this issue.

A multi-informant approach is considered the most reliable way to measure bullying in children (Shakoor et al., 2011), however, parents were the only informants used in this study. Parent and child reports of bullying only tend to modestly agree, however parents are credited as viable informants when children's reports are not available (Shakoor et al., 2011). Furthermore, good levels of agreement (.62) between children with intellectual and developmental disabilities and their parents have been

reported when completing the peer problem scale of their respective versions of the SDQ (Kaiser & Halvorsen, 2022). This does not negate the need to directly hear the experiences of children. Child self-reported bullying experiences are currently being collected by the team at Cardiff via the SEQ and will facilitate future investigation into the potential disparities between experiences reported by children and their parents.

The variables I was able to explore within my analyses were constrained by the data available. Therefore, although I considered several individual and contextual factors when assessing children's risk for bullying, many more that I was not able to consider have been identified as increasing children's likelihood of being bullied, for example, low self-esteem (O'Moore & Kirkham, 2001). Importantly, the association between children's ethnicity and bullying was not investigated here. The earlier ECHO and IMAGINE-1 studies only collected ethnicity data for children with a ND CNV and their biological parents. Sibling ethnicity was not recorded. As I could not know with certainty that children within the same family had the same biological parents, I could not assume sibling ethnicity from the data available. However, ethnicity as a risk factor for bullying could be explored in the future with the data collected by the more recent IMAGINE-2 and DiGEN studies, which have collected sibling ethnicity data.

Of the factors I did explore, my analyses do not indicate which of them likely contribute the most risk to children. Future research should explore these issues as results would be valuable to inform prevention strategies.

Analyses were cross-sectional meaning definitive conclusions about causal relationships between bullying and the associated variables cannot be concluded. The study's power ranged from .05 – 1.00. Therefore, power was too low in some of the analyses presented in this chapter to confidently presume findings reflect true relationships between variables (or lack of relationships).

The control group are not representative of the general population, however, there are strengths in utilising biologically related individuals as a control sample, specifically the increased ability to control for shared genetics and environment. Sibling controls have been cited as a preferable comparison group for children with a

ND CNV compared to general population samples for this reason, as well as being an easily accessible control sample (Arnold et al., 2001).

Children with a ND CNV were recruited from NHS genetic clinics, and therefore the ND CNV sample likely suffers from ascertainment bias. The reasons for referral to genetic testing are many and include intellectual disability, developmental delay, as well as psychiatric, behavioural, cardiovascular and neurological problems (Chawner et al., 2021). Therefore, to have been referred for genetic testing, children will have likely shown an impairment in these domains noticeable enough to have been flagged as warranting further investigation. Thus, children within this study could be more severely affected than those who comprise community samples, highlighting the need for population-based studies. Nevertheless, it is still important to understand the difficulties experienced by children with known genetic diagnoses to inform support and interventions for them and their families.

## 2.7 Conclusion

This chapter aimed to clarify whether children with a ND CNV are at risk for peer bullying compared to their unaffected siblings and elucidate some of the risk factors associated with bullying experiences in this group.

Having a ND CNV was positively associated with parent reported bullying experiences. This finding can be partly explained by children's neurodevelopmental profile and age, but additional individual and contextual factors beyond the scope of this study almost certainly contribute.

Siblings of children with a ND CNV may be at increased risk of victimisation compared to the general population, however this claim needs to be further investigated, along with the bullying experiences of siblings of children with a SEN in general.

These findings contribute to our understanding about the environmental influences on this group's outcomes. Given that environmental factors can to some extent be modified, and that bullying in childhood and adolescence can have far reaching and persistent impacts on individuals, this study presents an important area for intervention that could make a significant impact on children's outcomes.

This chapter has focused on one element of school experience, namely bullying, which falls under the broad category of social experiences at school. In the next chapter, I will shift focus to a more academic sphere of school experience, specifically, the extent to which children's support needs are met at school to help them access their school's curriculum.

# 3 Access to, and parent satisfaction with, support received at school for children with neurodevelopmental copy number variants

## 3.1 Chapter overview

Education is known to have a significant influence on a person's life outcomes. Support at school for children with SEN helps them to access education and experience its associated benefits, but recent investigation into the UK SEN provision system indicates that there are significant failings with its ability to provide for all children. Anecdotal accounts from parents of children with ND CNVs taking part in studies conducted by the Rare Genetic Research Group at Cardiff University, and members of the Cerebra Steering Group support this conclusion. Their testimonies also suggest parents of children with a ND CNV may face additional challenges when trying to secure educational support for their children compared to parents of children with SEN without a rare genetic cause. However, there does not appear to be any published evidence regarding the factors impacting access to, or parental satisfaction with such support in this group.

The aim of this chapter is to investigate factors associated with access to, and parental satisfaction with the educational support received by children with ND CNVs. Both quantitative and qualitative methods were used. Within this chapter, I define SEN and provide national statistics which characterise pupils in receipt of SEN support in England. I outline some of the issues with the current provision system as highlighted by a recent review into its operation, discuss some of the reasons behind said issues, and highlight the children who could be most vulnerable to having unmet need.

Nearly 25% of parents of children with an ND CNV reported they were unsatisfied with the support received by their child at school. Children with ADHD, ODD or CD were more likely to have unsatisfied parents, whereas parents of children who had an EHCP were more likely to be satisfied. In turn, children who met criteria for indicative ASD and ID, and those who had a *de novo* ND CNV were more likely to have an EHCP. Qualitative thematic analysis of information provided by parents in response to an open-ended question within the parent version of the SEQ provided further insight into the circumstances in which parents were unhappy with their

child's support at school. Issues impacting parent satisfaction included unhelpful learning environments, barriers to accessing higher level of support, limited understanding of children's needs, limited resources, as well as inadequacy regarding the amount and type of support received.

The results of this chapter indicate that children with a ND CNV are not always supported at school appropriately according to their parents. Steps should be taken to address inadequacies to ensure all children with ND CNVs have the best chance of reaching their full potential at school and in later life. Findings also provide tentative evidence that inequalities in educational provisions could impact the phenotypic variation observed amongst children with ND CNVs and thus support the notion that investigation into environmental experiences, and specifically school experiences, is important to better understand the variable presentation of this group.

## 3.2 Introduction

In the UK, children with SEN have a learning difficulty or disability which impairs their ability to access their school's curriculum (DfE, DoH, 2015). They are entitled to additional support at school to 'help them achieve the best possible education' and to prepare them for adulthood. Maintained schools (i.e., those which are publicly funded) receive extra monetary provision from their LA to provide such support, however children whose needs exceed what can be provided despite this additional funding should be in receipt of further support as outlined by an EHCP. EHCPs are statutory documents that detail the additional provision required by a child across education, health and social care which should be enabled by the child's school and the LA (DfE, DoH, 2015). Children with SEN made up 17.9% of pupils in English maintained primary, secondary and special schools in 2022-2023 (ONS, 2023). Of those, 24.6% had an EHCP (4.2% of pupils overall). ASD was the most common type of need for those with an EHCP whereas speech, language and communication needs was most common for children with SEN but without a plan. Boys made up 72.4% of pupils who had an EHCP.

In 2014, the UK government passed the Children and Families Act (2014) which made substantive changes to the SEN provision system, aiming to increase families'

involvement in decision making processes regarding children's support, to better integrate statutory services, to extend SEN support up to 25 years of age and to ultimately improve the outcomes of children with SEN. Prior to the revisions, children with an ECHP would have been in receipt of what was then called an 'educational statement'. The 'legal test' to secure a statement and an EHCP did not change (DfE, DoH, 2015) meaning those considered eligible for a statement and a plan should be the same. Therefore, children who were historically statemented should in principle meet criteria for an EHCP if they were being assessed for such need today.

The aims of the Children and Families Act sought to address issues within the previous SEN system, however significant failures in realising these aims were identified during recent reviews into SEN provision (DfE, DHSC, 2022; HCEC, 2019). It was concluded that the current system is one of 'unmet need and strain' (HCEC, 2019). Indeed, 81.3% of LAs in England spent over budget in 2017-2018 (NAO, 2019). Overspend was attributed to insufficient funding, as well as increases in the number of children attending special schools. The latest figures from the ONS (2023) reported a 5% increase in the number of pupils enrolled in a specialist placement relative to the previous year, and it is estimated to cost LAs £2,500 more to fund a child attending a special school, compared to a child with an EHCP in a mainstream school (NAO, 2019). Rises in the number of children in specialist placements are thought to reflect the increase in input parents now have regarding their child's support and could indicate that parents are not satisfied with the support mainstream schools can offer.

Widespread overspending is indicative of financial strain across regions, however some demographic groups could be particularly vulnerable to having unmet need. The DfE's NAO (2019) reported substantial variation between LAs regarding the proportion of children identified with SEN and who had an EHCP, sometimes referred to as the 'postcode lottery'. At the time the report was published, there had been no specific investigation into the reasons why this variation had been observed, however the DfE proposed it reflected 'local context and practice'. Additionally, the HCEC (2019) stated that parents' social capital has a 'huge' impact in whether their child receives further educational support. Parents and teachers perceive the SEN system to be rigged towards 'wealthy' and 'well-educated' parents, who are assumed

to be better informed of their rights, more confident to take on the system and better able to access additional aid to secure educational support (e.g., from charities). With this in mind, we might expect children from higher socioeconomic groups to be more likely to receive SEN, yet this is not observed. Children eligible for free school meals are in fact more often reported to have SEN (ONS, 2023). This might suggest that such assumptions by parents and teachers are incorrect, or reflect data which report that psychological and neurodevelopmental conditions are more common in children with lower socioeconomic status (SES) (Reiss, 2013). It has been proposed that the recorded number of children who have SEN could be an underrepresentation of the true number (Shaw, 2017), and that children from lower socioeconomic backgrounds are at particular risk of falling under the radar. Therefore, although a higher proportion of children with lower SES make up the number of children with SEN in the UK, they are also likely to make up a higher percentage of children with SEN who have not been picked up by services and remain unsupported.

Further indication of failure within the system and cause for concern is the reported exclusion rate of children with SEN from school compared to their peers. Children with SEN are more often excluded in all nations of the UK (ONS, 2023; Welsh Government, 2022; Scottish Government, 2022; DfE, NI, 2023) and again, certain groups of children who have SEN are at greater risk compared to others. The Timpson Review (2019) stated that children with ECHPs are less likely to be excluded compared to those with general SEN provision, indicating that EHCPs offer some kind of protection from exclusion. This observation could in part be explained by government guidance (DfE, 2022) which requires an emergency review of a child's EHCP to be undertaken prior to their potential exclusion to ensure the needed provisions are being provided (and implemented if they are not), and thus adds an intermediatory step which could better protect children with an EHCP from exclusion. Equally, children with an EHCP might display less behaviour which prompts exclusion because their needs are potentially better supported or because their disability is so much more severe that their opportunity to transgress is more limited. The Timpson Review also indicated that a child's reported primary need impacted their likelihood for exclusion. Children with social, emotional and mental health (SEMH) needs who did not have an EHCP were more likely to be excluded

compared to those with additional needs arising from a different cause, for example a physical disability. Children with SEMH needs experience difficulties which may make them ‘withdrawn or isolated’, or ‘display challenging, disruptive, or disturbing behaviour’ (DfE, DoH, 2015). These behaviours could be indicative of underlying psychiatric conditions (e.g., anxiety, depression, eating disorders, ADHD, attachment disorder) or ‘physical symptoms which are medically unexplained’. Interestingly, children with SEMH needs with an EHCP were less likely to be excluded compared to other children with SEN without an EHCP, as well as pupils without SEN, further highlighting the protective value of EHCPs. Additional research supports the notion that children with SEMH needs are an at-risk group for exclusion. A systematic review conducted by Whear et al. (2014) indicated that children with ADHD, severe depression and problems in behaviour, peer relationships and prosocial skills are more often excluded, suspended or expelled from mainstream school compared to their peers. Parker et al. (2019) also found evidence that children with ADHD and CD are at greater risk of exclusion.

In a recent Ofsted report (2019), teachers were noted to be lacking in support from external agencies as to how to manage issues beyond their expertise, including SEN. Ofsted concluded the lack of support for educators restricts the subsequent support they can provide children. Lack of support for teachers also contributes to the poor mental wellbeing observed within the profession (Ofsted, 2019). The mental health of teaching staff is notably worse than the mental health of the general population (Health and Safety Executive, 2022), indicating that increased support for educators could benefit both them and pupils with SEN. Furthermore, the psychological wellbeing of teachers is associated with that of their pupils (Harding et al., 2018). Therefore, strategies implemented to better support children should not be at the detriment of school staff or vice versa, or the success of these strategies could be limited.

The information presented paints a bleak picture of SEN provision in the UK, suggesting that even when children have their needs identified, they are not always supported adequately. This begs the question, how well are children with ND CNVs being supported at school? Are some children better supported than others and what factors might contribute to differences in provision? Whilst mindful not to make

sweeping statements based on a few parents' personal accounts, anecdotal testimonies from members of the Cerebra Steering Group and parents taking part in the research studies conducted by the Rare Genetic Research Group at Cardiff University indicate that parents of children with ND CNVs are unsatisfied with the support their child's school provides them and that they can experience great difficulty obtaining it. Challenges commonly cited include lack of funding for schools and LAs to implement support, as well as challenges more specific to this group, such as limited understanding about needs associated with children's genetic conditions. This suggests that whilst parents may face barriers which are commonly experienced by families of children with SEN, they may experience additional challenges specific to having a child with a rare genetic form of SEN when trying to secure educational support.

To improve access to effective support, it is important to determine whether children with an ND CNV who need support are in fact satisfactorily supported at school, and which children are most vulnerable to being missed by services.

Parental satisfaction literature pertaining to general ID populations typically report on satisfaction with a range of school related factors, or education in general. For example, Perry et al. (2020) investigated factors related to 'school satisfaction' in parents of children with severe and multiple developmental disabilities in Canada. Parents of children with lower adaptive skills and higher maladaptive skills were less satisfied, as well as parents of children in regular class settings (i.e. not in a special class placement). Higher parent score on a measure of positivity was the strongest predictor of parent satisfaction.

Studies that have explicitly investigated satisfaction with SEND support are seemingly few in the UK. Research conducted in America may provide some insight, although results may not be completely applicable across nations. Investigating parental satisfaction with elements of their child's 'Individualized Education Programme' (IEP), a legal document similar to EHCPs in the UK, Slade et al. (2018) reported that 61% of parents of children with ASD were dissatisfied with at least one element of the IEP (out of a total of four investigated: 1) IEP content, 2) support provided, 3) perceived level of agreement between IEP content and support provided

and 4) effectiveness of the IEP team). Forty-two percent were dissatisfied with 3 or all elements.

Male's (1998) findings may contribute our understanding of parental satisfaction with SEND provision in the UK, however, results are very likely too outdated to apply to current circumstances. Overall, parents of children with severe or profound and multiple learning difficulties were satisfied with their child's special school placement, although 1 in 5 reported they would prefer their child to learn within a special class within a mainstream school. Parents also indicated that their level of involvement in their child's education was not as high as they would ideally like. A more recent UK based study compared parental satisfaction with educational provision between parents of children with DS, WS and ASD (Van Herwegen et al., 2018). Satisfaction was lowest for parents of children with ASD. This group were also the least likely to receive one to one support and have parents report that their school met their child's SEND needs. However, parents within each group reported they were concerned about the level of knowledge professionals had about their child's condition and how to support them. There was also no difference between the groups regarding satisfaction with the level of educational progress parents felt their child had made within the past year.

Research conducted by Parsons et al. (2009) investigated parental satisfaction for in the UK regarding children with SEN in general versus focusing on children with a particular type of SEN. They reported that parents of children with SEN and disabilities in England, Scotland and Wales rated the support received by their child from their school as positive, overall. Satisfaction with the support provided was 'the norm rather than the exception'. Parents of children who had a then 'educational statement' (51.1%) were more satisfied compared to those who did not. Notably, 12% of parents reported they did not know if their child had a statement. Parents were also more satisfied if their child attended a special school compared to a mainstream school. Parents from higher socioeconomic backgrounds were more likely to report they were aware of government legislation pertaining to disability rights, which was in turn associated with higher reported levels of satisfaction. Children who were identified as having a 'psychological difficulty' had the least satisfied parents, compared to those who were identified with 'language and ASD needs', 'sensory needs', or 'motor needs'. Children with motor conditions were the

group whose parents were most satisfied, although information was not available as to the specific support they received, nor how this compared to what support other children had.

The findings of Parsons et al. (2009) indicate that whilst there are pockets of children who are vulnerable to not having their educational needs met, the majority of children with SEN and disabilities were satisfactorily supported according to their parents. Nevertheless, as the authors state, such findings are not an excuse for 'political complacency', and effort should be made to ensure that all children receive quality education. It is also worth noting that since this study was conducted, real terms funding for public services has decreased. Funding for schools specifically is predicted to be 3% below 2010 levels by 2024-2025 (Sibieta, 2022). As funding deficits have contributed to a strained SEN provision system, described as failing to meet need (HCEC, 2019), the conclusions of Parsons et al. (2009) may not be applicable to families' experiences today.

This study aims to explore the level of satisfaction reported by parents of children with a ND CNV and whether findings align with the picture of SEN support painted by parents' anecdotal accounts described above, and the conclusions of the SEN review and Parsons et al. (2009). I sought to employ both quantitative and qualitative methods to investigate the reasons why some children may not receive a satisfactory level of support. I used data collected via the SEQ (Chapter 2) by the Rare Genetic Research Group. Given the importance of education for a person's immediate and long-term socioeconomic and health outcomes (Raghupathi & Raghupathi, 2020), the extent to which children are supported at school could in part explain the marked phenotypic heterogeneity observed between individuals with ND CNVs. Better understanding regarding which children are vulnerable to insufficient support and why this might be, could inform the development and enacting of national and local policy to make support provision more equitable.

## 3.3 Aims

The aims of this chapter were to investigate the following questions:

**Aim 1: Are children with a ND CNV more likely to have an EHCP compared to their unaffected control siblings?**

Given children with ND CNVs have been found to be at higher risk for physical, developmental and psychiatric conditions (Chawner et al., 2021), I expected that a higher proportion of children with a ND CNV would have an EHCP.

**Aim 2: What are the predictors associated with having an EHCP in children with a ND CNV?**

Predictors:

*a) Child and family demographic factors*

Nationally, boys are more likely to have an EHCP compared to girls (ONS, 2023) and so I predicted that this trend would be reflected within my sample. If parent and teacher assumptions are correct (HCEC, 2019) and the system favours families with higher SES, I also expected children with higher SES to be more likely to have an EHCP.

*b) Child neurodevelopmental and psychological factors*

As the most common primary reported need of children with an EHCP is ASD (ONS, 2023), I predicted children who displayed more ASD traits to be more likely to have an EHCP. Given the reported difficulties in obtaining SEN support, I did not expect all children who met criteria for a given neurodevelopmental or psychological condition to have an EHCP. Furthermore, not all children with one of these conditions may need an EHCP. Conversely, I expected children with SEMH related needs, ADHD or behavioural conditions as less likely to have an EHCP.

*c) ND CNV inheritance status: inherited or* de novo*?*

No previous study of which I am aware has investigated the link between inheritance status and support from public services, however previous findings indicate that children with inherited ND CNVs present with more SEMH related problems (Cunningham et al., 2022; Wolstencroft et al., 2022). Children with SEMH are seemingly at risk of being under supported (The Timpson Review, 2019; Whear et al., 2014; Parker et al., 2019). Children with an inherited ND CNV are also more likely to be from deprived socioeconomic backgrounds (Wolstencroft et al., 2022),

another assumed risk factor for unmet need (HCEC, 2019). Therefore, I predicted children with an inherited ND CNV as less likely to have an EHCP.

**Aim 3: Are parents of children who have an ND CNV satisfied with their child's educational support?**

Given the recently noted failures of the SEN support system (DfE, DHSC, 2022; HCEC, 2019) and personal accounts heard from parents, I predicted that parents would report that they were unsatisfied with the support provided by their child's school. This hypothesis is out of keeping with the findings of Parsons et al. (2009), however schools are reportedly in a worse economic position compared to when their study was conducted (Sibieta, 2022), which could impact current parent satisfaction.

**Aim 4: What are the individual and contextual predictors of parent satisfaction with educational support received by children with a ND CNV?**

Individual factors:

*a) Child and family demographic factors*

Assuming the belief is correct that wealthier and more highly educated parents are better able to obtain an EHCP for their child (HCEC, 2019) and given that having an EHCP is associated with higher parent satisfaction (Parsons et al., 2009) and lower exclusion rates (The Timpson Review, 2019), I expected parents of children from higher socioeconomic backgrounds to be more satisfied with their child's educational support.

*b) Child neurodevelopmental and psychological factors*

I predicted SEMH needs or behavioural conditions in children to be negatively associated with parent satisfaction. This was based on the findings of Parsons et al. (2009) and reports of higher exclusion rates in these children (Whear et al., 2014).

*c) ND CNV inheritance status: inherited or* de novo*?*

For reasons explained above, I predicted children with an inherited ND CNV to be less likely to have an EHCP. As EHCP status has been associated with parent satisfaction (Parsons et al., 2009) and school exclusion (The Timpson Review, 2019), I predicted parents of children with an inherited ND CNV to be more likely unsatisfied with their child's support.

*d) EHCP status: child has an EHCP: yes or no?*

I expected parents of children with an EHCP would be more likely satisfied with their child's educational support compared to parents of children who did not have an EHCP.

Contextual factors:

*d) School placement: mainstream or special?*

Again, considering the findings of Parsons et al. (2009), I also expected that children attending special school would more likely have satisfied parents.

**Aim 5: In what circumstances are the educational needs of children with ND CNVs not supported?**

I expected parents to report that their child's educational needs were not supported due to challenges commonly experienced by parents of children with SEN in general (e.g., funding), as well as challenges more specific to parents of children with ND CNVs, for example limited awareness of rare genetic conditions. This hypothesis was based on parents' anecdotal accounts.

## 3.4 Methods

### 3.4.1 Sample

Children with an ND CNV and unaffected siblings who took part in the ECHO, IMAGINE-1, IMAGINE-2 and DiGEN studies (see Chapter 2) made up the study sample.

Data from 413 children overall were used in this analysis. The presence of a ND CNV was confirmed via genotyping of biological samples provided by participants. Genotyping was undertaken at Cardiff University's DPMCN. For participants who did not have sample genotyped, the presence of the ND CNV was established via clinical genetics reports. If the presence of an ND CNV could not be confirmed for a child, they were not included in the analysis. **Table 3.1** provides a breakdown of the ND CNV group by ND CNV type.

*Table 3.1 Breakdown of ND CNV group by ND CNV type*

| ND CNV | *n* | % |
|---|---|---|
| 1q21.1 duplication | 11 | 4 |
| 1q21.1 deletion | 14 | 5 |
| 15q11.2 deletion | 32 | 11 |
| 15q13.3 duplication | 9 | 3 |
| 15q13.3 deletion | 14 | 5 |
| 16p11.2 duplication | 24 | 9 |
| 16p11.2 deletion | 48 | 17 |
| 22q11.2 duplication | 29 | 10 |
| 22q11.2 deletion | 69 | 25 |
| NRXN1 | 16 | 6 |
| TAR duplication | 8 | 3 |
| More than one priority ND CNV | 6 | 2 |
| **Total** | **280** | **100** |

### 3.4.2 Measures

The information collected by each of the studies listed above is mostly similar, however additional assessments and questions have been administered to participants over time to address new and developing research aims. Consequently, whilst some of the data analysed for the purposes of this chapter were available for all participants, other data were only available for participants who took part in the more recent IMAGINE-2 and DiGEN studies. Thus, the size of the participant sample varied between some of the aims explored. The participants who had data available for each of the measures utilised for the purposes of this chapter will be highlighted below. Unless otherwise specified, each of the studies collected information from all participants using the following measures.

### Demographics

Demographic information for all the children taking part was collected from a primary caregiver, and included the child's age and biological sex, their mother's educational qualifications and approximate family income. The latter two variables were used as a marker of socioeconomic status. When known, the inheritance status of all participants with an ND CNV was recorded (i.e., inherited or *de novo*). The type of school children attended (mainstream or special) was also recorded.

### EHCP status

Prior to the 2014 Children and Families Act, an EHCP was known as a 'Statement of Educational Need'. To reflect this change in legislation and terminology, there are slight differences between studies regarding the questions posed to parents to determine whether their child was entitled to educational support to the level outlined by an EHCP. **Table 3.2** presents the questions parents were asked in each of the studies. If parents answered yes to all the questions they had data recorded for, their child was coded positively for having an EHCP in this study. If there were discrepancies between the answers provided to these questions, the child was coded as 'unknown' and not included in the analyses. This was because I could not be as confident that the parent truly knew whether their child had such support.

*Table 3.2 Questions asked in the ECHO, IMAGINE-1, IMAGINE-2 and DiGEN studies to determine EHCP status*

| Study | Year study was established | Question/s used to determine EHCP status |
|---|---|---|
| ECHO | 2010 | Has your child been educationally statemented? - Self-report questionnaire<br>Have they [child] received an educational statement? - Interview with researcher |
| IMAGINE-1 | 2014 | Has your child been educationally statemented? - Self-report questionnaire<br>Have they [child] received an educational statement? - Interview with researcher |
| DiGEN | 2019 | Has your child been educationally statemented or have they got an Education, health and care (EHC) plan? - Self-report questionnaire<br>Does your child have a Statement of Educational Need? - Self-report questionnaire<br>Have they [child] received an educational statement? - Interview with researcher |
| IMAGINE-2 | 2020 | Has your child been educationally statemented or have they got an Education, health and care (EHC) plan? - Self-report questionnaire<br>Does your child have a Statement of Educational Need? - Self-report questionnaire<br>Have they [child] received an educational statement? - Interview with researcher |

### School Experiences Questionnaire

The SEQ was briefly introduced in Chapter 2. I designed a separate parent and child version of the SEQ in 2019 to explore the aims of this thesis. Therefore, it was only administered to families who took part in DiGEN and IMAGINE-2. The SEQ explores children's bullying experiences (see Chapter 2), as well as their general experiences at school, for example whether they enjoy school and if they get along with their teachers.

Unless parents needed assistance from the research team with completing self-report questionnaires, they completed the SEQ themselves. Within the SEQ parents were asked "Has your child's school provided the right support for your child?" and could answer either 'yes' or 'no'. This question was included to measure the parents' satisfaction with their child's educational support. Parents who responded 'yes' were coded as 'satisfied' with their child's educational support. Those who responded 'no' were coded as 'unsatisfied'. Unsatisfied parents were then asked to "please briefly

explain why your child's school has not provided your child with the right support" and provided a free text response.

### Child and Adolescent Psychiatric Assessment

Parent responses to the Parent CAPA (introduced in Chapter 2) determined the presence of ADHD, ODD, CD, generalised anxiety disorder (GAD) and depression. If children met criteria for any of these conditions, they were coded positively for the associated condition. If they did not meet criteria, they were coded negatively.

Few participants met criteria for CD or depression. Thus, I merged the CD variable with ODD to make an ODD/CD variable. Children were coded positively if they met criteria for either condition. I did the same for GAD and depression to make an 'emotional problems' (EP) variable.

### Social Communication Questionnaire

Parent responses to the SCQ (introduced in Chapter 2) were used to derive a measure of indicative ASD. Children who scored above the threshold for indicative ASD (≥15 points) were coded 'yes' for meeting criteria. Those who did not meet criteria were coded 'no'. For children who had missing data for <10% of the SCQ items (N = 30), I pro-rated their missing items with their overall SCQ mean performance score, which was calculated using the items they had completed information for. I then calculated their total score and coded appropriately.

### Wechsler Abbreviated Scale of Intelligence

Children's IQ scores were calculated based on their performance on the WASI (introduced in Chapter 2). The WASI gives a measure of FSIQ which is based on children's performance on four subtests (two VIQ and two PIQ subtests: FSIQ-4) or two subtests (one VIQ and one PIQ subtest: FSIQ-2). 'Very small' differences have been noted between FSIQ-4 and FSIQ-2 performance scores (Wolraich et al., 2008). Children with an FSIQ score <70 were coded positively for ID. Those with a score of ≥70 were coded as not having ID.

Of children who had IQ data available, 24.7% (N = 102/413) could only complete one of the PSIQ subtests. Thus, their FSIQ score represented their FSIQ-2 score. The FSIQ score of the remaining 75.3% of children represented their FSIQ-4 score.

Developmental Coordination Disorder Questionnaire (DCDQ)

The DCDQ (Wilson et al., 2009) was used to screen for the presence of developmental coordination disorder (DCD) in children. Parents complete the DCDQ on behalf of their child and performance scores are calculated. Three sub-test scores measuring control of movement, and general and fine coordination skills are combined to compute an overall performance score. Total score performance thresholds are dependent on age (**Table 3.3**), and scores range from 15-75. Lower scores are indicative of poorer coordination. Performance on the DCDQ does not provide a definitive diagnosis of DCD but is an indicative marker. Children would need to undergo further assessment for a clinical diagnosis to be determined (Wilson et al., 2009).

*Table 3.3 DCDQ overall performance thresholds relative to participant age*

| Participant age | DCDQ score which is indicative of DCD |
|---|---|
| 5 years – 7 years, 11 months | 15-46 |
| 8 years – 9 years, 11 months | 15-55 |
| 10 years – 15 years, 11 months | 15-57 |

'Any neurodevelopmental or psychological condition'

I created an 'any condition' variable which described whether a child met criteria for any of the neurodevelopmental or psychological conditions (ADHD, indicative ASD, ID, indicative DCD, ODD/CD plus EP). Children who met criteria for at least one condition were coded 'yes' and those who did not meet criteria for any condition were coded 'no'.

### 3.4.5 Analysis

Data were first loaded into SPSS version 27 for data cleaning, and then imported into RStudio for statistical analysis. ND CNV group and control group characteristics were statistically compared using chi-square analysis for categorical variables. Age was the only continuous variable. Shapiro-Wilk test revealed that age was not

normally distributed for either the control or ND CNV group, and so Mann-Whitney U test was used to calculate between group differences in age.

**Aim 1: Are children with a ND CNV more likely to have an EHCP compared to their unaffected control siblings?**

Chi-square analysis was performed to determine between group differences in EHCP status. Logistic regression was then performed to control for the following demographic covariates: child age and sex, and mother's education level. ECHP status was entered into the model as the dependent variable and participant group as the independent. Mother's education status and family income data were missing for 17.9% (N = 74) and 19.6% (N = 81) of participants respectively. Therefore, I conducted two sensitivity analyses whereby I first added mother's education into the model as an additional covariate, and then family income.

To ensure the assumptions of logistic regression were met, I checked for the presence of multicollinearity between the independent variables within each of the models by using variance inflation factor (VIF). VIF values above 5 indicate that a problematic correlation between two or more variables could exist within the model (Menard, 2001). VIF for all the predictors within each of the models described above did not surpass 5. Logistic regression also assumes a linear relationship between each continuous variable within the model and the logit of the dependent variable. Age was the only continuous variable included in these analyses and did not violate the linearity assumption within any of the models.

**Aim 2: What are the predictors associated with having an EHCP in children with a ND CNV?**

To investigate the impact of neurodevelopmental and psychological profile on EHCP status, a separate logistic regression was performed for each of the associated variables respectively. The given neurodevelopmental/psychological variable (ADHD, indicative ASD, ID, indicative DCD, ODD/CD, EP or 'any condition') was entered into the model as the independent variable and child's age and sex, and mother's education were entered as covariates. EHCP status was entered as the dependent variable. I was not able include participant group as an additional covariate because the subsample of control participants who had an EHCP was too small to provide

findings which were not skewed once the neurodevelopmental/psychological variables were also added into their respective model. Each regression was followed up with sensitivity analysis into which family income was added as an additional covariate.

Next, I added inheritance status as an additional covariate. Inheritance status was not added to the first set of regressions as only 64.9% of the ND CNV group had available data. Therefore, adding inheritance status into the model at a later stage allowed investigation into EHCP status in a larger sample size first. Sensitivity analyses were then performed.

None of the assumptions were violated for any of these regression models.

After running the regressions into which inheritance status was included, I ran chi-square analyses to compare the inherited and de novo group regarding each neurodevelopmental/psychological variable, mother's education, and family income to gain more insight into any differences observed between inheritance status groups.

**Aim 3: Are parents of children who have an ND CNV satisfied with their child's educational support?**

I compared the overall frequency of children whose parents reported they were satisfied with the support provided by their child's school to the frequency of those whose parents reported they were unsatisfied.

**Aim 4: What are the individual and contextual predictors of parent satisfaction with educational support received by children with a ND CNV?**

The first set of logistic regressions as explained above were rerun but with parent satisfaction status added into the model as the dependent variable. First a given neurodevelopmental/psychological variable was added into a respective model, with child's age and sex, and mother's education added into the model. The same sensitivity analyses were then performed.

Logistic regression could not be performed for indicative DCD or 'any condition' as sub sample sizes were insufficient (there were 0 participants who did not meet

criteria for ‘any condition’ or for DCD whose parents were unsatisfied with their educational support). Therefore, Fisher’s exact tests were performed to test for between group differences in ‘any condition’ and indicative DCD between children whose parents were satisfied and those whose parents were unsatisfied.

Inheritance status was then added into the model as an additional covariate, and the relevant sensitivity analyses were performed.

Next, to determine the association between EHCP and parent satisfaction, inheritance status was substituted for EHCP status. Again, the follow up sensitivity analyses were run.

Lastly, to establish whether there was an association between ‘type of school’ attended and parent satisfaction, EHCP status was substituted for ‘type of school’ within the logistic regression. Sensitivity analyses were performed.

Additional regressions which included the sibling sample could not be performed because parent satisfaction data were not collected for the siblings.

None of the assumptions for logistic regressions were violated by any of the models above.

Post-hoc Power Analysis

I used the ‘pwr’ package in R to perform post-hoc power analyses for quantitative findings relevant to **Aims 1, 2 and 4**. To detect associations between the independent and dependent variables in each regression model, I used the lower bound of the 95% confidence interval in my calculations to avoid ‘winner’s curse’ and assumed a significance level of 0.05 throughout.

Multiple Testing Correction

To correct for multiple testing, I applied the BH-FDR method to each statistical analysis, assuming an alpha of 0.05.

**Aim 4: In what circumstances are the educational needs of children with ND CNVs not supported?**

I conducted thematic analysis (TA) (Braun & Clarke, 2006) to analyse the free text data provided by participants in response to the SEQ item: “Please briefly explain why your child’s school has not provided your child with the right support.”

Braun and Clarke’s method of TA (2006), termed ‘reflexive’ TA (RTA) (Braun & Clarke, 2019), is a well-established method of qualitative data analysis. RTA is a method which incorporates truly qualitative values (Braun & Clarke, 2021), as explained in greater detail in Chapter 4. That is, RTA does not emphasise the need for inter-rater reliability, objectivity, large sample sizes or generalisability of findings, which are qualities often associated with quantitative studies. Instead, RTA recognises and values the subjectivity of participant experience, as well as the influence the researcher’s own subjectivity and life experience will inevitably have on findings, which should be actively reflected on throughout data collection and analysis (Clarke & Braun, 2013). Thus, the use of two coders is not necessary, or a marker of reliable findings. Researchers analyse their data inductively, whereby they approach the dataset with their research aims in mind, but without prior identified ‘codes’ (defined below) for which they are searching for.

Five stages to RTA have been outlined (Braun & Clarke, 2006, 2019), followed by a sixth ‘write up’ stage. I describe the process I followed when analysing the free text responses in relation to the first 5 stages, below.

1) Familiarisation

First, I collected all the responses and read through each of them in turn to ‘immerse’ myself in the data (i.e., to get an overall impression of the data). Familiarisation is an ‘active’ process and so I noted the initial thoughts and impressions I had when reading the responses to encourage proper engagement with the data. I also considered and noted how my subjectivity might influence the way I interpreted the data (see below).

2) Generating initial codes

Next, I read through the data again and noted the reasons stated as to why children had not received the right support at school according to their parents. These defined my initial codes, which were recorded in an Excel spreadsheet. Braun and Clarke (2006) define codes as ‘semantic or latent’ features identified within the data

which are of interest to the researcher. I then reread the question responses to ensure all the data had been categorised and coded under the correct code/s.

3) Generating initial themes

Themes are ‘patterns of shared meaning underpinned or united by a core concept’ (Braun & Clarke, 2019). To help identify my initial themes, I created a table within Excel and grouped codes which were thematically similar into the same column. Each column was given a heading which thematically tied the codes within it together. These headings defined my initial themes.

4) Reviewing themes & 5) Defining and naming themes

To ensure my themes were sound, I reviewed whether each of them properly described the codes within them and made the appropriate corrections if they did not (e.g., switched codes into a more applicable theme, renamed the theme, merged two themes together). I then read through the entire dataset once more to ensure that my themes encapsulated their content fully. Themes were settled once I was happy that all relevant content within parents’ responses were accurately reflected.

Reflexivity

As noted above, Braun and Clarke (2013) stipulate that researchers should actively reflect on how their subjective experiences, ideas and opinions may influence the way in which they conduct their data collection and analysis throughout the process, and the implications of this. Parents provided the qualitative data analysed here via self-report questionnaire. Thus, there was no real opportunity for my subjective position to influence what parents shared. However, free text answers do not provide as great an opportunity as for example an interview or focus group for participants to share their experiences, and so parents may have felt restrained as to what they could share when providing their answers.

In terms of analysing the data, as a 29 year old female who does not have children, or any close personal relationships with a child with SEN or parent of a child with SEN, I did not consider myself to have any personal experiences which were obviously relatable to those which parents were sharing and would cause me to potentially approach the data from a highly biased standpoint in terms of prior individual experience. However, professionally, I have had many conversations with

parents of children with ND CNVs or other neurodevelopmental conditions and have often heard about their struggles in accessing support from all public services, not just education. Furthermore, I have not heard about access to support from the point of view of professionals on the other side (i.e., the ones who are approached by parents for support). Therefore, I recognised that I may interpret the data, not only with my prior knowledge about the barriers parents can face when trying to access support in mind, but that my impressions of such a process could be biased towards parents' position and less inclusive of professionals' experiences.

## 3.5 Results

### Sample characteristics

**Table 3.4** presents between group differences in sample characteristics between the ND CNV and control groups for the overall sample. The age range of participants was 6-16.

*Table 3.4 Overall sample characteristics*

| | Total Sample | | | ND CNV Group | | | Control Group | | | Statistics | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N (413) | Mean (SD) | N (%) | N (280) | Mean (SD) | N (%) | N (133) | Mean (SD) | N (%) | z | $X^2$ *(df)* | p-value |
| Child's age | 413 | 10.21 (2.82) | | 280 | 10.13 (2.81) | | 133 | 10.38 (2.86) | | -0.819 | | 0.41 |
| Sex: | 413 | | | 280 | | | 133 | | | | | |
| Male | 237 | | 57.4% | 165 | | 58.9% | 72 | | 54.1% | | .847 *(1)* | 0.36 |
| Female | 176 | | 42.6% | 115 | | 41.1% | 61 | | 45.9 | | | |
| Mother's education: | 339 | | | 266 | | | 73 | | | | | |
| University degree | 119 | | 35.1% | 90 | | 33.8% | 29 | | 39.7% | | .873 *(1)* | 0.35 |
| No university degree | 220 | | 64.9% | 176 | | 66.2% | 44 | | 60.3% | | | |
| Family income: | 332 | | | 263 | | | 69 | | | | | |
| ≤£19,999 | 100 | | 30.1% | 90 | | 34.2% | 10 | | 14.5% | | 10.12 *(1)* | **<.05** |
| ≥£19,999 | 232 | | 69.9% | 173 | | 65.8% | 59 | | 85.5% | | | |
| Inherited status: | 182 | | | 182 | | | - | | - | | | |
| De novo | 72 | | 39.6% | 72 | | 39.6% | - | | - | | | |
| Inherited | 110 | | 60.4% | 110 | | 60.4% | - | | - | | | |
| | | | | | | | | | | | | |
| Psychiatric characteristics: | | | | | | | | | | | | |
| ADHD | 391 | | | 269 | | | 122 | | | | | |
| Yes | 118 | | 30.2% | 107 | | 39.8% | 11 | | 9.0% | | 37.69 *(1)* | **<.001** |
| No | 273 | | 69.8% | 162 | | 60.2% | 111 | | 91.0% | | | |
| Indicative ASD | 391 | | | 271 | | | 120 | | | | | |
| Yes | 148 | | 37.9% | 137 | | 50.6% | 11 | | 9.2% | | 60.56 *(1)* | **<.001** |
| No | 243 | | 62.1% | 134 | | 49.4% | 109 | | 90.8% | | | |
| ID | 361 | | | 240 | | | 121 | | | | | |
| Yes | 64 | | 17.7% | 62 | | 25.8% | 2 | | 1.7% | | 32.25 *(1)* | **<.001** |
| No | 297 | | 82.3% | 178 | | 74.2% | 119 | | 98.3% | | | |

*Table 3.4 continued*

| | Total Sample | | | ND CNV Group | | | Control Group | | | Statistics | | |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| | N (413) | Mean (SD) | N (%) | N (280) | Mean (SD) | N (%) | N (133) | Mean (SD) | N (%) | z | $X^2$ *(df)* | p-value |
| Indicative DCD | 351 | | | 227 | | | 124 | | | | | |
| Yes | 225 | | 64.1% | 202 | | 89.0% | 23 | | 18.5% | | 172.91 *(1)* | **<.001** |
| No | 126 | | 35.9% | 25 | | 11.0% | 101 | | 81.5% | | | |
| ODD/CD | 396 | | | 266 | | | 130 | | | | | |
| Yes | 54 | | 13.6% | 48 | | 18.0% | 6 | | 4.6% | | 13.37 *(1)* | **<.001** |
| No | 342 | | 86.4% | 218 | | 82.0% | 124 | | 95.4% | | | |
| Emotional problems | 360 | | | 268 | | | 92 | | | | | |
| Yes | 42 | | 11.7% | 36 | | 13.4% | 6 | | 6.5% | | 3.17 *(1)* | 0.08 |
| No | 318 | | 88.3% | 232 | | 86.6% | 86 | | 93.5% | | | |
| Meets criteria for at least one condition | 354 | | | 262 | | | 92 | | | | | |
| Yes | 296 | | 83.6% | 254 | | 96.9% | 42 | | 45.7% | | 130.77 *(1)* | **<.001** |
| No | 58 | | 16.4% | 8 | | 8.0% | 50 | | 54.3% | | | |
| EHCP status: | 408 | | | 275 | | | 133 | | | | | |
| EHCP | 190 | | 46.6% | 181 | | 65.8% | 9 | | 6.8% | | 125.63 *(1)* | **<.001** |
| No EHCP | 218 | | 53.4% | 94 | | 34.2% | 124 | | 93.2% | | | |
| | | | | | | | | | | | | |
| Type of school: | 340 | | | 216 | | | 124 | | | | | |
| Mainstream | 287 | | 84.4% | 165 | | 76.4% | 122 | | 98.4% | | 28.97 *(1)* | **<.001** |
| Special | 53 | | 15.6% | 51 | | 23.6% | 2 | | 1.6% | | | |
| | | | | | | | | | | | | |
| Parent satisfaction: | 108 | | | 108 | | | - | | - | | | |
| Satisfied | 83 | | 76.9% | 83 | | 76.9% | - | | - | | | |
| Unsatisfied | 25 | | 23.1% | 25 | | 23.1% | - | | - | | | |

**Aim 1: Are children with a ND CNV more likely to have an EHCP compared to their unaffected control siblings?**

A larger proportion of children with an ND CNV were reported to have an EHCP compared to children in the unaffected sibling group (65.8% vs 6.8% respectively; $X^2$ = 125.63, p = <.001). Logistic regression showed that children with a ND CNV were more likely to have an EHCP after controlling for child's age and sex (OR: 27.35, 95% CI 13.94 – 60.41, p = <.05, power = 1.00). Results remained the same in follow up sensitivity analyses whereby mother's education and family income were added into the regression model respectively.

**Aim 2: What are the predictors associated with having an EHCP in children with a ND CNV?**

**Table 3.5** presents the characteristics of the ND CNV group only, broken down by whether they had an EHCP and between group differences between those with and without an EHCP.

*Table 3.5 Characteristics of the ND CNV group with EHCP data available*

| | N | | | Mean (SD) | | Statistics | | |
|---|---|---|---|---|---|---|---|---|
| | **N (275)** | **EHCP** | **No EHCP** | **EHCP** | **No EHCP** | **z** | **$X^2$ *(df)*** | **p-value** |
| *Demographics:* | | | | | | | | |
| Mean age in years (SD) | 275 | 181 (65.8%) | 94 (34.2%) | 10.21 (2.82) | 9.95 (2.80) | -0.77 | | 0.44 |
| Sex | 275 | | | | | | 0.58 *(1)* | 0.45 |
| Male | 164 (59.6%) | 105 (64.0%) | 59 (36.0%) | | | | | |
| Female | 111 (40.4%) | 76 (68.5%) | 35 (31.5%) | | | | | |
| Mother's highest level of qualification | 260 | | | | | | 2.86 *(1)* | 0.09 |
| University degree | 88 (33.8%) | 64 (72.7%) | 24 (27.3%) | | | | | |
| No university degree | 172 (66.2%) | 107 (62.2%) | 65 (37.8%) | | | | | |
| Approximate family income | 258 | | | | | | 0.14 *(1)* | 0.71 |
| ≤£19,999 | 88 (34.1%) | 60 (68.2%) | 28 (31.8%) | | | | | |
| >£19,999 | 170 (65.9%) | 112 (65.9%) | 58 (34.1%) | | | | | |
| Inheritance status | 177 | | | | | | 9.41 *(1)* | **<.05** |
| De novo | 70 (39.5%) | 55 (78.6%) | 15 (21.4%) | | | | | |
| Inherited | 107 (60.5%) | 60 (56.1%) | 47 (43.9%) | | | | | |
| | | | | | | | | |
| *Psychiatric characteristics:* | | | | | | | | |
| ADHD | 265 | | | | | | 0.65 *(1)* | 0.42 |
| Yes | 105 (39.6%) | 72 (68.6%) | 33 (31.4%) | | | | | |
| No | 160 (60.4%) | 102 (64.0%) | 58 (36.0%) | | | | | |
| Indicative ASD | 266 | | | | | | 5.22 *(1)* | **<.05** |
| Yes | 134 (50.4%) | 97 (72.4%) | 37 (27.6%) | | | | | |
| No | 132 (49.6%) | 78 (59.1%) | 54 (40.9%) | | | | | |
| ID | 236 | | | | | | 18.46 *(1)* | **<.001** |
| Yes | 61 (25.8%) | 52 (85.2%) | 9 (14.8%) | | | | | |
| No | 175 (74.2%) | 95 (54.3%) | 80 (45.7%) | | | | | |

*Table 3.5 continued*

| | N | | | Mean (SD) | | Statistics | | |
|---|---|---|---|---|---|---|---|---|
| | **N (275)** | **EHCP** | **No EHCP** | **EHCP** | **No EHCP** | **z** | $\chi^2$ ***(df)*** | **p-value** |
| Indicative DCD | 222 | | | | | | 1.48 *(1)* | 0.22 |
| Yes | 198 (89.2%) | 132 (66.7%) | 66 (33.3%) | | | | | |
| No | 24 (9.8%) | 13 (54.2%) | 11 (45.8%) | | | | | |
| ODD/CD | 262 | | | | | | 0.88 *(1)* | 0.77 |
| Yes | 48 (18.3%) | 31 (64.6%) | 17 (35.4%) | | | | | |
| No | 214 (81.7%) | 143 (68.8%) | 71 (33.2%) | | | | | |
| Emotional problems | 264 | | | | | | 0.17 *(1)* | 0.68 |
| Yes | 35 (13.3%) | 22 (62.9%) | 13 (37.1%) | | | | | |
| No | 229 (86.7%) | 152 (66.4%) | 77 (33.6%) | | | | | |
| Any psychiatric condition | 257 | | | | | | | |
| Yes | 246 (96.9%) | 167 (67.1%) | 82 (32.9%) | | | | | **0.02*** |
| No | 8 (1.2%) | 2 (25.0%) | 6 (75.0%) | | | | | |
| | | | | | | | | |
| Type of school | 211 | | | | | | | |
| Mainstream | 160 (75.8%) | 94 (58.8%) | 66 (41.3%) | | | | 27.55 *(1)* | **<.001** |
| Special | 51 (24.2%) | 50 (98.0%) | 1 (2.0%) | | | | | |

**Fisher's exact test used to test for between group differences.*

**Table 3.6** presents the results of multivariate logistic regressions which explore the association between EHCP status and neurodevelopmental/psychological presentation whilst controlling for demographic factors.

Indicative ASD, ID and 'any condition' were the only significant predictors of whether children had an EHCP (indicative ASD: OR = 1.86, 95% CI 1.09 – 3.21, p = <.05, power = .06; ID: OR = 6.18, 95% CI 2.76 – 15.85, p = <.001, power = .88; any condition: OR = 5.78, 95% CI 1.28 – 40.40, p = <.05, power = .06). ADHD, indicative DCD, ODD/CD and EP did not significantly predict EHCP status (ADHD: OR = 1.30, 95% CI 0.75 – 2.26, p = .35, power = .19; indicative DCD: OR = 1.72, 95% CI 0.71 - 4.11, p = .22, power = .12; ODD/CD: OR = .83, 95% CI 0.42 – 1.65, p = .58, power = .74; EP: OR = .92, 95% CI 0.43 - 2.04, p = .84, power = .74).

Sensitivity analyses whereby family income was added as an additional covariate into each of the models did not reveal any change in findings.

*Table 3.6 Logistic regression results: EHCP status and neurodevelopmental/psychological presentation whilst controlling for demographic factors*

| | EHCP Status | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | EHCP | No EHCP | OR | 95% conf.int | SE | z-score | p-value | $R^2$ |
| A) | 250 | 164 | 86 | | | | | | |
| **Meets criteria for ADHD** | | | | 1.30 | 0.75 – 2.26 | 0.28 | 0.93 | 0.35 | 0.02 |
| Age | | | | 1.10 | 1.00 – 1.22 | 0.05 | 1.93 | 0.05 | |
| Female | | | | 1.03 | 0.60 – 1.77 | 0.28 | 0.10 | 0.92 | |
| Mother has a university degree | | | | 1.70 | 0.96 – 3.08 | 0.30 | 1.79 | 0.07 | |
| | | | | | | | | | |
| B) | 252 | 166 | 86 | | | | | | |
| **Meets criteria for indicative ASD** | | | | 1.86 | 1.09 - 3.21 | 0.28 | 2.24 | **0.03** | 0.03 |
| Age | | | | 1.06 | 0.96 - 1.17 | 0.05 | 1.15 | 0.25 | |
| Female | | | | 1.22 | 0.71 - 2.11 | 0.28 | 0.71 | 0.48 | |
| Mother has a university degree | | | | 1.62 | 0.91 - 2.94 | 0.30 | 1.61 | 0.11 | |
| | | | | | | | | | |
| C) | 223 | 139 | 84 | | | | | | |
| **Meets criteria for ID** | | | | 6.18 | 2.76 – 15.85 | 0.44 | 4.14 | **<.001** | 0.09 |
| Age | | | | 1.05 | 0.94 – 1.17 | 0.06 | 0.83 | 0.41 | |
| Female | | | | 1.15 | 0.64 – 2.08 | 0.30 | 0.46 | 0.65 | |
| Mother has a university degree | | | | 1.62 | 0.87 – 3.07 | 0.32 | 1.50 | 0.13 | |
| | | | | | | | | | |
| D) | 213 | 138 | 75 | | | | | | |
| **Meets criteria for indicative DCD** | | | | 1.72 | 0.71 - 4.11 | 0.44 | 1.22 | 0.22 | 0.02 |
| Age | | | | 1.05 | 0.95 - 1.17 | 0.05 | 0.93 | 0.35 | |
| Female | | | | 1.10 | 0.61 - 1.99 | 0.30 | 0.32 | 0.75 | |
| Mother has a university degree | | | | 1.64 | 0.89 - 3.07 | 0.31 | 1.57 | 0.12 | |

*Table 3.6 continued*

| | EHCP Status | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | EHCP | No EHCP | OR | 95% conf.int | SE | z-score | p-value | $R^2$ |
| E) | 247 | 164 | 83 | | | | | | |
| **Meets criteria for ODD/CD** | | | | 0.83 | 0.42 – 1.65 | 0.35 | -0.56 | 0.58 | 0.02 |
| Age | | | | 1.10 | 1.00 – 1.22 | 0.05 | 1.88 | 0.06 | |
| Female | | | | 1.02 | 0.59 – 1.77 | 0.28 | 0.06 | 0.95 | |
| Mother has a university degree | | | | 1.66 | 0.93 – 3.02 | 0.30 | 1.70 | 0.09 | |
| | | | | | | | | | |
| F) | 249 | 164 | 85 | | | | | | |
| **Meets criteria for EP** | | | | 0.92 | 0.43 - 2.04 | 0.39 | -0.21 | 0.84 | 0.02 |
| Age | | | | 1.10 | 1.00 - 1.22 | 0.05 | 1.87 | 0.06 | |
| Female | | | | 1.07 | 0.62 - 1.85 | 0.28 | 0.24 | 0.81 | |
| Mother has a university degree | | | | 1.61 | 0.91 - 2.91 | 0.29 | 1.62 | 0.11 | |
| | | | | | | | | | |
| G) | | | | | | | | | |
| **Any condition** | 243 | 157 | 78 | 5.78 | 1.28 – 40.40 | 0.83 | 2.10 | **0.04** | 0.03 |
| Age | | | | 1.10 | 1.00 – 1.22 | 0.05 | 1.77 | 0.08 | |
| Female | | | | 1.17 | 0.67 – 2.04 | 0.28 | 0.54 | 0.59 | |
| Mother has a university degree | | | | 1.52 | 0.84 – 2.81 | 0.31 | 1.37 | 0.17 | |

When inheritance status was added into each of the models as an additional covariate, indicative ASD and ID remained the only significant predictors of having an EHCP (indicative ASD: OR = 2.12, 95% CI 1.08 – 4.27, p = <.05, power = .06; ID: OR = 5.88, 95% CI 2.04 – 21.47, p = <.05, power =.36).

Inheritance status was a significant predictor of having an EHCP in each of the models. Those with an inherited ND CNV were less likely to have an EHCP in each of the models (ADHD model: OR = 0.38, 95% CI 0.18 – 0.77, p = <.05; indicative ASD model: OR = 0.36, 95% CI 0.17 – 0.73, p = <.05; ID model: OR = 0.41, 95% CI 0.19 – 0.87, p = <.05; DCD model: OR = 0.31, 95% CI 0.13 – 0.67, p = <.05; ODD/CD model: OR = 0.42, 95% CI 0.20 – 0.85, p = <.05; EP model: OR = 0.40, 95% CI 0.19 – 0.80, p = <.05; any condition model: OR = 0.37, 95% CI 0.17 – 0.77, p = <.05). Findings were unchanged after conducting the sensitivity analyses.

**Table 3.7** shows the number and percentage of children within the ND CNV group who were included in the inheritance status regression models above (not the sensitivity analyses) who met criteria for each neurodevelopmental/psychological variable broken down by inheritance status. Information about mother's education and family income are also included.

*Table 3.7 Number and percentage of children within the ND CNV group broken down by inheritance status.*

| | De novo - N (%) | | Inherited - N (%) | | Statistics | | |
|---|---|---|---|---|---|---|---|
| Meets criteria for: | Yes | No | Yes | No | $X^2$ *(df)* | p-value | Power |
| ADHD | 26 (38.8%) | 41 (61.2%) | 45 (46.4%) | 52 (53.6%) | 0.93 (1) | 0.34 | 1.00 |
| Indicative ASD | 34 (50.7%) | 33 (49.3%) | 55 (55%) | 45 (45%) | 0.29 (1) | 0.59 | 0.96 |
| ID | 19 (32.2%) | 40 (67.8%) | 16 (17.4%) | 76 (82.6%) | 4.43 (1) | **0.04** | 1.00 |
| Indicative DCD | 51 (91.1%) | 5 (8.9%) | 77 (87.5%) | 11 (12.5%) | 0.44 (1) | 0.51 | 1.00 |
| ODD/CD | 10 (14.9%) | 57 (85.1%) | 23 (24.0%) | 73 (76.0%) | 1.99 (1) | 0.16 | 1.00 |
| Emotional problems | 9 (13.4%) | 58 (86.6%) | 13 (13.5%) | 83 (86.5%) | 0.00 (1) | 0.98 | 0.05 |
| 'Any condition' | 64 (100%) | 0 (0%) | 93 (94.9%) | 5 (5.1%) | 3.37 (1) | 0.07 | 1.00 |
| | | | | | | | |
| Mother has university degree | 31 (43.7%) | 40 (56.3%) | 27 (26.7%) | 74 (73.3%) | 5.35 (1) | **0.02** | 1.00 |
| Income = ≤£19,999 | 20 (29.0%) | 49 (71.0%) | 40 (40.4%) | 59 (59.6%) | 2.31 (1) | 0.13 | 1.00 |

## Aim 3: Are parents of children who have an ND CNV satisfied with their child's educational support?

**Table 3.8** presents the characteristics of children who had parent satisfaction data available (N = 108/280; 38.6%). Just over 75% (76.9%; N = 83/108) of parents were satisfied with the support their child's school had provided their child.

*Table 3.8 Characteristics of the ND CNV group with parent satisfaction data available*

| | N | | | Mean (SD) | | Statistics | | |
|---|---|---|---|---|---|---|---|---|
| | **Total (108)** | **Satisfied** | **Unsatisfied** | **Satisfied** | **Unsatisfied** | **z** | **$X^2$ *(df)*** | **p-value** |
| *Demographics:* | | | | | | | | |
| Mean age in years (SD) | 108 | 83 (76.9%) | 25 (23.1%) | 10.88 (2.73) | 11.16 (2.76) | -0.41 | | 0.68 |
| Sex | 108 | | | | | | 0.26 *(1)* | 0.61 |
| Male | 61 (56.5%) | 48 (78.7%) | 13 (21.3%) | | | | | |
| Female | 47 (43.5%) | 35 (74.5%) | 12 (25.5%) | | | | | |
| Mother's highest level of qualification | 106 | | | | | | 1.73 *(1)* | 0.19 |
| University degree | 60 (56.6%) | 43 (71.7%) | 17 (28.3%) | | | | | |
| No university degree | 46 (43.4%) | 38 (82.6%) | 8 (17.4%) | | | | | |
| Approximate family income | 107 | | | | | | 0.45 *(1)* | 0.50 |
| ≤£19,999 | 41 (38.3%) | 30 (73.2%) | 11 (26.8%) | | | | | |
| >£19,999 | 66 (61.7%) | 52 (78.8%) | 14 (21.2%) | | | | | |
| Inheritance status | 62 | | | | | | 0.08 *(1)* | 0.78 |
| De novo | 27 (43.5%) | 20 (74.1%) | 7 (25.9%) | | | | | |
| Inherited | 35 (56.5%) | 27 (77.1%) | 8 (22.9%) | | | | | |
| | | | | | | | | |
| *Psychiatric characteristics:* | | | | | | | | |
| ADHD | 104 | | | | | | 6.34 *(1)* | **<.05** |
| Yes | 42 (40.4%) | 27 (64.3%) | 15 (35.7%) | | | | | |
| No | 62 (59.6%) | 53 (85.5%) | 9 (14.5%) | | | | | |
| Indicative ASD | 105 | | | | | | 2.92 *(1)* | 0.09 |
| Yes | 63 (60.0%) | 45 (71.4%) | 18 (28.6%) | | | | | |
| No | 42 (40.0%) | 36 (85.7%) | 6 (14.3%) | | | | | |
| ID | 87 | | | | | | 0.00 *(1)* | 0.97 |
| Yes | 28 (32.2%) | 21 (75%) | 7 (25%) | | | | | |
| No | 59 (67.8%) | 44 (74.6%) | 15 (25.4%) | | | | | |

*Table 3.8 continued*

| | N | | | Mean (SD) | | Statistics | | |
|---|---|---|---|---|---|---|---|---|
| | **Total (108)** | **Satisfied** | **Unsatisfied** | **Satisfied** | **Unsatisfied** | **z** | **$X^2$ *(df)*** | **p-value** |
| Indicative DCD | 100 | | | | | | | 0.20* |
| Yes | 91 (91.0%) | 70 (76.9%) | 21 (23.1%) | | | | | |
| No | 9 (9.0%) | 9 (100%) | 0 (0%) | | | | | |
| ODD/CD | 104 | | | | | | | **<.05*** |
| Yes | 16 (15.4%) | 8 (50%) | 8 (50%) | | | | | |
| No | 88 (84.6%) | 72 (81.8%) | 16 (18.2%) | | | | | |
| Emotional problems | 104 | | | | | | | **<.05*** |
| Yes | 19 (18.3) | 11 (57.9%) | 8 (42.1%) | | | | | |
| No | 85 (81.7%) | 69 (81.2%) | 16 (18.8%) | | | | | |
| Any psychiatric condition | 104 | | | | | | | **1.00*** |
| Yes | 103 (99.0%) | 79 (76.7%) | 24 (23.3%) | | | | | |
| No | 1 (1.0%) | 1 (100%) | 0 (0.0%) | | | | | |
| | | | | | | | | |
| Type of school | 100 | | | | | | 2.68 *(1)* | 0.10 |
| Mainstream | 70 (70.0%) | 53 (75.7%) | 17 (24.3%) | | | | | |
| Special | 30 (30.0%) | 27 (90%) | 3 (10%) | | | | | |
| | | | | | | | | |
| EHCP | 103 | | | | | | 5.15 *(1)* | **<.05** |
| Yes | 78 (75.7%) | 64 (82.1%) | 14 (17.9%) | | | | | |
| No | 25 (24.3%) | 15 (60%) | 10 (40%) | | | | | |

**Fisher's exact test used to test for between group differences.*

**Aim 4: What are the individual and contextual predictors of parent satisfaction with educational support received by children with a ND CNV?**

**Table 3.9** presents the results of logistic regressions which explored the association between parent satisfaction and neurodevelopmental/psychological presentation whilst controlling for demographic factors. As logistic regression could not be performed for ‘any condition’ Fisher’s exact test was performed and revealed no significant difference in parent satisfaction between children who met criteria for ‘any condition’ and those who did not meet criteria for ‘any condition’ ($p = 1.00$) (**Table 3.8**).

*Table 3.9 Logistic regression results: Parent satisfaction and neurodevelopmental/psychological presentation whilst controlling for demographic factors*

| | | Parent Satisfaction | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Satisfied | Unsatisfied | OR | 95% conf.int | SE | z-score | p-value | $R^2$ |
| A) | 102 | 78 | 24 | | | | | | |
| **Meets criteria for ADHD** | | | | 0.31 | 0.11 - 0.82 | 0.51 | -2.31 | **0.02** | 0.07 |
| Age | | | | 0.94 | 0.78 - 1.13 | 0.09 | -0.66 | 0.51 | |
| Female | | | | 0.72 | 0.28 - 1.89 | 0.49 | -0.67 | 0.50 | |
| Mother has a university degree | | | | 1.67 | 0.63 - 4.66 | 0.51 | 1.01 | 0.31 | |
| | | | | | | | | | |
| B) | 103 | 79 | 24 | | | | | | |
| **Meets criteria for indicative ASD** | | | | 0.42 | 0.13 – 1.19 | 0.56 | -1.57 | 0.12 | 0.04 |
| Age | | | | 0.94 | 0.78 – 1.12 | 0.09 | -0.73 | 0.46 | |
| Female | | | | 0.76 | 0.29 – 1.99 | 0.48 | -0.56 | 0.57 | |
| Mother has a university degree | | | | 1.38 | 0.51 – 3.90 | 0.51 | 0.62 | 0.53 | |
| | | | | | | | | | |
| C) | 86 | 64 | 62 | | | | | | |
| **Meets criteria for ID** | | | | 1.08 | 0.37 - 3.38 | 0.56 | 0.15 | 0.88 | 0.02 |
| Age | | | | 0.98 | 0.81 - 1.18 | 0.10 | -0.25 | 0.81 | |
| Female | | | | 0.58 | 0.21 - 1.55 | 0.50 | -1.08 | 0.28 | |
| Mother has a university degree | | | | 1.40 | 0.51 - 4.00 | 0.52 | 0.64 | 0.52 | |
| | | | | | | | | | |
| | 102 | 78 | 24 | | | | | | |
| **Meets criteria for ODD/CD** | | | | 0.22 | 0.07 – 0.69 | 0.59 | -2.59 | **0.01** | 0.08 |
| Age | | | | 0.97 | 0.80 – 1.16 | 0.09 | -0.35 | 0.73 | |
| Female | | | | 0.63 | 0.24 – 1.67 | 0.49 | -0.92 | 0.36 | |
| Mother has a university degree | | | | 1.81 | 0.68 – 5.10 | 0.51 | 1.16 | 0.25 | |

*Table 3.9 continued*

| | Parent Satisfaction | | | | | | | | |
|---|---|---|---|---|---|---|---|---|---|
| Independent predictors | Total | Satisfied | Unsatisfied | OR | 95% conf.int | SE | z-score | p-value | $R^2$ |
| E) | 102 | 78 | 24 | | | | | | |
| **Meets criteria for EP** | | | | 0.32 | 0.11 - 0.98 | 0.56 | -2.04 | **0.04** | 0.06 |
| Age | | | | 1.01 | 0.84 - 1.21 | 0.09 | 0.06 | 0.96 | |
| Female | | | | 0.64 | 0.24 - 1.66 | 0.49 | -0.93 | 0.35 | |
| Mother has a university degree | | | | 1.81 | 0.69 - 5.04 | 0.5 | 1.18 | 0.24 | |

ADHD, ODD/CD and EP were the only significant predictors of parent satisfaction within their respective models. Children who met criteria for any of these conditions were less likely reported as receiving the right support at school (ADHD: OR = 0.31, 95% CI 0.11 - 0.82, p = <.05, power = 1.00; ODD/CD: OR = 0.22, 95% CI 0.07 - 0.69, p = <.05, power = .99; EP: OR = 0.32, 95% CI 0.11 - 0.98, p = <.05, power = .98). Indicative ASD and ID did not predict parental satisfaction (indicative ASD: OR = 0.42, 95% CI 0.13 – 1.19, p = .12, power = 1.00; ID: OR = 1.08, 95% CI 0.37 - 3.38, p = .88, power = .53). All results were unchanged in the sensitivity analyses when income was added into the model.

When inheritance status was added into the model as an additional covariate, ODD/CD was the only remaining significant predictor of parent satisfaction. Again, children who met criteria for ODD/CD were less likely reported as receiving the right support at school (ODD/CD: OR = 0.13, 95% CI 0.02 – 0.69, p = <.05, power = .98). Inheritance status was not associated with parent satisfaction in any of the models (ADHD model: OR = 1.29, 95% CI 0.37 – 4.62, p = .69; indicative ASD model: OR = 1.51, 95% CI 0.43 - 5.42, p = .52; ID model: OR = 1.71, 95% CI 0.46 – 6.68, p = 0.43; ODD/CD model: OR = 1.43, 95% CI 0.37 - 5.72, p = 0.60; EP model: OR = 1.39, 95% CI 0.38 – 5.13, p = 0.62). Sensitivity analyses did not result in any change to findings.

Next, inheritance status was substituted with EHCP status within each of the models. Children who had an EHCP were more likely to have satisfied parents within each of the models (ADHD model: OR = 4.99, 95% CI 1.53 – 17.71, p = <.05; indicative ASD model: OR = 4.19, 95% CI 1.36 – 13.62 p = <.05; ID model: OR = 3.36, 95% CI 1.07 – 10.72, p = <.05; ODD/CD model: OR = 3.54, 95% CI 1.15 – 11.13, p = <.05; EP model: OR = 3.40, 95% CI 1.13 – 10.33, p = <.05). ADHD and ODD/CD were the only significant neurodevelopmental/psychological predictors of satisfaction within their respective models (ADHD: OR = 0.20, 95% CI 0.06 – 0.58, p = <.05, power = 1.00; ODD/CD: OR = 0.21, 95% CI 0.06 – 0.69, p = <.05, power = .99). Adding income into each of the models when conducting the sensitivity analyses only resulted in one change. Indicative ASD became a significant predictor of parent satisfaction, with children meeting criteria less likely to have satisfied parents (OR = 0.29, 95% CI 0.08 – 0.94, p = <.05, power = 1.00).

Finally, EHCP status was substituted with school type status within each of the models. Children who met criteria for ADHD or ODD/CD were more likely to have unsatisfied parents (ADHD: OR = 0.27, 95% CI 0.08 – 0.81, p = <.05, power = 1.00; ODD/CD: OR = 0.11, 95% CI 0.01 – 0.59, p = <.05, power = 1.00). The type of school children attended predicted parent satisfaction in the ODD/CD model only, with parents more likely satisfied if their child attended a special school (OR = 7.00, 95% CI 1.45 – 60.03, p = <.05). Sensitivity analyses did not change the findings of any of the other models.

Multiple Testing Correction

All findings survived BH-FDR multiple testing correction.

**Aim 5: In what circumstances are the educational needs of children with ND CNVs not supported?**

Twenty-four out of a possible 25 participants (96%) provided an answer to the SEQ item: “Please briefly explain why your child’s school has not provided your child with the right support.”

The following themes were identified when analysing the responses: barriers to higher level support; unhelpful learning environment; amount and type of support; limited resources; and limited understanding. **Figure 3.1** shows the proportion of data each theme represents within the data. **Figure 3.2** is a visual diagram which presents how themes, and their respective codes are linked to one another.

a) Barriers to higher level support

Children’s needs were not supported in some cases because of difficulties in obtaining higher level support from their LA (local authority). Some of these challenges were experienced because of school related factors whereas others were beyond the control of the school. Barriers identified are presented in **Table 3.10**, as well as some verbatim quotes provided by parents.

Barriers to higher level support prevented the school from accessing the guidance and expertise of professionals who conduct needs assessments and consequently,

impacted schools' ability to understand a child's difficulties and implement the right support.

> *"Awaiting a statutory needs assessment to determine an accurate picture of his abilities and needs. Until now, school hasn't been sure of his needs due to no assessments." (Parent quote)*

Thus, this theme was linked to the theme 'limited understanding'.

*Table 3.10 Barriers to higher level support*

| **Barrier** | **School factor vs beyond school factor** | **Further explanation** |
|---|---|---|
| Child's diagnosis (or lack of) | Cannot be determined from the data provided | One parent highlighted the specific need to have an ASD diagnosis to access further support: *"We are currently trying to seek specialist provision but without an ASD diagnosis we are limited".* |
| No school backing | School | Several parents reported they did not have the support of their child's school to apply for further support. Parents' requests for help had been *"ignored"* and *"refused".* Lack of support from particular members of staff, including the SENCO [Special Education Needs Coordinator] and headteacher was noted, indicating their importance in securing additional support. |
| LA processes and procedures | Beyond school | One parent noted that factors associated with the ways in which LAs operate act as a barrier, even when they have the backing of the school: *"I recognise that school are doing all they can to get this support, but local authority processes and decisions are causing severe delays."* Such issues were exacerbated by COVID, and one parent noted she had sought a private educational psychologist assessment because of the added delays in obtaining publicly funded assessments over the pandemic. |

b) The amount and type of support

In many instances, parents felt their child was not supported at school correctly because they did not receive either the right type or right quantity of support.

The absence of an EHCP was noted by several parents, supporting the notion that parents deem EHCPs as beneficial to obtaining the right support.

> *"Fundamentally, no school is able to really provide the right support without an EHCP…" (Parent quote)*

However, for those who did have an EHCP, there were also instances where the support stipulated within the EHCP had not been implemented.

> *"In preparation for transition to secondary school, she was awarded 20 hours support through an EHCP. This support was never provided." (Parent quote)*

The specific types of support noted as missing or insufficient included academic, behavioural, psychological, one to one support and as noted above, support around transition to secondary school. One parent also reported that the type of support provided was reactive and not preventative after having been informed by the school's SENCO that *"they wait until a crisis happens and then they offer support".* In the parent's view, this was *"counterproductive"*.

c) Limited understanding

Parents noted that teachers sometimes failed to understand their child's needs. Examples provided included particular neurodevelopmental conditions and traits such as pathological demand avoidance, dyslexia and dyspraxia, as well as ADHD, but specifically in girls and those with an inattentive subtype. Lack of understanding about children's genetic diagnosis was also mentioned with one parent describing the school's lack of understanding about 22q11 DS. Evidence of diagnostic overshadowing also indicated limited understanding of children's genetic diagnosis.

*“Since we got the genetic diagnosis they [the school] use that as the reason for everything. They [the school] used to complain a lot, but since the diagnosis they have retracted a lot of what they said and now deny that he struggles.” (Parent quote)*

One parent noted *“being quiet and compliant doesn't mean support isn't required”* indicating that schools may not understand that children experiencing difficulties may present in different and subtle ways.

Parents also described a lack of knowledge about the support their child was receiving. This was attributed to poor communication from the school.

*“…they have him in different supports, but I don't feel I’m kept informed great” (Parent quote)*

*“She has interventions - but not enough and feedback is rather vague.” (Parent quote)*

This might indicate that children are better (or worse) supported than their parents are aware, and that better communication between school and home could impact parents’ satisfaction with educational support.

As noted above, children’s EHCPs were not always implemented. A lack of understanding by teachers as to what was included in a child’s EHCP was noted, possibly explaining why such support had not been provided, and highlighting the link between this theme and ‘amount and type of support’.

*“There's also poor recognition to what's on the EHCP…” (Parent quote)*

d) Unhelpful learning environment

Factors included under ‘unhelpful learning environment’ were those which created unhelpful settings for children to learn in. For example, class sizes were reported to be too large for effective learning. One parent stated that their child was in a classroom within a special school with *“too many pupils with extreme needs and behaviours”* which resulted in *“volatile”* classes and *“very little access to learning”*. Another parent also commented that there was *“no safe space”* for their child at school, and *“multiple changes”* to their child’s one to one support. Lack of funding was implicated in creating such environments, highlighting the link between this theme and the theme, ‘limited resources’.

e) Limited resources

Many parents reported that their child’s school had limited access to certain resources which in turn hindered the schools’ ability to implement support. Funding was often cited as lacking and as one of the reasons why EHCPs were not followed.

| |
|---|
| *“My son is now in a SEN school which is drastically underfunded. Once it was a good school and his education was good.” (Parent quote)* |

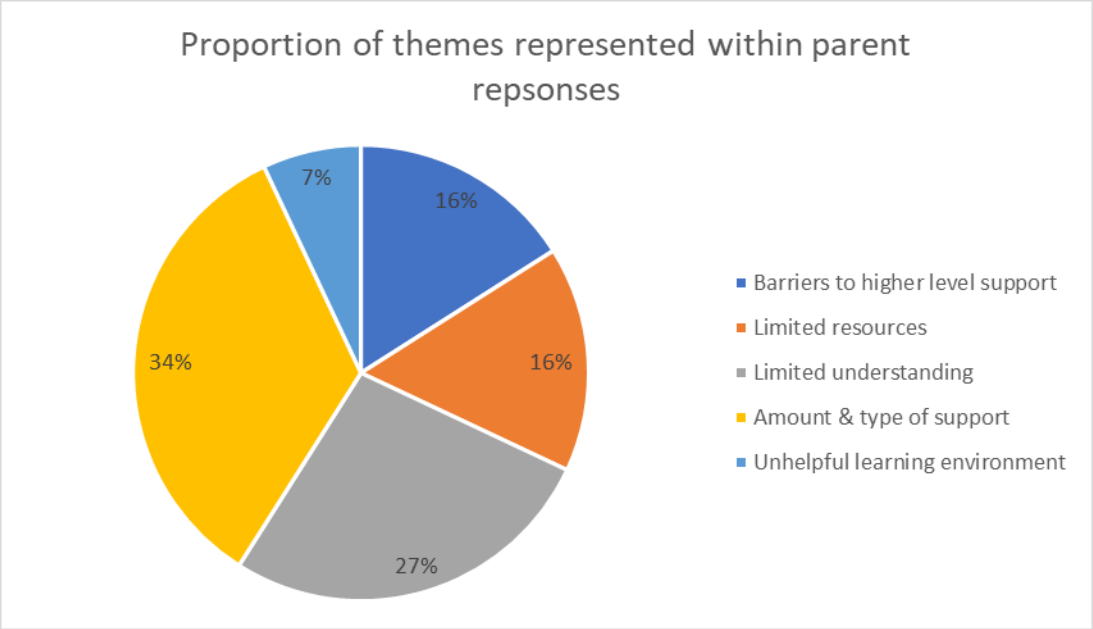
| |
|---|
| *“School are also having their funding cut. They are doing their best with what they have but she needs more.” (Parent quote)* |

| |
|---|
| *“They often don't follow the EHCP. They blame funding.” (Parent quote)* |

Staff shortages were commented on, as well as limited staff training and skills to support children appropriately. One parent reported:

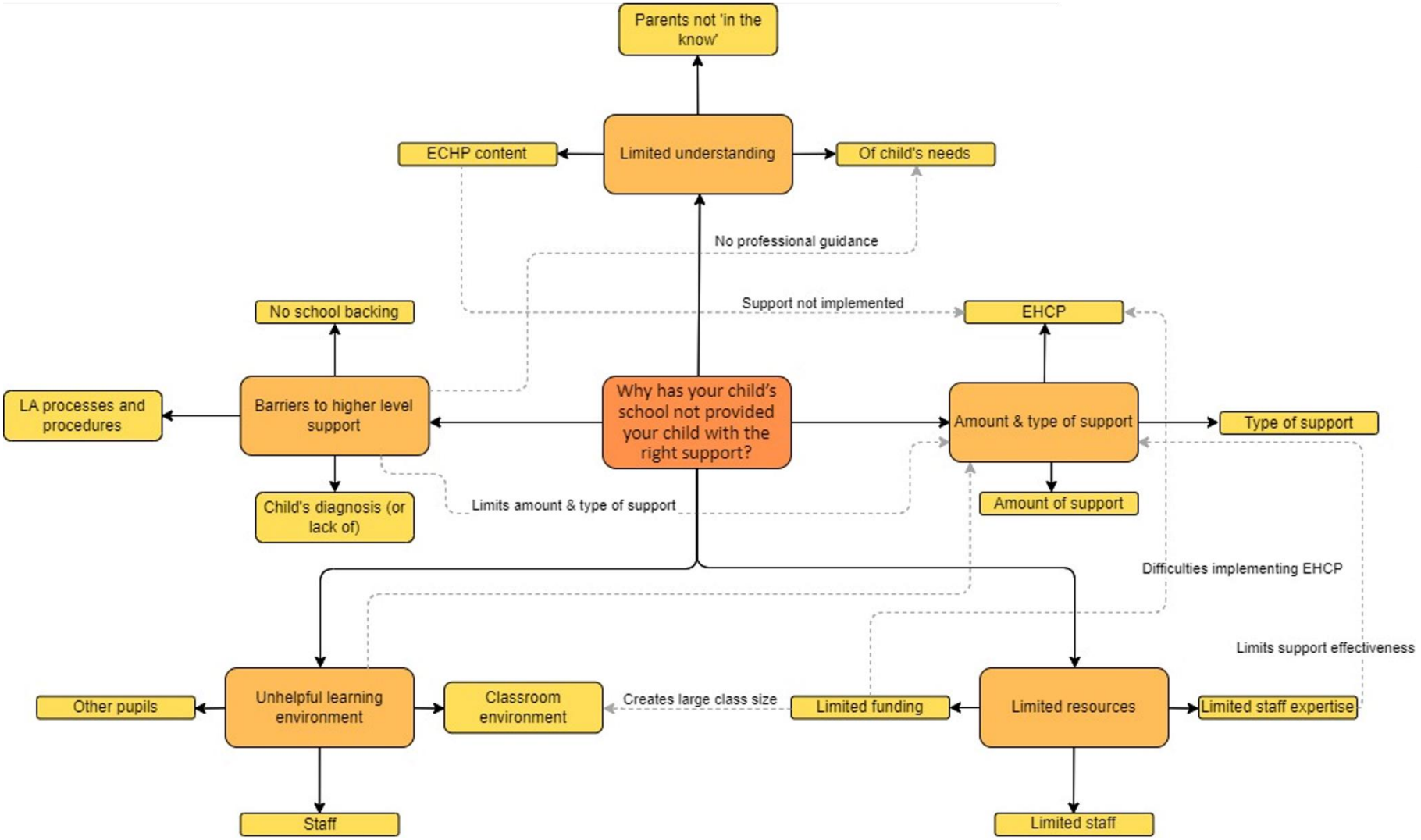
*"… the issues lay with insufficient funding, not enough support staff… and I didn't see any evidence that SENCO staff had the relevant skill set or training to effectively support the learning differences/ difficulties of SEN students." (Parent quote)*

*Figure 3.1 The proportion of data each theme represents within parent responses to the question: "Please briefly explain why your child's school has not provided your child with the right support."*




The theme 'amount and type of support' represented the theme which made up the largest proportion of the data. 'Unhelpful learning environment' made up the smallest proportion. Excerpts of data could be included within multiple themes when appropriate.

*Figure 3.2 Diagram presenting links between themes and codes*



The darker orange bubbles which tail off from the central bubble represent the 5 main themes identified. These bubbles are referred to as ‘theme bubbles’. The lighter orange bubbles which tail off from a given ‘theme bubble’ detail specific factors noted by parents that are thematically tied by their theme bubble (i.e., codes). The light grey dotted lines between bubbles draw explanatory links between separate themes and codes to better depict the relationships between them.

## 3.6 Discussion

This is the first known study to explore parent satisfaction associated with school support received by children with a ND CNV. It is also the first to explore the link between satisfaction and demographic factors, neurodevelopmental and psychological factors, ND CNV inheritance status, school type and EHCP status. Furthermore, I am not aware of another study to investigate the accessibility of EHCPs in this group. To date, the only evidence seemingly available regarding the educational support for children with ND CNVs specifically was anecdotal and so this study adds empirical evidence to these reports.

This is also the first quantitative study I am aware of to investigate parent satisfaction with educational support received by children with likely SEN since Parsons et al. (2009). Since their study, public services, including education, have faced real terms funding cuts (Sibieta, 2022) and recent reviews into the operation of the SEN provision system concluded it was failing to meet the needs of all pupils (DfE, DHSC, 2022; HCEC, 2019). Therefore, updated investigation into the perceptions of educational provision for children with likely SEN was warranted.

This study was inspired by subjective testimonies of parents of children with a ND CNV regarding their school support. These stories highlighted the obstacles parents can face when trying to obtain this kind of provision and the frustration associated with such experiences, and so this study is likely of considerable relevance to parents.

### 3.6.1 Findings and implications

In line with Parsons et al. (2009), the majority of parents with available data reported their child’s school had provided them with the right support (76.9%). This was a

positive yet surprising finding considering the conclusions of the recent evaluations of the SEN system (DfE, DHSC, 2022; HCEC, 2019), as well as the experiences the Rare Genetics Research Group and I have often heard from parents. The findings of this study suggest that many schools are doing a good job in supporting this group of vulnerable children. This should be acknowledged, as many teachers report feeling undervalued in their work (Ofsted, 2019), which is in part contributing to the decision made by some to leave the profession. Better acknowledgement could help educators feel better appreciated, and subsequently help to improve retention rates.

Although problems with educational provision may not be as widely felt by parents as first assumed, we must not disregard the fact that almost a quarter of parents reported they were dissatisfied with their child's support. This is arguably the finding that should be given the most importance here. Although the 'status quo' is seemingly working for most children, it is unacceptable to excuse the need to make improvements for those for whom it is not.

The qualitative insights provided by parents implicate unsatisfactory learning environments for children with ND CNVs, barriers to accessing provision at a higher level than what schools can provide alone, limited understanding of children's needs and limited resources, all of which seemed to impact the amount and type of support children receive. Funding deficits were often noted under the theme 'limited resources', suggesting that increased funding for schools and LAs could in part alleviate some of these issues. However, the current budget outlined by government is estimated to be unsuccessful in meeting the financial needs of schools in future years, with spending per pupil expected to be 3% lower by 2024-2025 compared to 2010 levels (Sibieta, 2022). Longitudinal research which tracks parents' satisfaction and children's outcomes alongside the level of funding received by schools could provide important insight into the importance of appropriate funding for satisfactory SEN provision. Additional future research could also investigate the qualitative issues identified here to gain a greater understanding of these issues compared to what could be obtained via the self-report questionnaire utilised in this study. Teachers' limited ability to access the expertise they needed in order to effectively support children when that child did not have higher-level SEN support was also

noted. This was reflective of issues concluded by Ofsted (2019) impacting teacher wellbeing.

Quantitative analysis revealed that parents of children who met criteria for ADHD, ODD/CD or EP were more likely unsatisfied with their child's educational support. This finding is in line with those of Parsons et al. (2009), who reported that parents of children with 'psychological difficulties' were the least likely to be satisfied with the support provided by their child's school compared to children with alternative SEN. Furthermore, this finding supports the conclusions of exclusion studies, which report higher rates of exclusion amongst children SEMH related SEN. This implies a failure of early identification of need in this group and thus, limited contact with professional support services (Whear et al., 2014). The current study adds to the literature which indicates children with behavioural and emotional difficulties are a particularly likely group to have unmet need which in turn could impact educational experience. Only ODD/CD survived all the multivariate regressions into which different covariates were added, suggesting that this group could be particularly vulnerable. Increased vulnerability could reflect stigma and negative perceptions towards ODD (Clarke & Van Ameron, 2015), leading children with ODD to be less supported (Fadus et al., 2020), despite poor outcomes observed in this group (Burke et al., 2014). It is possible that ADHD and EP did not remain significant predictors of satisfaction in all the multivariate regressions due to relative decreases in sample size. The ORs within the respective models still indicated that parents of children with ADHD or EP remained less often satisfied when the additional covariates were added, however future analyses in a larger sample would help to determine whether these observations failed to reach significance because of sample size or true effects.

Other than in the ODD/CD model, school type was surprisingly not associated with parent satisfaction. This was not consistent with the findings of Parsons et al. (2009) and could indicate that the challenges facing mainstream schools in meeting need are now being more widely felt by special schools too. This was implied in one parent's qualitative response when they reported that their son's special school which had previously been a *"good school"* had become *"drastically underfunded".*

Parent satisfaction was consistently associated with EHCP status, with parents of children with an EHCP more likely satisfied with their child's support at school. This

finding was in keeping with Parsons et al. (2009). Children with an EHCP are also less likely to be excluded compared to children with SEN who do not have an EHCP (Timpson, 2019) again, suggesting children with such support could be more likely to have their needs satisfied. Alternatively, lower exclusion rates in children with an EHCP could reflect the guidance which better protects such pupils from exclusion compared to those with general SEN support (DfE, 2022).

Demographic factors were not associated with either parent satisfaction or EHCP status indicating that children with ND CNVs across demographic groups are vulnerable to not having their needs supported adequately at school and could face challenges securing EHCP support. Of course, not all children with an ND CNV may need an EHCP. Indeed, 19% of children who did not have an EHCP had parents who reported they were satisfied with the support their child received at school. However, if teachers' and parents' assumptions are correct and the SEN system favours wealthier and more highly educated parents (HCEC, 2019), the fact that families with higher SES were no more likely to have an EHCP could indicate that socioeconomic advantage/disadvantage may not apply to parents of children with ND CNVs. Instead, there may be challenges experienced by families with a child with a ND CNV felt by parents from all sociodemographic groups. Limited understanding of children's genetic condition was noted by some parents within their free text responses and may reflect an example of such a challenge. The severity of children's challenges could also confound this finding and is discussed further below. In reality, the relationship between SEN support and SES is complex and nuanced, with many additional influential variables not studied here playing their part. Thus, these are all tentative hypotheses which certainly do not explain the full picture.

It could be argued that it is unreasonable and indeed unrealistic to expect all professionals working with children to have in-depth knowledge about each of the rare genetic conditions they may encounter. However, as discussed by Reilly (2012), general understanding of aetiology of need could help professionals identify specific need within the classroom, as well as plan for potential future challenges, aiding early implementation of the most appropriate and targeted interventions. However, to make this ideal a reality, professionals need to have access to information to help them understand the needs associated with ND CNVs. Researchers and parent

groups have created resources and guidelines for educators to inform them about ND CNVs and to help them support children with these conditions in the classroom. For example, researchers at the University of Surrey and the Cerebra Network for Neurodevelopmental Disorders have recently created an online interactive resource website for teachers, providing information to help educators understand genetics, as well as the neurodevelopmental conditions commonly observed in children with genetic syndromes (https://www.findteacherresources.co.uk/). It also offers ideas and provides additional resources as to how to meet the needs of this group. Effort should be made to increase awareness of existing resources for schools, as well as identifying and addressing their remaining information needs.

Just under 70% of children in this study had an EHCP. Indicative ASD, ID and 'any condition' were the only neurodevelopmental/psychological measures which predicted EHCP status. This could indicate that children with indicative ASD and ID presented with the most severe needs, or that teachers and LAs are most familiar with the difficulties associated with these conditions. Increased awareness of ASD is likely a positive impact of the efforts of advocacy groups who have done much work to get the needs of those with ASD on the political agenda (e.g., The Autism Act, 2009). Indeed, ASD is noted as the most common primary need of children with an EHCP (ONS, 2023). However, there is evidence that clinicians in other nations (Belgium and Australia) can sometimes feel pressure to diagnose individuals with ASD because they can act as a 'ticket' to additional provision, including educational provision (Jacobs et al., 2018; Skellern et al., 2005). The need for an ASD diagnosis to access specialist support was in fact noted by parents in this study, which could indicate an overreliance on ASD diagnoses to grant access to support in the UK too. Whilst it is positive that children with ASD are getting the support required, it is important that other SEN needs are given just as much attention to ensure that all children are adequately supported and to decrease the demand on ASD diagnostic services which are struggling to keep up with demand (British Medical Association, 2019).

Indicative ASD and ID both remained significant predictors of EHCP status after controlling for demographic covariates and inheritance status, which itself was consistently predictive of EHCP status. Children with an inherited ND CNV were less

likely to have an EHCP, which could signify that parents with the genetic condition themselves may face additional challenges compared to parents who do not have the genetic condition in securing support for their children. Wolstencroft et al. (2022) reported that children with inherited ND CNVs were at increased risk for certain neurodevelopmental and psychiatric conditions. Cunningham (2022) also observed increased nonspecific emotional and behavioural problems in children with inherited ND CNVs. Neurodevelopmental and psychological group differences regarding inheritance status were inconsistent in this study (i.e., the de novo group more often met criteria for some conditions and the inherited group more often met criteria for others) and were largely non-significant. However, children with a *de novo* ND CNV were more likely to meet criteria for ID, which was in turn positively associated with EHCP status and could partly explain why children with a *de novo* CNV were more likely to have an EHCP.

Wolstencroft et al. (2022) also reported that children with inherited ND CNVs were more likely to live in lower socioeconomic areas and suggested cognitive impairment within affected parents as a potential explanation. Whilst not a significant finding, children in this study who had an inherited ND CNV were more often reported as having a lower family income compared to the *de novo* group. Furthermore, mothers of children with an inherited CNV were less likely to have a university degree. Both these findings could suggest that the inherited group in this study were from lower socioeconomic backgrounds. Should the hypothesis of Wolstencroft et al. (2022) be correct, cognitive impairment in parents with a ND CNV could disadvantage them in obtaining educational support for their child and they therefore may need additional support from services to ensure their child's needs are appropriately supported to help prevent intergenerational cycles of disadvantage. Equally, children with inherited CNVs in this study could have more often lived in lower income households for alternative reasons, for example a higher proportion of their parents could have been unable to work because of their caring responsibilities. Therefore, further investigation is needed to substantiate the hypothesis that parents who have a ND CNV have additional difficulties securing support for cognitive reasons.

### 3.6.2 Limitations

This study has several limitations. Firstly, there is no gold standard research measure to assess whether children require additional support at school. Even formal assessments conducted by LAs do not always correctly identify need, indicated by tribunal outcome figures which show the percentage of rulings which are upheld in full or in part (i.e., families are granted all or some of the support they had requested but which had previously been denied by the LA). For example, The SEN Tribunal for Wales Annual Report (2022) reported that 9% of hearings in 2020-2021 had these outcomes. Nine percent might appear a relatively small figure, but notably 26% of tribunal cases had already been conceded by LAs before the tribunal date.

Formal assessments conducted by LAs involve acquiring a range of information from several sources including the child's parents and school, medical professionals, educational psychologists, social workers and in some instances the child themselves. Acquiring information to such a degree was not possible within the scope of this project and therefore I used the neurodevelopmental and psychological profile of children as obtained from interview with the primary caregiver as an indication as to whether children could need such support. Arguably, not all children who meet criteria for a such conditions will need SEN support or an EHCP, however this is an exploratory study and still provides an indication that not all children who may need support receive it, and that there are differences in the accessibility of support for different children. Furthermore, the parent satisfaction measure provided additional indication that some children are not supported to the level parents deemed adequate.

Regarding my measure of satisfaction, a single item within the SEQ was utilised and therefore could suffer from issues associated with single item measures (discussed in Chapter 2). However, as noted in Chapter 2, single items could be favourable when measuring unambiguous concepts (Wanous et al., 1997). The question "Has your child's school provided the right support for your child?" is arguably unambiguous. On the other hand, this question might have benefitted from increased specificity to prompt parents to consider all the elements of support children could receive at school (e.g., support with schoolwork, social support, emotional support). Without such direction, parents will have answered this question with their

understanding of what support at school should look like, which may not align with what can feasibly be provided, or the support their child could be legally entitled to.

This study only collected information from parents, yet parent opinion regarding whether they receive the right support at school could differ to the opinions of the child's teacher and to those of the child themselves. In fact, some of the parents in this study reported they were not well informed about the support their child received, indicating that parents may not always have the information to be able to properly assess whether their child is being supported adequately. Additionally, parents were the only reporter of ECHP status, however, Parsons et al. (2009) reported 12% of parents were unsure as to whether their child had a statement. Whilst I excluded participants who gave discrepant answers to the self-report and researcher interview measures of EHCP status to eliminate parents who were likely unsure about the support currently received by their child, I cannot be sure that the ECHP status recorded for all children included in these analyses was accurate.

Child's ethnicity was not controlled for in any of the analyses presented, for the same reasons described in **Chapter 2**. Ethnicity will be an important factor to consider in future studies investigating parental satisfaction and should be possible as sample size for the IMAGINE-2 and DiGEN studies improve.

Some of the study findings suffered from low power and therefore I cannot be confident that the null hypothesis has been correctly rejected in all analyses presented. Some of the models presented in this chapter likely suffer from low overall and sub-sample size. Continued data collection will help to increase the power of future studies and determine whether these findings are replicated in a larger sample. Furthermore, my analyses were cross sectional and therefore, causal conclusions cannot be made.

The $R^2$ associated with each of the logistic regression models performed were small, indicating that the effect of each of the predictors analysed on parent satisfaction and EHCP status was minimal. Additional factors as to why parents are unsatisfied with their child's support and why children do or do not have an EHCP should be also investigated. For example, children are eligible for EHCPs for reasons other than

their neurodevelopmental and psychological presentation which could provide a stronger explanation of EHCP status (e.g., physical health conditions).

## 3.7 Conclusion

The aim of this chapter was to assess whether children with ND CNVs are adequately supported at school according to their parents and to identify factors which may impact children's access to educational support.

Nearly 25% of parents reported that their child's school had not provided them with the right support. Children's neurodevelopmental and psychological profile predicted parent satisfaction, and children with emotional and behavioural conditions represented a particularly vulnerable group. This was in keeping with the conclusions of other researchers and the recent SEN review.

Parents of children with an EHCP were more likely satisfied with their child's educational support, which again, replicated previous findings. Neurodevelopmental and psychological profile also predicted EHCP status. Children with ID and ASD were more likely to have an EHCP, possibly indicating that these children display more obvious or well understood needs, or that the activism of ASD advocacy groups has had beneficial impacts.

Children with an inherited ND CNV were less likely to have an EHCP, indicating that parents with ND CNVs themselves may face additional challenges compared to parents who do not have the condition when trying to obtain support for their children, or that challenges experienced by both groups are harder to overcome for the former. Issues associated with lower socioeconomic status may be implicated.

These findings could help to explain some of the observed differences between children with ND CNVs. Those in receipt of satisfactory educational support are likely better equipped with knowledge and skills to help them in immediate and later life, likely improving their outcomes. Findings could also help to identify children who are at most risk of being missed by services, either because of the needs they present with or because of additional challenges associated with ND CNVs experienced by parents.

So far, this thesis has presented findings regarding the social challenges experienced by children with ND CNVs, as well as access to, and satisfaction with additional educational support. The next chapter provides further qualitative information on these experiences, providing more personal and in-depth accounts to help ‘fill some of the gaps’ regarding the topics already covered.

# 4 Obtaining educational support and the impact of support once received: The experiences of mothers of children with rare genetic conditions associated with high risk of neurodevelopmental difficulties

## 4.1 Chapter overview

Life at school for children with a ND CNV is likely difficult for some. This group are at increased risk of bullying by their peers compared to controls and, according to their parents, some are reportedly inadequately supported at school. However, our understanding about the factors which lead to difference in experiences at school for children with ND CNVs is incomplete. Furthermore, the consequences of these experiences on the child and their family can only be inferred by considering studies investigating these experiences within the general population, or populations with similar challenges.

This chapter presents qualitative data collected via semi-structured interviews with 17 mothers who had a child with a rare genetic condition associated with high risk of neurodevelopmental difficulties. Findings provide detailed insights into the factors which affect access to SEN support, and the impact of support once received, not only on children with ND CNVs, but also their family. It also provides better understanding of parents' experiences of obtaining SEN support for their child and how these experiences impact their own mental health.

The following main themes were identified after conducting framework analysis (FA): a fight against 'the system'; shining a light on social inequalities; and the benefits and limitations of support. Mothers reported they must engage in a continuing 'fight' with public services to secure and maintain SEN support and to navigate a SEN provision system which is under resourced and lacks knowledge about the challenges experienced by the children it aims to support. The system highlights inequalities within society and could also perpetuate certain inequalities. Children who were supported at school showed improved social and academic experiences, however challenges remained when schools could not implement truly inclusive school environments.

Results of this chapter support the need to improve the SEN support system to make it more accessible for all families so that all children have an equal opportunity to experience its benefits and to protect the mental health of mothers when navigating it.

## 4.2 Introduction

Findings presented in the previous chapters of this thesis suggest that the school days of children with a ND CNV can be challenging. Children with ND CNVs are more likely than their unaffected siblings to be bullied by their peers (Chapter 2) and a significant proportion of children with a ND CNV (almost 25%) receive an unsatisfactory level of educational support according to their parents (Chapter 3).

Experiences between children were variable and the previous chapters have, for the first time, elucidated some of the factors which seem to contribute to bullying experiences and parent satisfaction with the support received by their child at school. Children who met diagnostic criteria for ADHD, probable ASD and psychotic experiences were particularly at risk for bullying, as well as children who displayed higher symptom counts of these 'conditions'. Higher neurodevelopmental burden (i.e., the number of neurodevelopmental 'conditions' children met criteria for) was also associated with higher risk of being bullied. Risk also seemed to increase with age. Parents of children who met criteria for ADHD, or ODD or CD were more likely to report they were unsatisfied with their child's educational support whereas parents were more likely satisfied if their child had an EHCP. Parents of children with probable ASD, ID or a *de novo* ND CNV were more likely to have an EHCP indicating that educators may be more attuned to the needs of children with ASD and ID, and that parents with a ND CNV themselves might experience increased challenges when trying to obtain higher levels of support for their children. However, our understanding about which children are more or less likely to experience peer problems or be unsupported remains incomplete. Chapters 2 & 3 do not explain the full picture, and there remain unexplored factors which likely impact bullying and support experiences at school which warrant further investigation.

The previous chapters also do not tell us about the impacts of bullying and SEN support experience on children. Research exploring the impact of peer victimisation

in the general population shows that such experiences leave children vulnerable to immediate and long-term negative impacts on psychological and physical health and also limit their later socioeconomic opportunities relative to their peers (Takizawa et al., 2014, 2015). The literature investigating the impact of insufficient educational support is limited, however if we consider educational support as an enabler for children with SEN to access their school's curriculum (i.e., to access education), it could be argued that without such support children are unable to experience the benefits of education. These include increased opportunity for social mobility (UNESCO, 2019) and health equity (CSDH, 2008). By generalising the impacts of victimisation and lack of access to education within the general population to children with ND CNVs, we can assume that children with a ND CNV who are bullied by their peers, or whose needs are unsupported at school will also be vulnerable to these outcomes. However, without research investigating the impact of these experiences in this group specifically, these assumptions can only be made tentatively.

Assumptions can also only be made regarding the efficacy of proposed interventions which aim to mitigate against harmful experiences. For example, after failings were identified within the current SEN provision system (DfE, DHSC, 2022; HCEC, 2019), the UK government have outlined plans to address the issues (DfE, DHSC, 2023). However, without investigating the experiences of children with ND CNVs specifically, additional challenges potentially experienced by this group will remain unaddressed by current plans. Therefore, further investigation which gathered detailed accounts of children's school experiences and their impacts on children to enrich the findings of the previous chapters was justified.

There has been little consideration within this thesis thus far regarding the impact of children's experiences on the rest of their family. The Cerebra Steering Group advised that a child's experiences not only influence the child, but also the lives of their family members. Similarly, the school experiences of children may be impacted by family factors outside of the school. For example, the steering group noted that if a child's parents are struggling with their own mental health, they might have limited wherewithal to secure SEN support for their child. Indeed, mothers of children with ND CNVs are at increased risk for psychiatric problems compared to mothers in the

general population (Baker et al., 2021; Niarchou et al., 2022). Therefore, it is important to study children within the wider context of their family.

After taking a predominantly quantitative approach to studying the school experiences of children with a ND CNV, this chapter sought to gain a more in depth understanding about children's experiences and how these experiences impact both the child and their family by implementing qualitative methods.

### 4.2.1 The differences between quantitative and qualitative methods

The fundamental differences between quantitative and qualitative methods can be understood in greater depth by considering their core aims and values (**Table 4.1**). Whilst quantitative approaches seek to test pre-existing theory and hypotheses, truly qualitative approaches aim to generate theory by better understanding phenomena within a particular social context (Rocco et al., 2003). The principal aims of the two approaches are reflected within their opposing philosophical positionalities. Positionality broadly refers to a researcher's individual view of the world, which can be understood by qualitative and qualitative ontological and epistemological assumptions.

Easterby-Smith et al. (2021) defined ontological assumptions as the those which we make about the nature of reality. In other words, whether our understanding of the world is based on subjective or objective reality (i.e., how true one individual's reality is for another individual). Traditional qualitative approaches argue for multiple subjective realities, however, quantitative approaches value objective reality and truth which can be generalised beyond one individual's experience.

Epistemological assumptions are strongly linked to ontology and are the assumed acceptable foundations of knowledge (Bahari, 2010) (i.e., acceptable methods of acquiring knowledge). Qualitative methods which adopt a subjectivist school of thought place importance on participant and researcher subjectivity during data collection, analysis and interpretation. Findings are recognised as being influenced not only by participants but also the researchers' own life experiences and political, social and cultural values. Such values are reflective of interpretivist ontological assumptions. Quantitative methods however align with a positivist ontological perspective which posits that experience can and should be measured via objective

methods (e.g., standardised questionnaires). They seek large and representative samples with the aim to reliably generalise findings to describe experiences of wider populations.

The conflicts between underlying quantitative and qualitative assumptions have been referred to as 'the paradigm wars' (Bryman, 2006). Some researchers, who are sometimes referred to as 'purists' remain heavily in one camp, favouring one approach over the other. However mixed methods research is now common, indicating that disciplines which traditionally favoured one practice are beginning to recognise the contributions both methods can make to their work. A mixed methods approach is followed when researchers collect and analyse both quantitative and qualitative data at some stage within the process of a single study to obtain a more complete understanding of their research question (Creswell, 2012).

*Table 4.1 The aims and values which underlie qualitative and quantitative methods.*

| | **Qualitative** | **Quantitative** |
|---|---|---|
| Principle orientation to the role of theory in relation to research | Inductive; generation of theory | Deductive; testing of theory |
| Epistemological position | Interpretivism | Positivism |
| Ontological position | Subjectivism/ constructionism | Objectivism |

*Adopted from Bryman (2004)*

## Reasons for taking a qualitative approach for this study

The findings of Chapter 2 & 3 provide evidence that bullying, and unsatisfactory SEN support are likely problems facing some children with ND CNVs. However, the results do not give deeper insight into the reasons why experiences may differ beyond those discussed above, or the impact of such experiences on children or their family. This knowledge is important to better understand whether the assumed negative impacts of these experiences are indeed harmful, whether existing support strategies and policies are likely to be effective, and finally, which children and families support strategies could be targeted to.

The open nature of qualitative inquiry provides participants the opportunity to answer questions in a way that best reflects their experiences, giving researchers a better

understanding about the range of contextual factors which contribute to participant experience (Sofaer, 1999). I would be less able to gather a good understanding of such nuance if quantitative methods were used, whereby participants' answers are confined to closed multiple choice questions predefined by myself, the researcher.

The understanding of nuance can be helpful when investigating a new area by informing researchers about which questions are the 'right' ones to ask participants at a later date via quantitative methods to reliably depict experience at scale (Sofaer, 1999). Justification to study the school experiences of children further was provided by Chapters 1 & 2, but qualitative enquiry was deemed a helpful next step to best ensure that future quantitative investigation taps into the factors most strongly associated with certain school experiences (i.e., the right questions are asked) and provide a meaningful reflection of such experiences and impactful research translation.

Throughout this thesis, I have justified the need to conduct this research, and built my hypotheses, based largely on studies which explored the school and social experiences of children within the general population, or children with conditions associated with ND CNVs (e.g., children with neurodevelopmental conditions). Although children without an ND CNV who experience similar difficulties may face comparable challenges at school, it is important to understand whether there are experiences unique to children with ND CNVs specifically. Without such understanding it will be difficult to inform support which addresses all the potential challenges experienced by children with these genetic variants. Again, the opportunity qualitative methods provide to participants to share their unique experiences more openly was deemed an effective way of better understanding whether there are any experiences unique to children with ND CNVs. This insight will hopefully enable researchers to build future theories, hypotheses and research designs which are more specific to this group.

## 4.3 Aims

**Aim 1:** Explore the experiences of parents when obtaining educational support for their child with a rare genetic condition associated with high risk of neurodevelopmental difficulties.

**Aim 2:** Explore the impacts of parents' experiences of obtaining educational support on them and their family.

**Aim 3:** Explore the impact of educational support on children who have a rare genetic condition associated with high risk of neurodevelopmental difficulties and their family.

## 4.4 Methods

### 4.4.1 Recruitment

#### Participants

Similar to Gallo et al. (2008), I adopted a 'non-categorical approach' to genetic conditions when recruiting participants. As applied to this study, this approach assumes that children with genetic conditions who share common neurodevelopmental challenges experience similar challenges at school regardless of their specific genetic condition. Therefore, individuals were eligible to take part in this study if they were a parent of a child who had a ND CNV or a child with another rare genetic condition associated with neurodevelopmental conditions. I decided to take this approach in the hope that it would increase the size and demographic diversity of the sample.

I had initially intended to also recruit children with such genetic conditions, as well as teachers into this study. However, due to the COVID-19 pandemic and the subsequent pressure on schools during this time, I was unsuccessful in recruiting any teachers into the study. Children were also difficult to recruit. In response to COVID-19 related government guidance to restrict travel and social contact, and considering the clinical vulnerability of this group, I had planned to conduct the interviews with children via online video call. However, when trying to recruit children, several parents advised that it would be difficult to engage the children in the interview online. Therefore, I decided to halt the recruitment of both teachers and children and recruit parents only.

Sampling strategy

Given that participants were recruited on the basis of having a child with a relevant genetic condition, a purposive sampling method was followed. To obtain a greater understanding about the breadth of participant experience I sought to recruit as diverse a sample as possible in regard to the characteristics listed within **Table 4.2**. However, given the rarity of the conditions of interest, I was aware that the characteristics represented would be largely dependent on who volunteered to take part in the study.

'Saturation' is a common method used by qualitative researchers when justifying their sample size. Saturation was first defined by Glaser and Strauss (1967) and signals the point at which researchers are satisfied that no (or not 'enough') new insight into the phenomenon being studied will be obtained from any new participant recruited into the study. Researchers (Braun & Clarke, 2021b; Malterud et al., 2016) have argued that saturation is a positivist concept which has been applied to qualitative research. For example, if knowledge is subjective, how can researchers be confident that the inclusion of one or more participants would not add new insight to the reality of experience shared by the already existing sample. Considering I would be working relatively independently throughout all stages of this study, my approach to data analysis (discussed further below) was already misaligned with more positivist qualitative methodologies in that there was no second coder whom I would compare my analysis to and thus check its 'reliability', another marker of quality taken from positivist approaches. Therefore, saturation did not seem in keeping with my methodology. However, given the necessity to stipulate sample size when applying for ethical approval, I proposed that I would recruit approximately 15 parents. This figure was informed by the work of Guest et al. (2006) who reported that 88% of their codes were developed after analysing 12 out of 60 interviews. To allow for the remaining 12% of codes to be identified, but also considering the rarity of the genetic conditions I was investigating, I aimed to recruit approximately 15-20 participants.

*Table 4.2 Participant characteristics I sought to have represented within my qualitative sample.*

| |
|---|
| *Parent characteristics* |
| 1) Socio-economic status (measured by overall Index of Multiple Deprivation (IMD) score) |
| 1st quintile<br>2nd quintile<br>3rd quintile<br>4th quintile<br>5th quintile |
| 2) Ethnicity |
| White<br>Minority ethnic |
| 3) Sex |
| Male<br>Female |
| 4) Age (years) |
| <20<br>20-29<br>30-39<br>40+ |
| *Child characteristics* |
| 1) Ethnicity |
| White<br>Minority ethnic |
| 2) Sex |
| Male<br>Female |
| 3) Inheritance status |
| Inherited genetic condition<br>De novo genetic condition |
| *School related characteristics* |
| 1) Level of support received by child |
| Mainstream school with EHCP<br>Mainstream school without EHCP<br>Special school |
| 2) Stage of school |
| Primary school<br>Secondary school |
| 3) Overall, does the participant's child like school? (parent's opinion) |
| Yes<br>No |

Recruitment strategy

The opportunity to take part in the study was advertised via the Rare Genetic Research Group's social media pages which included their Twitter, Facebook, and Instagram pages. I also contacted charities such as Unique and Cerebra to ask for their help in circulating information about the study to their networks via their social

media pages and newsletters. Families who had previously taken part in the DiGEN and IMAGINE-2 studies were contacted by the Rare Genetic Research Group via email to inform them about the study.

Parents who were interested in taking part got in contact via email. I then contacted them by phone to explain the study in more depth, to determine whether their child had a genetic condition which made them eligible to take part and to schedule a future date for the interview to take place. This first conversation via telephone helped to build some initial rapport with participants before the interview date. I felt this was particularly important given that interviews were being conducted online.

After speaking with participants on the phone, I emailed them a copy of the study information sheet and consent form to allow them to familiarise themselves with these documents and contact me with any questions before the interview.

### 4.4.2 Measures

Developing the interview schedule

Considering the school experiences of children with ND CNVs is an understudied topic, there was little literature to formulate my interview questions from. To develop a schedule which facilitated exploration of the issues that were most pertinent to parents, I asked members of the Cerebra Steering Group to share with me via email which aspects of their child's school experience had been particularly challenging for them and their child and therefore should be explored further. After reading through their responses, I created an interview schedule to encapsulate the issues shared.

**Figure 4.1** shows the parent interview schedule. This was used as a guide when interviewing participants. The schedule had 4 main themes: your child in school; support at school, relationships, impact of experience.

*Figure 4.1 Final parent interview schedule*

**Parent interview schedule**

Begin each interview explaining the study, its background and its aims, and question topics.

Parent Interview

**Introductory questions**

1) Collect demographic data about the parent *(their age, sex, ethnicity, and postcode)*
2) Collect demographic data about the child *(their age, sex, ethnicity, their genetic condition, is the genetic condition inherited or de novo)*
3) School related information:
   a) What school year is your child in?
   b) What type of school does your child go to *(special or mainstream)*?
   c) Do they have an EHPC?
   d) Overall, do you think your child likes school?

**Theme 1: Your child in school**
1) How do you think your child generally feels when they are in school?
*- What do you think makes them feel this way?*
2) What does your child like about school?
3) What does your child dislike about school?

**Theme 2: Support at school**
4) What, if any, support does your child receive at school?
5) What support does your child need at school?
6) (If relevant): What has your experience been like to obtain support for you child at school?
7) How has the level of support your child receives impacted them?

**Theme 3: Relationships**
8) How does your child get along with other children at school?
*- Has your child got a best friend or close circle of friends?*
*- Has your child experienced bullying?*
9) How does your child get along with the teachers at their school?
10) How has your child's relationships with others at school impacted them?

**Theme 4: Impact of experience**
10) How has your child's experience of school impacted them?
*- e.g. at home, academically, socially, emotionally*
11) How has your child's experience of school impacted you and the rest of your family?

Conducting the interview

All interviews were conducted online and audio-recorded, with consent, via Zoom.

After a few minutes of general conversation to help ‘break the ice’, participants were given the opportunity to read through the participant information sheet in case they had not already done so. Participants then completed the consent form virtually via REDCap. REDCap is a secure online platform which researchers can use to build and manage databases. I sent participants a unique weblink via the ‘chat’ function in Zoom which took them to their individual consent form to complete within REDCap.

After the consent form had been completed by participants and checked by myself, I introduced the main themes of the interview to prepare participants about the topics that were to be covered. I made it clear that parents did not have to answer a question if they did not wish to and that they could take a break or ask me questions at any point during the interview.

To begin the interview, I first ran through the introductory questions to collect information about participants’ characteristics as per **Table 4.2**. Then, I asked participants the first question under the first theme on the interview schedule (‘Your child in school’). The interview followed a semi-structured format whereby participants were asked each question on the schedule, but individual follow up questions were posed to participants based on their respective answers to gather greater detail on the points raised. As this was the first known study to qualitatively explore this group’s school experiences, I let mothers answer questions at length. I did not want to limit their answers too soon as such information could provide insight into an important aspect of school experience not included in the interview schedule. However, if participants veered too far off topic, I tried to bring the interview back on course. I asked questions in the same order as listed on the interview schedule, however if participants had already provided information relevant to a later question whilst answering an earlier one, I did not repeat it. Likewise, if parents began talking about their child’s relationships within the ‘support at school theme’ for example, I stayed on topic and posed the questions about their child’s relationships first, and subsequently circled back to the support at school questions.

Once each item on the schedule had been covered, I asked parents if there was anything else they wished to share before we ended the interview. I then concluded the interview by thanking them and explaining the next steps to them (i.e., completion of data collection, data analysis and write up). I informed them that if they

thought of any further information that they had not already shared and would like to do so, that they could email me and that I would take this new information into consideration when analysing the data.

Finally, I followed up each interview by sending an email to participants to thank them once more and send them a copy of their signed consent form and a study debrief form. This document listed who to get in contact with if they had any concerns arising from taking part in the study.

### 4.4.3 Analysis

Before analysing the data, I transcribed 16 of the 17 interviews. One of the interviews was transcribed by a Cardiff University psychology student who was completing a year's professional placement with the Rare Genetic Research Group. To protect anonymity, all participants were given a pseudonym. Any other identifiable data (e.g., child's name) were excluded from the transcripts.

#### Framework Analysis

I conducted FA to analyse the interview data. FA is a type of thematic qualitative analysis developed by Ritchie and Spencer (2002) for the purposes of applied policy research, however has since become a widely used method in many disciplines (e.g., psychology (Parkinson et al., 2016) and health care (Gale et al., 2013)). Thematic analyses is a "family" of methods (Fugard & Potts, 2019) used to analyse qualitative data and involves the development of 'themes' (patterned meanings within data (Braun & Clarke, 2006)) to effectively describe data. The feature of FA which differentiates it from most other thematic approaches (e.g., reflexive thematic analysis) is the formation and application of an analytical framework throughout the analysis to help the researcher map out their ideas as they progress through the process.

##### *Why I used FA for this study*

Braun and Clarke (2021a) advise that there is rarely only one qualitative analysis method suitable for a given study and the choice of method is commonly based on

conceptual and pragmatic reasons, as well as the researcher's experience and comfort with the approach.

As a relatively new qualitative researcher, the use of a framework throughout the analytical process appealed to me as it provided some structure to help organise an otherwise overwhelming amount of text. Secondly, I was receiving guidance from a qualitative researcher who is highly experienced in FA. Therefore, I used FA because of the structure it provided and because I was confident that I had an appropriate level of input to make sure I was following the method correctly.

Secondly, FA is not wedded to a particular epistemological/ontological position and researchers can approach the data inductively or deductively. I adopted an inductive and iterative method of analysing my data in that I did not approach the data with predefined themes for which I was searching for evidence and themes were not settled on early but evolved throughout the analytic process.

I sought to speak to parents with different demographic characteristics and therefore had too large and diverse a sample compared to other thematic approaches such as interpretative phenomenological analysis (Smith, 1996) which requires a fairly small and homogenous sample.

Finally, Ritchie and Spencer (2002) advise that their method be used for studies with research questions which fall into one of the following categories:

1. Contextual questions: e.g., the nature of peoples' experiences, the needs of the study population.
2. Diagnostic questions: e.g., what factors underlie attitudes, why are decisions made, why do certain needs arise.
3. Evaluative questions: e.g., how things are achieved, how do experiences impact behaviour.
4. Strategic questions: e.g., what is needed to meet need, how can operations be improved.

Whilst these categories were originally specified for applied policy researchers, they can be applied to research questions of studies from other disciplines when evaluating whether FA is an appropriate method (Parkinson et al., 2016). I assessed

that my research questions were aligned with both contextual (experiences) and evaluative (impacts) type questions.

FA has a series of steps to follow (Ritchie & Spencer, 2002) which provided good instruction for a relatively inexperienced qualitative researcher. I will present each of the stages here and describe the processes I followed for each of them. In their paper, Parkinson et al. (2016) outline the way in which they conducted each of the stages. I found this paper particularly helpful as an additional resource to guide my analysis.

### Stage 1: Familiarisation

During the familiarisation stage researchers are looking to 'immerse' themselves in their data to gather an overview of the material they have collected. This is an important part of the process and seen in other methods of thematic qualitative analysis.

Although conducting the interviews provided me with good knowledge about their content, data collection does not suffice as quality familiarisation. Therefore, to engage with the data further, I transcribed the interviews myself as opposed to outsourcing transcription to a third-party service. As already mentioned, a placement student transcribed one of the interviews. This was partly due to time constraints and because the student was interested in listening to an interview to learn more about families' experiences. Once the interviews were transcribed, I relistened to all the interview recordings whilst also reading through the associated transcript. This served the purpose of checking for transcription errors, as well as helping me to familiarise myself with the data further.

### Stage 2: Identifying a Thematic Framework

After familiarising themselves with the data, researchers should build a thematic framework to help them to organise their data by 'key issues, concepts and themes' (Ritchie & Spencer, 2002). The framework is essentially a list of these key issues, concepts and themes, which will now be referred to as 'codes'.

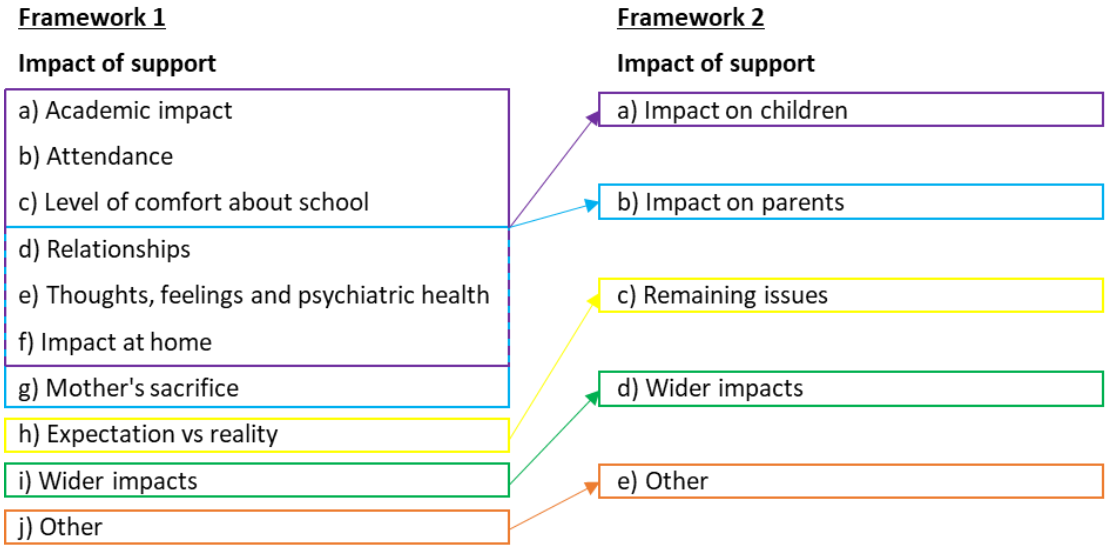
I selected a sub-sample of 4 transcripts and made notes about the points raised by the interviewee relevant to my research aims. I used Microsoft Excel to note my thoughts within separate columns which represented the study's topics of interest (e.g., obtaining support, impact of support). The transcripts for which I made detailed notes were selected based on the characteristic diversity of the participants as per **Table 4.2**.

I then studied these notes and identified recurring topics across and within transcripts relevant to my research aims. These recurring topics formed the main codes within my first framework. Data which were not explicitly related to the study's aims but which I interpreted as important to note were also considered within the framework. For example, my third aim was to explore how the level of support received by the child at school impacted the child and their family. However, impacts beyond the family were repeatedly noted within one of the selected transcripts and therefore also included within the framework.

Next, I applied the framework to 4 further interviews to refine the framework in light of additional information. To apply the framework, I loaded the additional 4 transcripts into NVivo and created what NVivo term 'nodes', which corresponded to each of the codes within my first framework. I then read through each of the 4 new transcripts, and logged excerpts of data under the applicable node. Relevant data which did not fit within my existing codes were logged under 'Other', a code which was included in my first and second framework for this reason.

**Figure 4.2** presents an example of how codes within the first framework were refined within the second. It does not provide the full list of codes, only those relevant to the 'impact of support' aim.

*Figure 4.2 Codes within the first and second 'impact of support' framework*




Stage 3: Indexing

Indexing is the process of applying the framework to the entire dataset to code all data. To do this, I loaded each remaining interview transcript into NVivo and followed the same process as described above to code each transcript with respect to my second framework. The transcripts which I had already coded in respect to the first framework, and the transcripts on which I built the initial framework were recoded using the new framework.

Stage 4: Charting

Once indexing has been completed, researchers begin the charting stage whereby they summarise and log participants' coded experiences within a table (i.e., a chart).

Using Microsoft Excel, I created a separate chart for each respective topic of interest relevant to the study resulting in 3 charts in total: 1) obtaining support, 2) impact of support, and 3) peer relationships. Column headings within the table corresponded to code names (e.g., impact on children, impacts on parents, etc) and each row corresponded to a single participant. Participants were charted in alphabetical order. **Table 4.3** provides an example of some of the charted summaries for one participant within the 'impact of support' chart. 'Good' participant quotes were also included in the chart along with a reference as to where the corresponding data could be found

within the transcript. Good quotes were those which clearly reflected the summarised point noted within the chart.

*Table 4.3 Example of charted summaries for one participant within the 'impact of support' chart*

| IMPACT OF SUPPORT | | | | | |
|---|---|---|---|---|---|
| Participant | Impact on children | Impact on parents | Remaining issues | Wider impacts | Other |
| [redacted] | Child is a completely different child in new school where he is supported (eg - in terms of behaviour and violence). Is more himself: *the teacher in the next school I got him into she actually said to me 'when I'm reading what they describe M as' she said 'I don't recognise him as the same child'...he had on occasions hit out at teachers... but it's not him at all and since getting out of there it's never happened again do you know it was just yeah different child completely (pg 1, line 25)* | When school don't undersatnd how to best support needs they work with parent to come to a solution. Mum says this *makes a hell of a difference (pg 2, line 60)* to parents themselves. Also indicates a certain level of respect for mother's knowledge and child's rights. *Nothing's too much trouble (pg 2, line 60)* - suggests parents don't feel like a nuisance | Even with one to one support at old, class teacher might still intervene which would draw attention to child from other children in class = increased stress. If one to one wasn't confident to be firm with the class teacher this would often happen - positive impacts of support can only be realised if a number of factors come together to work together | Inclusive schools foster a school ethos of acceptance and encouraged other children to want to help support children with SEN, promoting further inclusivity: *they had and ASD and a MLD base within the mainstream school. The teacher in that base made sure all the staff and all the children had awareness sort of sessions and things and then all the members of staff from the mainstream were invited to spend time in the bases and the whole kind of ethos was really like accepting difference in the school... they would kind of give awards to other children in the mainstream for supporting the children in the base and then they would wanna help d'you know so it was really positive (pg 5, line 168)* | |
| | One of the one to ones in previous school who mum credits as saving child - indicates how significant the impact can be on children | | Child is a very anxious child and mum puts that down to his early school experiences. His anxiety can still be triggered by things which remind him of old school indicating lasting impact on mental health. Mum worries this might have life long impacts: *when he started the new school it was all going really well and then all of sudden he started to get really stressed, really anxious, totally out of character and we realised that there was a support worker that had started there that had on a few occasions worked as like a bank support worker in this old school and he recognised her and that's like 6 years later and it completely threw him so I think it's had a massive effect on his mental health (pg 4, line 139)*. *the anxiety that it caused at the time and that hasn't left him (pg 7, line 241)* | | |

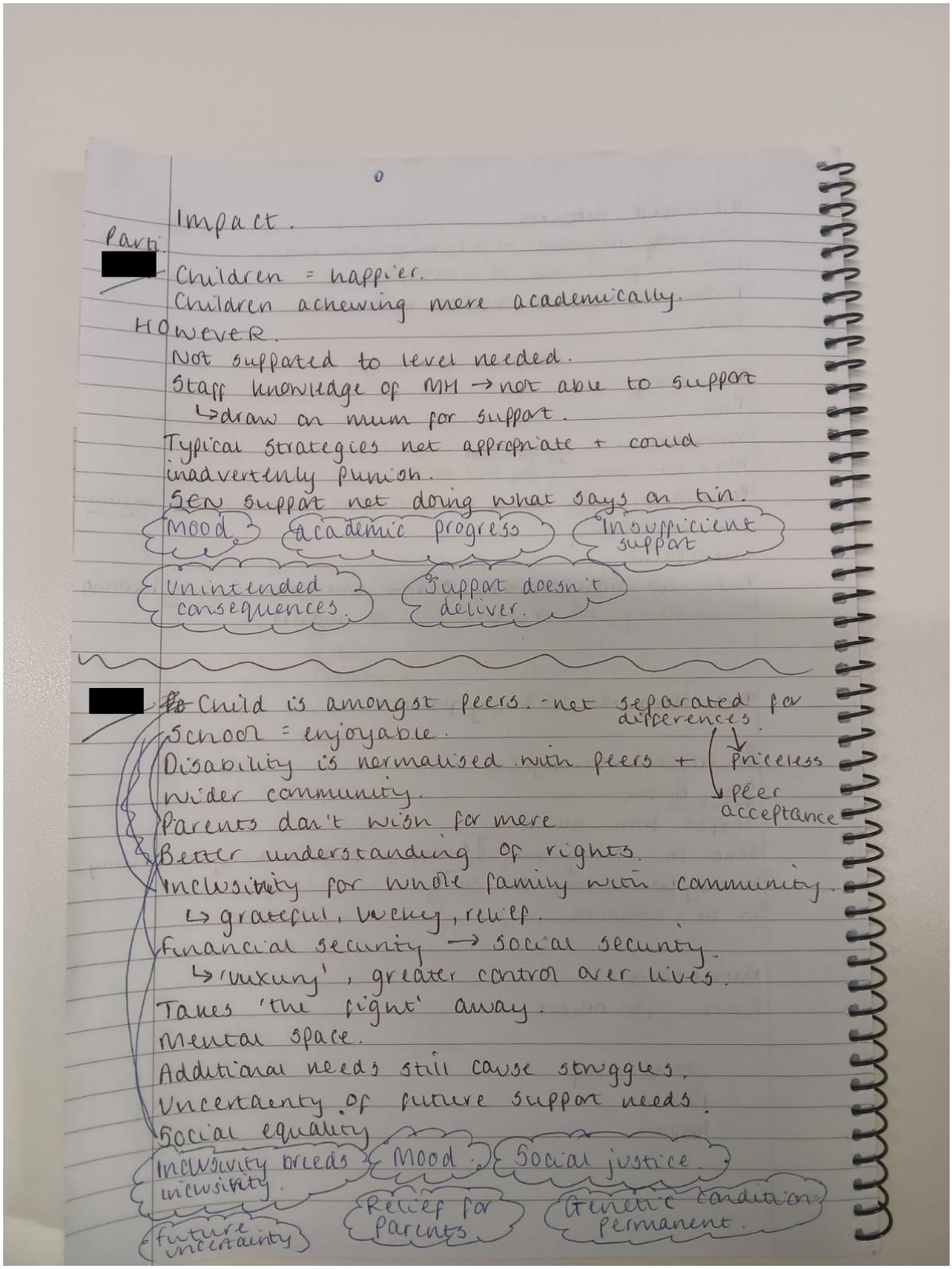
### Stage 5: Mapping and Interpretation

After all the data have been charted, the researcher uses their charts for the final stages of data interpretation. Ritchie and Spence (2002) comment that this stage is the hardest to describe in terms of the process that should be followed. Although they provide some examples as to how researchers might go about interpreting their data, I found the process outlined by Parkinson et al. (2016) particularly helpful for this stage.

I started by making notes about the charted information for the first 8 charted participants. Notes were made for each of the participants separately and with respect to a given chart. For example, notes were made for the first parent, 'Andrea', in respect to the information logged within the 'impact of support' chart, and then for the next parent, 'Emily' and so on. Notes were then made in the same order for the information logged in the 'peer relationships' chart and then the 'obtaining support' chart. Once the notes were made for a participant for a given chart, I summarised them into participant specific themes. **Figure 4.3** presents an example of participant specific notes and themes I created for Andrea and Emily in respect to their information logged under the 'impact of support' chart.

Next, I collected all the themes for each participant for a given chart/aim and grouped them in terms of common underlying meaning and topics. This process gave rise to my initial themes. Following this I noted the charted information for the next 5 charted participants and again summarised their notes into participant specific themes. I then incorporated these themes into my initial themes and made necessary adjustments to them when needed (e.g., renamed them). I then repeated this process for the remaining 4 charted participants. Once this final stage had been completed, I had identified my final themes and subthemes.

*Figure 4.3 Participant specific notes and themes created for 'Andrea' and 'Emily' for information logged under the 'impact of support' chart*



## 4.5 Results

### Sample

17 mothers were recruited into the study. Three of the mothers who took part had two children with the genetic condition and so information was provided for 20 children in total.

**Table 4.4** presents the characteristics of the overall sample and **Table 4.5** presents the characteristics of each participant individually.

*Table 4.4 Sample characteristics.*

| | Total N (17) | % |
|---|---|---|
| Mother's characteristics: | | |
| *Age* | 17 | |
| <20 | 0 | 0.0% |
| 20-29 | 1 | 5.9% |
| 30-39 | 4 | 23.5% |
| 40+ | 12 | 70.6% |
| *Sex* | 17 | |
| Male | 0 | 0.0% |
| Female | 17 | 100.0% |
| *Ethnicity* | 17 | |
| White | 17 | 100.0% |
| Minority ethnic | 0 | 0.0% |
| *IMD score* | 14 | |
| 1st quintile | 4 | 28.6% |
| 2nd quintile | 3 | 21.4% |
| 3rd quintile | 3 | 21.4% |
| 4th quintile | 3 | 21.4% |
| 5th quintile | 1 | 7.1% |
| | Total N (20) | % |
| Child's characteristics: | | |
| *Sex* | 20 | |
| Male | 13 | 65.0% |
| Female | 7 | 35.0% |
| *Ethnicity* | 20 | |
| White | 18 | 90.0% |
| Minority ethnic | 2 | 10.0% |
| *Inheritance status* | 18 | |
| Inherited | 8 | 44.4% |
| De novo | 10 | 55.6% |
| *Level of support* | 20 | |
| Mainstream school with EHCP* | 7 | 35.0% |
| Mainstream school without EHCP | 7 | 35.0% |
| Special school | 6 | 30.0% |
| *Stage of school* | 20 | |
| Nursery | 1 | 5.0% |
| Primary | 14 | 70.0% |
| Secondary | 5 | 25.0% |
| *Child likes school* | 20 | |
| Yes | 15 | 75.0% |
| No | 5 | 25.0% |

*Abbreviations: IMD score = Index of Multiple Deprivation score*
**includes one child who attended a mainstream independent school.*

*Table 4.5 Individual characteristics of mothers who took part in an interview and characteristics of their child/children*

| Mother | | | | Child | | | | | | |
|---|---|---|---|---|---|---|---|---|---|---|
| Participant | Age | Ethnicity | IMD score | Sex | Ethnicity | Inheritance status | School stage | School type | EHCP status | Likes school |
| Andrea* | 40+ | White European | 1 | F | Black British | De novo | Primary | Mainstream | No EHCP | Yes |
| | | | | F | Black British | De novo | Primary | Mainstream | No EHCP | No |
| Emily | 30-39 | White American | NA | M | White American | De novo | Nursery | Mainstream | EHCP ◊ | Yes |
| Isla | 40+ | White Mixed | 3 | M | White Mixed | De novo | Primary | Special | EHCP | Yes |
| Kayleigh | 40+ | White British | 4 | F | White British | De novo | Primary | Special | EHCP | Yes |
| Kylie | 30-39 | White British | 1 | F | White British | Inherited | Primary | Mainstream | No EHCP | Yes |
| Kirstie | 40+ | White British | 1 | M | White Welsh | De novo | Secondary | Special | Unknown | Yes |
| Lily | 40+ | White British | 4 | M | White British | Inherited | Secondary | Mainstream | No EHCP | Yes |
| Leah | 40+ | White British | Unknown | M | White British | De novo | Primary | Mainstream | EHCP ◊ | Yes |
| Lydia | 40+ | White British | 2 | F | White British | Unknown | Secondary | Mainstream | EHCP | No |
| Maureen | 20-29 | White British | 2 | M | White British | Unknown | Primary | Mainstream | EHCP | Yes |
| Megan | 40+ | White Irish | Unknown | M | White Irish | De novo | Secondary | Mainstream | No EHCP | Yes |
| Sarah* | 40+ | White British | 3 | F | White British | Inherited | Primary | Mainstream | No EHCP | No |
| | | | | M | White British | Inherited | Primary | Mainstream | No EHCP | No |
| Shelly | 40+ | White British | 5 | M | White British | De novo | Secondary | Special | EHCP | Yes |
| Thea* | 30-39 | White British | 3 | F | White British | Inherited | Primary | Special | EHCP | Yes |
| | | | | M | White British | Inherited | Primary | Special | EHCP | No |
| Tessa | 40+ | White British | 4 | M | White British | De novo | Primary | Independent | EHCP | Yes |
| Violet | 30-39 | White British | 1 | M | White British | Inherited | Primary | Mainstream | EHCP | Yes |
| Zoe | 40+ | White British | 2 | M | White British | Inherited | Primary | Mainstream | EHCP | Yes |

*Abbreviations: IMD score = Index of Multiple Deprivation score (IMD score for 'Emily' = NA because family did not reside in the UK).*
** Mothers who have two children with the genetic condition.*
*◊ Country equivalent of an EHCP (e.g., Individual Education Plan (IEP) in Ireland).*

## Themes

Three main themes were identified and broken down by subthemes.

1. A fight against ‘the system’
    a) Opponents and allies along the way
    b) The long and taxing road to victory
    c) To be continued…
2. Shining a light on social inequalities
    a) Intergenerational cycles of disadvantage
    b) Discrimination against disability
    c) Gender inequality
3. The benefits and limitations of support
    a) *“if he’s given the time he can achieve”*
    b) Improved social experiences
    c) Remaining issues

### Theme 1: A fight against ‘the system’

‘The system’ refers to the SEN provision service. ‘The system’ was explicitly referenced by 10 mothers and was deemed to be *“broken”*. Mothers described their experience as a *“fight”,* and used synonymous terms such as *“a battle”* and survivor talk.

> *“I’ve sort of gone through there and come out of the other side” (Lily)*

#### *Subtheme 1: Opponents and allies along the way*

Opponents along the way represented the various obstacles mothers had to overcome during their effort to obtain support.

Firstly, there were rules and requirements parents seemingly had to adhere to, but which did not reflect proper SEN guidance. For example, parents were told their child

was not *"needy"* enough compared to other children to receive support, or that their child needed an ASD diagnosis to be deemed eligible for an EHCP. Some of these rules could indicate misunderstandings of the SEN support system by parents or the professionals working within it. However, in some cases it seemed the school put these requirements in place to increase parents' chances of being ultimately successful in securing support because they knew that even if children needed it, parents would be likely be unsuccessful without proving additional certain markers of need. This was because the SEN system was under resourced, both in terms of money and personnel, and was under increasing pressure *"to save money"*.

> *"we got that autism diag[nosis] cause the school kept saying well we can't apply for the EHCP until we've got an official thing because although we'd got his diagnosis of [genetic condition] they didn't think that would be sufficient enough" (Violet)*

Some obstacles were deemed less well intentioned, however, with some parents reporting they had been purposely misled about what support was available, and that information had been intentionally withheld by LAs to protect resources.

> *"they told me they [children] had to display behaviours to be able to qualify to go there [a school]. [I've] since found out that's not quite right but they're limited on numbers, so they try and put people off" (Kirstie)*

The quote by Violet above also signifies the limited knowledge associated with these genetic conditions. This 'opponent' was noted by several parents. Lily was told by her child's school that they did not *"know enough*" about his genetic condition to provide support. This led to her child's paediatrician meeting with the school and diagnosing him with additional conditions which were more known to them.

*"the paediatrician was like 'well he has a diagnosis of the chromosome deletion, that is a diagnosis, you need to support his needs because that is a neurodevelopment issue' and they [the school] said 'well we don't know enough about that. If he had a diagnosis of say ADHD or ASD-', so he [the paediatrician] said 'okay then, I'll diagnose him right now. He's got ADHD and ASD' (Lily)*

After that meeting, the school "*bucked their ideas up"* according to Lily. However, Lily had previously told her child's paediatrician she did not want to label her child with additional diagnoses at that point in time, indicating that her wishes had needed to be disregarded for her child to receive the support he required. Lily's story also provides a good example of how having an ally onboard can have a considerable impact on parents' success in securing support.

To highlight the lack of knowledge of rare chromosomal conditions, some mothers compared the awareness of these conditions to Down Syndrome (DS), a more common chromosomal condition. They felt it would be beneficial for the gains being made by DS advocates to be applied to those with rarer conditions given their similarities in need.

*"the Down Syndrome Bill going through parliament at the moment…Unique and people like that have been saying well actually it should be a 'Rare Diseases Bill'. It's not just about Down Syndrome, all the things that are going in that bill are equally applicable to children who have another chromosomal difference" (Tessa)*

Whilst knowledge of genetic conditions was particularly low, knowledge about neurodevelopmental conditions in general was low amongst some professionals, including teachers, clinicians and local authority (LA) staff. Some professionals were cited as having an *"old fashioned"* views and therefore limited understanding of neurodevelopmental challenges (e.g., children who could make eye contact with others could not have ASD). Yet, their knowledge was at odds with the expertise some seemingly perceived to have, which led to the dismissal of the opinions of parents, and other professional experts. This was a particular point of frustration for parents.

*"I'm saying to the school 'I cannot get her to school, like she will not come, she's refusing'. They're like 'it's an attendance problem, she's got attachment issues with you, you mollycoddle her, you wrap her up in cotton wool' and all of this stuff so I'm like 'no, no, no, I can see that there's something else, but I can't put my finger on it' (Lydia)*

*"[child's paediatrician] put it in his statement that he's probably going to need assessing for ADHD. I mentioned this to his actual class teacher and his TA…'oh well he's not ADHD, I know what ADHD is, he's definitely not got ADHD'. Within two weeks of being within special school and seeing the paediatrician he was medicated for ADHD…it just shows that they have no idea what it can look like" (Isla)*

As suggested by Lily's story further above, support from allies could make a considerable difference to parents' success in securing educational support. Allies came in the form of educational psychologists, speech and language therapists, occupational therapists, lawyers, Members of Parliament (MPs), child commissioners, charities, other parents and more. Support often came from a child's school or nursery too. According to Maureen, she would not have come as far as she had without the support of her child's school, who had been a *"backbone"*, fighting her corner *"every step of the way"*. Violet's account of securing support was notable for the inclusive language she used ('we' versus 'I') and indicated the partnership between her and the school when applying for support. Her story also credited her SENCO as taking a leading and coordinating role throughout the EHCP process and providing guidance to her and her partner, which was seemed to be a rare experience.

*"we went straight to the school and said 'right this is the diagnosis. What do we do now?', and then she [the SENCO] got straight onto it [applying for an EHCP]" (Violet)*

However, support from allies was not always impactful. In Andrea's case, she had received backing from her children's private tutor but to no avail.

> *"she [children's tutor] told me that, "Andrea, trust me that is not normal". She came with me to the school, their tutor for two years, to tell them the significant problems they have in maths and they [the school] still don't want to draw a conclusion and discuss anything at all" (Andrea)*

*Subtheme 2: The long and taxing road to victory*

The road to victory was not short and spanned several years in some cases. This was in part related to some of the issues already noted. For example, it took Violet two years to obtain the ASD diagnosis that facilitated her child's EHCP application.

Parents' battles were sometimes won at tribunal, with a judge ruling in the family's favour. In some cases, it was won just by threatening to take the LA to tribunal, or after the LA conceded in the run up to the tribunal date. LAs were reported to sometimes overturn their initial decision to deny support at the last minute. This was regarded as an LA tactic to purposefully delay having to grant support.

> *"it went right up to the very, right at the day before the court case, and then they [the LA] turned round and conceded…the solicitor actually says that's what they do…'it won't go to court because I guarantee you like somewhere in that week before they will give you what you want'…they have no hope in hell of winning the case, they just keep it up to the very last minute" (Leah)*

Additional LA delaying *"tactics"* included drafting *"useless"* EHCPs which then needed to be redrafted, and LAs not sticking to statutory time limits to complete actions. LAs were also accused of tricking parents with *"lip service"* to appease them, but then providing no subsequent action.

*“I feel like local authorities sort of almost give lip service to it. ‘Oh, we’ll invite the parents in for a way forward meeting’ and then nothing happens so the parents like feel like they're getting somewhere and then they're just using it as another delaying tactic” (Tessa)*

Participants also recognised delays caused by more genuine reasons such as extra demands placed on public services during the COVID-19 pandemic. They also recognised the lack of resources within the system which meant staff within schools and LAs were overworked and could not implement the support they are likely morally aligned with.

*“Nobody goes into like special education or early education because they like hate kids so it's just gotta be like very disheartening and frustrating for them [staff] to not be able to execute what they know that these children [need]” (Emily)*

The demands on staff were having a negative impact on their wellbeing. Staff were reported as burnt out and, in some cases, taking sickness leave.

*“they’re completely overworked…my last caseworker who I actually thought was brilliant…she said that she had something like 150 live cases…well you know that’s two days a year basically on one child and that’s nowhere near enough…so I understand it’s stressful on them and I also understand that because of the processes with parents, parents then lose patience and they get the rough end of the stick…so yeah I can totally understand why the staff turnover’s high” (Tessa)*

Visibility of need could also influence the length of time it took for support to be obtained. For example, masking behaviour in children impacted the school’s ability to recognise and therefore support children’s needs. Masking was explicitly noted by two parents but, others reported a similar behaviour without labelling it as such.

*"[Child] is not going to start crying in the maths lesson…but you know how she says it to me, 'mummy I was crying in my head'. She hides it but she said that she cries in her head hard" (Andrea)*

Some parents reported that it had taken some time for them to come to terms with their child's disability. For example, Thea, who had two children with the genetic condition, noted that she had started the process of applying for support *"much earlier"* for her youngest child compared to her eldest, for whom she had gone through a process of acceptance first. Accepting children's disability was particularly difficult when mothers were repeatedly told by others, including professionals, that their child did not have an underlying need or diagnosis, and *"made to believe"* their child's difficulties were *"all in my head".*

*"people started saying well you know 'why do you need a diagnosis? Why do you need to carry on finding out what's wrong…maybe it's just [child], maybe you should just learn to deal with it' and when you've got teachers saying that to you as well and the SENCO you start thinking well maybe, maybe that's the way that I need to start thinking" (Shelly)*

One parent shared that she was even completing a college course on SEN to reassure herself that she was *"not being led like sideways"* by the school who were telling her that her children *"have absolutely no problem".* This was contradictory to the opinion of the children's speech and language therapist (SALT). The need to arm herself with knowledge is indicative of the level of distrust noted between some parents and schools.

The journey to eventual 'victory' was taxing. The expectation on mothers throughout the process often resembled that of a professional job, attending meetings, filling out paperwork, answering emails and background research. On top of this, mothers were juggling and coping with additional professional and personal stressors, including the challenges associated with their child being unsupported.

*"I almost had an emotional breakdown…it was dreadful. At the time I was working full time…it was quite a stressful job as it was. I was coming home to emails from school 'oh this has happened, that's happened'…then my grandma died as well so I was just, I'd got to the point where I just couldn't take anymore" (Lily)*

Lily was not the only mother to report that the fight had such an impact on her mental health. Others had been prescribed medication to treat their associated stress, anxiety and low mood.

When parents were successful in securing support, they were validated. The long process also revealed to them personal qualities that they had not appreciated about themselves before (e.g., their determination and patience). One mother was also diagnosed with ADHD after she was successful in obtaining an ADHD assessment for her son.

*Subtheme 3: To be continued…*

The fight was not always over after support had been formally secured. The legally binding nature of EHCPs assured parents that their child would get the support needed, however some reported that plans were only as good as what was written in them. If they were sparse or loose in content then it was easier for the school and LAs to *"interpret"* them in ways that best suited them, which was not always in alignment with what parents expected would be implemented.

In some instances, the school could not physically provide the support that had been agreed. Isla stated that the mainstream school her child had first attended was *"never set out to deal with"* children who had the additional support needs her child had. Kayleigh's child's school came to the same conclusion, but this meant that Kayleigh had to go through the process of amending her child's EHCP so that she could attend a special school which had the capability to support her. However, this was met with LA resistance.

There were some incidences where schools had reported to parents that certain support, was being provided when it had not been. This had included administering

medication to children and had prompted parents to make formal complaints the school's governors board.

> *"There were things recording as having happened with [child] that I then established actually hadn't happened, so I made a formal complaint to the governors after he left" (Shelly)*

Limited knowledge about how to best support children was also implicated. Andrea shared that her children's school did not know how to provide psychological support and therefore would turn to her for answers. Whilst parents appreciated joint problem solving, Andrea felt that in this case she had given them all her ideas already and that she was not *"an expert"*, but *"just a mother"*. In other cases, teachers were completely unaware of a child's needs as detailed in their plan.

> *"he had a really bad start with a teacher in primary school…he literally was crying everyday going into school and we went to talk to the teacher and she had him at the back of the room in the corner…I said you know 'what about his report and his [genetic condition]?...and she said 'what report?' and I said 'you never read the report?'" (Megan)*

Additionally, parents felt that they were still battling against the general attitude of, and language used by teachers regarding their child, which were reflective of the traditional approach that society has toward how education should be conducted, and a lack of appropriate teacher training which provides a good understanding about the learning needs of *"neurodiverse"* children.

> *"we live in a neurodiverse world…but I feel like teaching is still on the 'you sit down, you be quiet, you do the work', like he's never going to be like that…when he had his end of term report last year at the end of reception they put something like '[child] shows great potential in sports if only he'd listen', well he's not making a choice not to listen, he's in a sports hall, it's echoey, he suffers with sensory processing, he's trying to cope with all the noise and stay good but his cup is*

*overfilling at this point…it's almost like an accusation that he's being deliberately obtuse and I find that a lot…teachers need to do a stint with children with additional needs to fully understand as part of their overall training" (Tessa)*

Parents found it difficult to listen to what they felt was constant criticism of their child, and to be regarded as *"a problem parent"* when they challenged the school about their language and inadequate implementation of their child's SEN support.

The permanence of genetic conditions and other neurodevelopmental conditions mean that additional support could be required for the entirety of a child's time at school and parents expected future battles similar to what they had already experienced.

*"you've got years of this to come yet, [child] will be eligible for an EHCP till he's twenty-five. He's only seven now and we've already had to have a monumental fight…we've got years of this to go yet and then I guess we'll be fighting adult social care next after" (Tessa)*

Indeed, Shelly, whose child had an EHCP and was nearing the end of secondary school, was experiencing some issues with obtaining the most suitable college placement for her child and was in the midst of prepping to *"battle my heart out".* Maintaining the level of support currently received could also be a challenge during children's annual EHCP reviews.

Finally, parents reflected that the rarity of genetic conditions often means that what the future holds for their family is uncertain. Mothers who had good relationships with their child's school seemed to have more trust that they would manage their needs in the most appropriate way. Their preparations for a fight were not obvious.

*“they are such partners with us…we just don't know what to expect from him [child] as time goes on you know…but because they’re such a good district and because they partner with us so strongly and they are so innovative, I feel like we might really have a good chance of finding some sort of accommodation…we'll have to wait and see” (Emily)*

## Theme 2: Shining a light on social inequalities

### *Subtheme 1: Intergenerational cycles of disadvantage*

The need to fight for the limited resources within the system meant that some mothers might feel pitted against each other in accessing the limited resources that are available.

*“I know that we should all share and care but actually you have to fight like rocket” (Kayleigh)*

Yet some families had the social capital which enabled them to enter the fight from a stronger position compared to others. Several mothers noted their “*slight perks”* which had helped them access support. Tessa noted that not all families had the financial resources to employ legal aid when taking their LA to tribunal as her family had done, and Lydia commented that not all families could finance a private educational psychologist to assess their child as her family had. Violet reported working in a hospital gave her increased opportunity to get her child assessed for ASD which had been instrumental in her obtaining an EHCP. Lily, Kylie, Tessa and Maureen all had previous professional experience which they were able to utilise. Emily had also moved to an area where the houses and property taxes were more expensive to access the better resources which were subsequently available to her child.

Considering the fight for support was difficult despite their noted advantages, several mothers shared their concern for children whose family did not have access to such

resources and the social inequalities the current system helps to perpetuate.

> *"it worries me cause I used to work in a community in quite a deprived area…I see children there that have obviously got emerging needs and the parents are going to need to start fighting and it really worries me. It's like what if they haven't got any family to back them up? What if they've got their own issues and they're not able to do the fight? It's like that generation of children that just get lost isn't it and their life has you know, it's not gonna be good is it? It's not setting them up in the early days" (Kirstie)*

Yet despite fighting for the same resources, the support parents provided each other to access better educational provision was evident. Parents helped each other to complete applications forms, and online community support groups had been created with the aim to help parents apply for SEN support. However, parents needed the knowledge that these groups existed in order to access them, which was not always the case.

> *"you just have to go on Facebook. There's like groups galore where parents will literally come on and give you a video tutoring on how to apply for it [an EHCP]. There's so much stuff out there that parents don't realise" (Lydia)*

The strength of community between parents was clear just by reflecting on the motivation some of the mothers had for taking part in this study.

> *"the reason I do it [take part] for is because I hope it will help somebody else…sometimes when you read somebody else's story you think 'gosh', it resonates with you and you think 'okay, I'm on the right path. I might not be right all the time, but actually other people are going through this' and I think that helps cause even though we are all so connected we're not connected if you know what I mean…and I think when you have a child who has a disability you're even less connected because there's lots of people that don't understand. Until you've been there yourself, you don't know" (Kayleigh)*

*Subtheme 2: Discrimination against disability*

Several parents stated that the denial of support their child was legally entitled to was discriminatory. Children were attending school unsupported, which had consequent impacts on their ability to engage with learning which in turn, stifled their academic progress. Children's attendance was impacted, with children either refusing to attend school or parents pulling them out of school for their protection. Some children were delayed in starting school because the support parents felt was needed had not been provided. Parents argued that their child was being denied their right to education, yet this was not being addressed with a sense of urgency.

Maureen posited that there is a societal complacency working against children with disabilities. Society is prejudiced against what people with disabilities can achieve which consequently denies them the same opportunities their peers have to achieve their potential.

> *"it's society that holds him back cause people have this outlook on, okay he has got disabilities and he does struggle, but people can make it easier for him and I think the system needs to be updated in some way to help them children because they are kind of left behind and they do need that help because there's nothing saying they can't reach that potential they just need more help than say the same child that's normal I suppose" (Maureen)*

*Subtheme 3: Gender inequality*

Lastly, the social inequalities between men and women gender roles were highlighted. Several mothers shared that they were more heavily involved with and impacted by the process of obtaining their child's support compared to their male partners.

*“I feel like the pressure is ever so much on the woman…because men don't really understand it as much as the woman does. My partner…he's brilliant with her…but he will do all the laughy jokey stuff and the fun dad stuff…I deal with the serious stuff like the meetings and the appointments and all of the paperwork and all of the hard work getting her to school...it's very much left to me and I was frustrated…the weight was on my shoulders…and it nearly broke me and my partner up because I was angry at him that I was having to do all of it and then he was thinking ‘well what can I do? I have to go to work?’” (Lydia)*

Even for those who did not explicitly refer to the difference in their and their partner’s role, it was implied. Mothers often attended school meetings seemingly alone. In cases where one parent had stopped working, changed job, or had gone to part-time hours, it was always the mother in the partnership who was reported to have done so. They also often told their stories in the first person singular versus first person plural or third person (i.e., ‘I’ vs ‘we’, ‘he/she’ or ‘they’).

It should be noted that in Kylie’s case, she believed that her partner lacked confidence to make suggestions or necessary decisions regarding their child’s support as he felt his knowledge was comparatively less than Kylie’s, who had experience of working in a school. Additionally, his employer was less flexible than Kylie’s in letting him attend school meetings and he either had to take the time as annual leave or unpaid leave, which was not financially viable. Being less able to contribute led him to feel like *“a bit of an odd part”*.

## Theme 3: The benefits and limitations of support

### *Subtheme 1: “if he’s given the time, he can achieve”*

Academic progress was notable when children were better supported. Children showed improvements in specific core skills such and writing and language, and more generally within academic subjects.

*“his English was about two years behind at that time because he’d not had the support he needed to cope with it all. He’s now caught up with his English and he’s about a year and half ahead on his maths” (Lily)*

Children who had attended school over the pandemic were noted to progress well during this time because of the smaller class sizes and increased ability of teachers to work with children more closely. Some children were also noted to be happier within the smaller class.

*“they [the school] classed them [children] as vulnerable children and they were able to access education, they could go to school, so it was very clear that in a small group setting they were much happier and they achieved much better academically as well” (Andrea)*

Parents were also happy to notice an increase in their child’s confidence in light of their academic achievements, for example, more frequently speaking during class and answering questions. However, not all children had caught up with their peers fully and some parents therefore felt their child’s potential still had not quite been met in the way it may have if earlier intervention had been provided. Furthermore, one parent noted that some of the improvements their children had made was a result of the support which they had done with them at home, or for which they had privately paid. Another said that the improvements her child had made were down to his own character and determination, indicating she did not hold the support provided by her child’s school in high regard. Therefore, although parents were happy to observe progress, not all progress was attributable to the school.

*“it [academic progress] isn’t in my belief anything to do with the school and more to do with [child] being so determined. He wants to learn, he wants to know things, he’s very intrigued by things, he's inquisitive, he’s asking questions, he’s learning himself…he pushes himself” (Zoe)*

*Subtheme 2: Improved social experiences*

Increased support also helped to improve children's social experiences at school. Children who had moved from a mainstream school to a special school were amongst children who were more like them, and therefore were more included amongst their peers and within school activities (e.g., school plays), and able to *"fit in".*

> *"when they used to do things like that at mainstream schooling…he wouldn't really join in with the dancing and the fun so he was standing there sucking his fingers just watching everything, taking everything in…he did stand out from the crowd as being somebody that wasn't fitting in with his peers whereas now he blends in*" *(Shelly)*

Mothers also reported that their child was less teased in special school compared to in mainstream for differences between them and their peers (e.g., for not being able to do work others could do, for physical features associated with their genetic condition), which relieved mothers of worry and upset associated with the increased risk of bullying they perceived their child to be at.

The differences in bullying incidences between mainstream and special schools suggested that mainstream schools are not able to (or were choosing not to) provide truly inclusive environments for all children. Indeed, some approaches taken by mainstream schools to manage bullying excluded the child who had been bullied from participating in school activities other pupils had the opportunity to experience, and therefore punished the victim versus the bullies.

*"What they [the school] did with her is they removed her out of her class at the end of year four because of the bullying and they put her in a cupboard with a TA [teaching assistant] and another little girl and there she stayed until year six. That's no intervention whatsoever, is it? You know and I feel like that really impacted her…she was the one hidden away and I feel like that's really frustrating for her because she thinks well "what have I done wrong?", "Why am I the one being punished?" cause that's essentially what it is, it's a punishment, you're being removed from the main school. She wasn't allowed to take part in any of the year six production…she didn't go on any of the year six leaver trips, she's not in any of their photos, so all of the stuff they did for year six leaving she didn't participate in" (Lydia)*

Mothers were sometimes internally conflicted when trying to assess how to personally manage the bullying their child had experienced by those who the child considered to be their 'friends'. Whilst mothers did not want their child to be *"anywhere near"* other children who were unkind to them, they also did not want to *"punish"* them by not allowing them to interact with their 'friends'.

The school's approach to supporting children who were being bullied was important. Some parents excused other children for some of the negative behaviours directed toward their child because *"they're children, they don't know any better".* However, schools who took approaches like the above, whose approaches did not model acceptance towards children with SEN, and whose approaches did not raise awareness of disabilities, were regarded as helping to reinforce non-inclusivity. This was not only harmful to their child, but was regarded as disadvantageous to the other pupils, who would no doubt encounter and perhaps need to support others with disabilities in their later lives. Conversely, schools with zero tolerance for bullying and who disciplined bullies whilst protecting the anonymity of the child being bullied were reported to reduce the incidences of bullying and increase children's confidence that they were not going to be targeted by their peers. In instances where children were bullied again by the same child, mothers still felt confident that their school would support them with the issue well.

Unsatisfactory support with bullying was not confined to in person incidences at school, but also cyberbullying. Megan had been told by her child's school that they would not "*deal with"* incidences of cyberbullying perpetrated against her child by other pupils at the school *"unless it's impacting on his teaching and learning".* Yet the incidences she described were serious. Other children were using online platforms to make light of the recent suicide of her child's friend and posting subtle messages which made associations between her own child and suicidal thoughts and actions. Though apparent to Megan, the subtlety of the posts meant that her child did not always understand the meaning behind other children's messages, that they were being unkind to him, or the extent to which they were.

Limited understanding about the intent behind other children's actions and why other children would be unkind was a common theme throughout the interviews and made children vulnerable to being led to do things by others (e.g., hitting or spitting on other children), especially when they strived to be liked and accepted, and sometimes resulted in the child getting into trouble. As noted earlier, in some cases children even regarded those who were bullying them as their 'friends'. Some mothers worried that their child did not understand the concept of bullying and therefore would not be able to report incidences to an adult or would mistakenly report they had been bullied when they had not.

When children did have insight, the consequences of bullying included children becoming upset or showing reluctance when having to go to school, unwillingness to ask for help during lessons for fear of attention and teasing, difficulties sleeping, bed-wetting, dislike of lessons in which they were at particular risk (physical education (PE)), all of which signal the importance of effective anti-bullying support at schools.

Mothers also reported that their child did not have the communication skills to effectively defend themselves, thus signalling the need for improved social support. Children's limited ability to defend themselves and their lack of insight into the situations in which they might need to defend themselves sometimes acted as a signal to parents and teachers that a child could have an underlying SEN.

*“she [child] struggled with relationships with her peers a lot and that's how we noticed and recognised the issues because she had a skipping rope tied around her neck by two girls and they tightened it and knotted it, it wasn't like it was just wrapped around, they physically knotted it around her throat...but she allowed them to do it which was really odd” (Sarah)*

Two mothers reported that their child had bullied or been aggressive towards other children because of difficulties with sharing, communication and *“identifying right from wrong”*. Therefore, improved social support could see bullying behaviour towards other children minimised too.

Support with bullying did not only come via approaches taken by the school, but also from children’s various protectors at school. Several parents noted their child had other children looking out for them. This was particularly effective when those children were popular amongst their peers. Children were also protected by their siblings at school or by personal contacts of their parents (e.g., friends or family members who worked at the school). The presence of TAs also helped reduce instances of bullying. This seemed to be a comfort to parents who could not be at the school to support their child themselves.

Mothers did what they could to help their child socially (e.g., keep in touch with friends who had moved away, explained their child’s behaviour to other children who did not understand, provide the school with guidance and aides to minimise the risk of bullying), however their influence was limited given they could only help from afar.

*“she’s [child] got oromotor issues…so when she is eating like pasta or spaghetti or whatever she’s always got sauce around her face, so I specifically said to the school “please, you know other children don’t come home with yogurt or whatever on their face. I’ll provide the wet wipes, please, even if she can just wipe her own face, please help her to”, because you know when you’re in a mainstream school the children they say it to you straight, they don’t like think about how you might feel, so I was trying to help” (Kayleigh)*

Additional support helped relieve some children of anxiety and distress associated with school which meant they no longer displayed alienating coping behaviours (e.g., hiding underneath tables, *"lashing out"* at other pupils). Other pupils perceived them as less 'different' and children were better able to engage with their peers which helped them to foster friendships. This was the opposite for children for whom the support they should have received was not implemented properly. For example, Isla described the social impacts of her child not being allowed to use the continence products that had been provided to the school for him to use.

> *"there was no continence products so then children even at 4 and 5 years old will be not wanting to be near him because he's wet through constantly" (Isla)*

Improved relations were also noted between children and their mothers as children's behaviour improved with additional support received. Social support also improved children's resilience in coping with later instances of bullying compared to how they had been able to cope with earlier incidences.

Again, as mothers were not physically at school with their child, their child's social experiences were assessed via information from teachers, other children in their child's school, and their child. For example, children might come home and ask their mother to explain the behaviour of other children as it was not always obvious to them, which signalled to mothers that they were being teased.

> *"one time she came home and she said 'mummy the children are calling me weird. Why are they calling me weird?" (Kayleigh)*

However, information from their children could be limited and it was sometimes hard for mothers to assess what they were being told. For example, Maureen shared that her child typically only told her about his day when he was overwhelmed with emotions at home. She described she therefore had to get him to calm down before she could get a proper grip of what had occurred and *"what's actually just in his head"*.

Some mothers noted their child was happier to talk about their school day once support was put in place, and this helped them feel more included in their child's schooling and suggested to them that there was less negativity associated with school for them.

Social experiences were also assessed by their child's engagement with their peers outside of school hours (e.g., playdates, birthday parties and after school clubs) which again, appeared to increase with support. Children's improved social experiences also improved mothers' social networks as they were more able to get to know the parents of their child's friends.

> *"he got invited to a birthday party…it's the first birthday invite he's had in probably three or four years…so I stayed so I could get to know some of the parents cause obviously I've not sort of got to know anybody because we didn't go to the primary schools they went to so that was nice to get to know some parents as well" (Lily)*

The emotional impact children's social experiences had on mothers themselves was evident throughout the interviews. Lily reported she had been "emotional" when her child told her he had a best friend and had been invited to another child's birthday party. I also noticed that Leah had sometimes teared up when talking about her child's social experiences at school, which I interpreted to reflect her concern that her child might not be properly understood or accepted by his peers and her fear of potential bullying. Megan was seemingly more impacted by the cyberbullying experiences of her child than he was.

> *"it was just so upsetting and so stressful and your whole weekend where you should be safe at home and just doing what you have to do is now ruined by this and you spend a few days going over that in your head… it just takes away your peace" (Megan)*

Mothers also had a worry that the threat of bullying would always linger, even if was not a current problem, and that the risk would increase as they got older and the

differences between them and their peers became more apparent. Increased social difficulties were noted by Zoe who had older children with SEN.

> *"just from experience as a mum of other kids with special needs, year 4 now is the year when the maturity kicks in and they kind of end up being left behind…that gap widens between them and their peers so much. More socially than anything else, and this is when they kind of stop being invited to parties and the kids start going off in their smaller groups" (Zoe)*

### *Subtheme 3: Remaining issues*

Despite the social and academic improvements that came with increased support, additional provision did not solve all problems. Parents who were pleased with their child's support still noted behaviours at home which signalled to them that they were ongoing struggles at school (e.g., behavioural issues, reluctance to get ready for school in the morning, school refusal and difficulty getting to sleep). Some parents noted this was because their child was *"always gonna be someone that struggles with education".* Remaining challenges children faced compared to their peers made them feel *"sad"*, *"different"* and *"stupid".*

Others noted that their child's behaviour at home stemmed from being pushed to their potential, something the school were better able to do now given their better understanding of children's limits.

*"they [the school] do push the children. They know what the children can achieve. She finds it very difficult and then we do have some behavioural issues where she might act out or she'll talk back like she shouldn't… she needs one of us to lay with her to go to sleep and she has to talk to you about nonsense for half an hour before she goes to sleep and you need to keep saying 'its bedtime, bedtime, bedtime' and 'you have to go to sleep'…I think it's her way of like making sense of the day. I don't always understand why she has to just like say the same things over and over but it's like a calming thing for her and I do think even though she's happy at school, actually its taxing" (Kayleigh)*

The need for additional support at school did not go unnoticed by children's peers and could lead to teasing, which meant that it was better that support be provided by teachers 'in secret'.

*"because of his fine motor skills…when he'd be eating he sometimes gets quite messy…so we sent him in his bag with some baby wipes…on a number of occasions we heard this couple of lads come out and say "oh, there's [child] the baby"…it took ages for me to work out why they were saying it…I was asking him and then he started saying "I don't want to take these [wipes] to school anymore mammy, I don't need them"…so we mentioned it to the teaching assistant and she says "right I know, I'm gonna get wipes that are just basic no 'baby' or anything written on them and then if he does get into a point where he does get a bit messy that he needs a bit of a clean then he knows he can just go to my drawer and get them" (Violet)*

Support was not able to undo the lasting psychological impacts of previous negative experiences that had occurred whilst children were unsupported, which were sometimes reflective of trauma.

*"when he started the new school it was all going really well and then all of sudden he started to get really stressed, really anxious, totally out of character and we realised that there was a support worker that had started there that had on a few occasions worked as like a bank support worker in this old school and he recognised her and that's like 6 years later and it completely threw him so I think it's had a massive effect on his mental health" (Kirstie)*

Isla also reported that her child now had extreme anxiety around using public toilets which had stemmed from her child's previous school attempting to toilet train him, despite being incontinent and against the instruction of both her and the occupational therapists who had assessed him and provided guidance to the school as to how they should support him. This had impacted the activities they could do as a family away from the home and raised concerns about potential physical health implications of her child not using the toilet when needed.

Whilst support increased the opportunities for inclusivity, the point made above that some children will *"always"* struggle with education relates to an earlier point made by Tessa and Maureen: that education today does not cater for the learning needs of children with SEN as well as it does for their peers. This viewpoint calls into question whether current school systems are providing the truly inclusive environments of which they should aim for.

The benefits of inclusive practices when they were implemented were praised by parents.

*"what was so exceptional about this district is that because they have this funding they have the ability to have a paraprofessional with our children in a general education classroom so our paraprofessional helps our son navigate the classroom and the activities so that he is doing these activities alongside his peers and it's just very normalised for him to be in the classroom" (Emily)*

Emily also noted that the other children in her son's class were understanding and accepting of her son's behaviour which had the potential to *"frustrate"* them (e.g., shouting and pulling on other children's face masks). His peers showed kindness and patience towards him, and he was extremely popular amongst them. Other mothers also noted that the children who showed understanding toward their child often had personal understanding of SEN from having a family member (e.g., a sibling, a cousin) with a neurodevelopmental condition.

The inclusive nature of Emily's child's school was regarded as promoting inclusivity within the wider community which she could not value enough. Additional benefits included relief from the fight for support, additional brain space, which was not being taken up with worry, greater understanding of the support parents are entitled to and greater social equality. Emily noted that she was *"lucky"* that her son had the support he did as she knew that truly inclusive practices were rare. In fact, several mothers who reported that their child attended a supportive school reported they were lucky, indicating they were aware that there are differences between the quality of SEN support between schools.

Despite some schools' best efforts to identify and address needs, their limited capability to teach children amongst their classmates resulted in them being taken out of lessons for their additional supports and then *"plonked"* back into their own class where they were then expected to catch up with what their peers had learnt whilst they had been absent. Taking children out of lessons also limited their ability to experience the benefits of working with and learning from their typically developing peers and make close friends with the children in their own class. Additionally, if children's TAs were not available to them, they could 'miss out' on what their peers were doing at that moment in time. For example, Kayleigh's daughter had sometimes missed out on playing with the other children at breaktime when her TA was not immediately available to help her back into her shoes after changing out of her PE clothes.

The perceptions and exclusionary behaviour of other pupils' parents was an issue noted by some mothers, who felt that parents model the level of acceptance their own child will later show towards children with SEN.

*“the other parents as well don’t want to invite him to things because you know he’s the kid that’s still in nappies… his anxiety makes him not speak so he wouldn’t speak to the parents and they just saw him as being a strange child” (Isla)*

Lastly, working a fulltime job was still not possible for some mothers after support had been secured.

## 4.6 Discussion

This chapter presents the first known study to explore the qualitative experience of mothers of children with a genetic and a neurodevelopmental condition when trying to obtain SEN support, the impact of those experiences and the impact of support once secured. Importantly, this study focussed not just on the impacts on children but also those on the rest of their family, particularly their mother. This approach was taken after learning from the Cerebra Steering Group that too often the needs of children with SEN are considered separate from their family context, which can be counterproductive to fully understanding the experiences of this group. Therefore, it is hoped that these findings will not only further our understanding of children’s school experience but will also highlight important additional contextual factors that should be considered when future efforts to minimise negative experiences will hopefully be enacted.

### 4.6.1 Findings and implications

Findings indicate that it can be a significant challenge for parents to obtain the level of support deemed necessary for their child at school. This was reflective of the SEN review’s and HCEC’s findings (DfE, DHSC, 2022; HCEC, 2019). Their “fight” against a “broken” system highlighted the failings and unsustainability of the current SEN provision system. Parents navigating it were clearly frustrated by it and in some cases disadvantaged by it. Staff working within it were reportedly overworked and disheartened, and the children who should have been receiving support were often being neglected by it.

Proving that children required SEN support was difficult. Children sometimes masked their difficulties which made it more challenging for parents to get

professionals ‘on side’. Masking is used largely amongst autistic people to describe the actions they take to hide or ‘camouflage’ their autism. However, masking is likely common amongst individuals with neurodevelopmental conditions and traits in general (Pearson & Rose, 2021). Collaborative working between schools and parents help to improve children’s academic and behavioural outcomes (Cox, 2005), however when parents were not believed, untrusting relationships formed between them and their child’s school. Raising awareness about masking behaviour amongst educators may improve parent-school relationships and ultimately help to improve outcomes for children.

Staff also often held misconceptions about how neurodevelopmental traits and conditions could present in children, which also indicated a limited level of understanding about such conditions. Yet this was at odds with the seemingly high levels of confidence some school staff had to dismiss the concerns of not only mothers, but also the expert opinions of other professionals (e.g., educational psychologists, paediatricians). Teachers accumulate expertise from first-hand experience with and observation of children within the classroom and have been proven good (although not perfect) at identifying the existence of behavioural and emotional problems in children (Loades & Mastroyannopoulou, 2010). Indeed, several of the mothers in this study were grateful to their child’s school and nursery for highlighting potential SEN. Therefore, this point is not to disregard teachers’ professional knowledge, but to advocate for better utilisation of the knowledge of other experts too. The level of knowledge about rare genetic conditions specifically was even less. Thus, harnessing parents’ knowledge of their child’s difficulties is particularly important for this group, especially as neurodevelopmental conditions can present differently in children with genetic conditions (Niarchou et al., 2015). Additionally, although they are limited in number, awareness of quality information resources which do exist for educators should also be raised.

Once need was proven, support was still difficult to secure. The HCEC (2019) reported that schools are running on insufficient budgets which has diminished the quality of SEN support they can provide without the additional funding provided by an EHCP. Inadequate lower-level SEN support has contributed to the “magnetic” pull of EHCPs, yet to qualify for one, children must display more severe needs. There

was some evidence that schools could be advising parents to seek additional diagnoses to increase their chance of being successful in applying for an educational plan (e.g., an ASD diagnosis). Parents may wish to seek additional diagnoses for their child, however they should not be required to do so in order to secure SEN support when need is already apparent, as seemed to be the case for some of the children in this study. ASD diagnostic pathways in the UK are overwhelmed (British Medical Association, 2019) and children waiting for an assessment purely for the purpose of securing an EHCP are likely increasing demand for assessments which are already scarce.

The EHCP application process also involves significant administrative effort for schools and local authorities (HCEC, 2019), however, again, they often do not have the resources to cope with such administrative burden. This results in the delays at various stages of the process as noted by mothers here, staff burnout and high staff turnover, which subsequently compromises the efficiency of the system even further, as well as its sustainability. By enabling schools to provide better lower-level support, public services might be relieved of some of the pressures associated with fewer parents feeling the need to apply for an EHCP. Children might also receive support earlier, likely improving their outcomes (Ramey & Ramey, 1998) and improving the quality of life for parents who no longer need to engage in the fight to obtain support.

Parents who had secured SEN support were often still engaged in some kind of battle with services, or at least expected to be in the future. What was detailed in a child's EHCP did not always align with what their school could provide which gave parents false hope. In other cases, school staff were not always aware of the support that had been stipulated or how to provide it. These findings help partly explain why almost a quarter of parents of children with an ND CNV might be unsatisfied with the support provided by their child's school (Chapter 3) and substantiate some of the reasons provided in the previous chapter as to why parents did not believe their child's school had provided them with the right support. Continued difficulties were also associated with staff members' limited knowledge of neurodevelopmental conditions which led parents to feel their child was judged and, in some cases, unfairly punished by their teachers. Parents believed that teacher training should better prepare educators to support children with SEN by providing them with better

understanding of neurodevelopmental conditions and traits. Teacher training has previously been identified as an avenue to improve knowledge of need and support for children (Nash et al., 2016).

It was also noted that the education system within the UK adopts an old-fashioned way of teaching which does not, in practice, allow for the level of inclusivity so strongly advocated for within policy (DfE, DoH, 2015). The discrepancy between policy and practice could suggest that schools and staff do not have the capability, or want, to implement working practices which can offer true inclusion. The measures used by governments to evaluate the performance and quality of schools, namely publicly available pupils' examination and school inspection results, have been criticised because they deprioritise inclusionary practice (Shaw, 2017). Ofsted (2019) has recommended the DfE work more closely with teachers before policies are passed to ensure their aims reflect what can feasibly be implemented on the ground and that they do not contend with the instructions of potentially competing policies which already exist.

The findings of this study also provide qualitative evidence that the educational environment of children with ND CNVs may help to partly explain phenotypic variability observed within this group. Mothers reported marked social, academic and psychological benefits of SEN support, suggesting that the level of support received at school could have a significant impact on children's phenotype.

The stories of the mothers taking part in this study support parents' and teachers' suspicions that the system favours those with higher social capital (HCEC, 2019). Mothers noted their *"slight perks"* which had aided them in their struggle for support. I am not aware of a study to specifically investigate how SES influences access to SEN support in a larger and more representative UK sample, but those from less deprived backgrounds have been found to have better and more timely access other public services, for example, health services (Harris et al., 2011). If the SEN system is in fact inequitable, then procedures within the current system should be changed to make them more accessible to all parents. Otherwise, certain groups of children may be disadvantaged in experiencing the associated benefits of education.

The social advantages in accessing support noted in this study could also provide some context for the observation that children with inherited ND CNVs, who are more likely from deprived backgrounds (Wolstencroft et al., 2022) are less likely to have and EHCP (Chapter 3) compared to children with *de novo* conditions. However more research is needed to explore the links between SES, ND CNV inheritance status and SEN support before they can be confirmed (or discounted).

Findings also provide some of the first contextual evidence as to the potential reasons why high rates of psychopathology are found in mothers of children with ND CNVs (Baker et al., 2021; Niarchou et al., 2022). The stress associated with acquiring SEN support was significant and several mothers had been prescribed psychiatric medication over this period. Further investigation is needed to validate the experiences of the mothers taking part in this study within a larger sample, but removing the barriers to access within the current SEN system could see improved maternal mental health. Generally, the mothers taking part in this study made minimal reference to their partner's involvement in obtaining support. One mother indicated that her partner felt he was less knowledgeable and less able to take the needed time off work to attend school meeting, and therefore felt less confident to make significant contributions to securing support. These observations substantiate the points above about relative individual advantages and disadvantages in securing support, but also indicate that efforts could be made by services and employers to engage with fathers specifically and enable them to take a more active role in their child's support. This may help alleviate some pressure from mothers and also empower fathers.

The government have outlined the approaches they will take to address the failings within the SEN system (DfE, DHSC, 2023). Such measures include the enforcement of national standards to provide parents, schools and LAs with more guidance about the support that should be in place for a given child, improving professional training in SEN, utilisation of digital technology to make the EHCP application process easier to navigate, and improving experiences around transition periods. On paper, these measures address some of the issues identified by mothers taking part in this study and so will hopefully make a positive difference for children with ND CNVs and their family. However, a common theme throughout the interview was lack of funding for

the system, and if predictions are correct, this will remain a problem within the coming years despite the UK Government delivering ‘record investment’ for the SEN system. Sibieta (2022) predicted continued financial pressures for schools, and particularly for schools which rely more heavily on support staff (e.g., special schools), indicating that educators may continue to struggle to provide SEN support to pupils in need, and that children and their family will continue to experience negative associated impacts.

The consequences of an unfit system on children and their family are too great to be ignored. Beyond the more socially positioned argument discussed above which advocates for equality of opportunity for all children, the long-term potential national economic consequences of not supporting children early are detrimental. Children who are denied quality education are more likely to experience later health problems in adulthood (Raghupathi & Raghupathi, 2020) and are less likely to be in fulltime employment as adults (Mitra, 2011). Countries’ economic growth is reportedly ‘driven by increasing employment’ (HM Treasury, 2021). Therefore, the apparent obstacles parents of children with genetic conditions and SEN face in accessing support should be addressed by the UK Government, who prioritise the economic growth of the country (Prime Minister’s Office, 2023). The impact of the SEN support system on parents (e.g., poor mental health and difficulties remaining in fulltime employment) should be further cause for concern for the same reason.

Lastly, governments should be concerned that the consequences of current practice, as portrayed by mothers taking part in this study, seems to in some cases deny children with SEN their human right to quality education as stipulated within the 1998 Human Rights Act.

### 4.6.2 Limitations

Whilst representative samples and generalisability are not markers of quality in qualitative studies which adopt a constructionist research design, the insight gathered about the range of experiences in securing SEN support and its impacts would likely have had greater breadth if other demographics were better represented in my sample (e.g., fathers, parents from minority ethnic backgrounds). Similarly, the recruitment of children and teachers could have deepened my understanding about

the impact of support on children, the challenges facing schools in implementing support and parent-school relationships from an alternative perspective. Given that travel and social interaction restrictions associated with the pandemic are no longer in effect, the views of children and teachers could now be collected.

Some readers could question the acceptability of including *'Emily'*, a US based mother, in this study when all other participants were UK based. When Emily made contact to volunteer to take part, she shared at that point her experiences had been positive. It was decided it was important to learn about such experiences and that her account could be written up as a case study to illustrate, and learn from, good practice, albeit within a different SEN system. However, when analysing Emily's data, it was apparent that her positive experiences and the impacts of them mirrored the more negative experiences and impacts UK mothers were sharing (e.g., improved inclusivity, social equity and mothers' mental health and positive parent-school relationship). Furthermore, I learnt about the American SEN provision service and there were evident similarities between practice in the UK and US. Thus, her experiences actually reinforced the findings of the UK sample as opposed to altering them, and therefore her account was included within the main findings to strengthen the conclusions of the study.

The interviews were conducted with mothers online. Although this increased the geographical area from which I was able to recruit, online interviews could have impacted the level of rapport I was able to build with the mothers taking part. Comparing to my previous experiences of conducting interviews in person, I did not regard the online interview process to impact my ability to form good relationships with participants. However, as I did not collect information from mothers about how they experienced participating online, I cannot evaluate whether they felt unable to disclose events and feelings they otherwise might have if the interviews were conducted in person.

Although I had guidance from an experienced qualitative researcher throughout my analysis, I worked largely independently during all stages of data collection and analysis. Some of my interpretations may have benefitted from other researchers being more heavily involved in the analytic process, however as I approached the data from a constructionist standpoint, the subjectivity of my conclusions is not

necessarily a weakness of this study. Nevertheless, I recognise that another researcher could draw different conclusions than those presented here based on the same data collected.

Finally, although I had previous experience of conducting qualitative research, my prior experiences were predominantly in data collection and conducting interviews and so I did not consider myself to be highly experienced in analysing qualitative data. As noted already, I benefitted from the guidance of an experienced qualitative researcher, however as she was not a member of my main supervisory team, there were limits to her level of involvement. I attended qualitative data analysis courses to improve my understanding, skills and confidence in conducting thematic analysis.

## 4.7 Conclusion

This chapter aimed to enrich the findings of Chapters 2 & 3 by collecting in-depth accounts from mothers about their child's life at school. It also aimed to better understand how mothers experience the SEN support system and the subsequent impacts on their mental health.

Securing SEN support for children was reported to be challenging. Limited resources and lack of knowledge about SEN in general, but also these genetic conditions specifically, were cited as major challenges for parents to overcome. Whilst parents were 'fighting' for provision, their child was not supported to the level parents deemed necessary, which seemed to have negative implications for their social and academic functioning at school. Mothers' mental health was also negatively affected by their experience of securing SEN support.

Effective support was regarded as having transformative effects on children's academic progress, behaviour and psychological wellbeing. However, remaining issues were still noted and suggested that schools cannot (or do not) offer truly inclusive environments for children with SEN. When high levels of inclusion were facilitated, parents implied that they were happy with their child's experience of school and trusted the school to navigate future uncertainties.

This study has suggested that for the SEN support system to operate efficiently and for it to implement truly inclusive practices for children with ND CNVs, it needs to be

adequately equipped in terms of funding, personnel, and expertise. Additional factors could be important, however these were the main issues identified here.

In the next chapter I will discuss the findings and implications of this thesis as a whole. I also suggest topics for future research to address the methodological limitations of this work and to develop our understanding of the school experiences of children with ND CNVs beyond what has been presented.

# 5 General discussion

The overall aim of this thesis was to explore the school experiences of children with a ND CNV to better understand whether these experiences could help elucidate some of the unexplained phenotypic heterogeneity associated with these genetic variants. It also aimed to consider the impact of children's educational experiences on their parents, who have been found to be at increased risk for psychological problems.

This chapter will review the implications of the findings of this thesis as a whole, as well its methodological weaknesses, and future research directions. First, a reminder of the previous chapters' key findings is provided.

## 5.1 Overview of key findings

### Chapter 2

Children with ND CNVs were at increased risk for peer bullying compared to their unaffected siblings (52.4% vs 27.4% respectively; adjusted OR = 4.87, 95% CI 2.74 – 8.64, p = <.001). The presence of ADHD, indicative ASD and PE were predictive of bullying experiences, as were higher symptom counts of these 'conditions'. Increased neurodevelopmental burden and age were also positively associated with bullying experience. Children with a ND CNV did not seem to be at increased risk for bullying compared to their siblings for displaying neurodevelopmental characteristics associated with the neurodevelopmental conditions. Other factors such as physical appearance could provide a better explanation as to why bullying is more common in this group.

### Chapter 3

Almost 25% of parents who had a child with a ND CNV were dissatisfied with the support received by their child at school. Children with ADHD, ODD or CD were more likely to have dissatisfied parents. Parents of children with an EHCP were more likely to be satisfied. Children meeting criteria for indicative ASD, ID or who had a *de novo* ND CNV were more likely to have an EHCP.

Qualitative data provided insight into the circumstances in which parents were dissatisfied with their child's educational support. Unhelpful learning environments, difficulties accessing higher levels of statutory support, limited understanding of children's needs, limited resources, and inadequacy regarding the amount and type of support were implicated.

Chapter 4

Mothers of children with ND CNVs reported that they engage in a continuing 'fight' with public services to secure and maintain SEN support. Parents were fighting against an under resourced SEN provision system and limited knowledge about neurodevelopmental conditions and ND CNVs amongst professionals. Parents' social capital contributed to their success in obtaining support. Children who were supported at school showed improved social and academic experiences, however challenges remained when schools could not implement truly inclusive school environments.

## 5.2 Key findings and implications of this thesis

1. School can be a difficult place for children with a ND CNV.

Bullying was a common experience amongst children with a ND CNV. Furthermore, a considerable proportion of their parents were unhappy with the support received by their child at school. Chapter 4 highlighted the importance of SEN support for positive academic and social school experiences.

These findings are perhaps unsurprising when we consider the wider literature which reports that children with challenges associated with those observed in children with ND CNVs are at increased risk of bullying (Chapter 1). Equally, parents of children with neurodevelopmental conditions without a known genetic cause have been found to experience great difficulty in securing SEN support for their child (Chapter 1). Given this broader literature, it is reasonable to ask what the added utility is of specifically studying the experiences of children with ND CNVs.

Firstly, although this group presents with similar challenges to other children and thus needs similar support, the findings of Chapter 4 indicate that having a ND CNV

could act as a barrier to access. This was because genetic diagnoses were too poorly understood by professionals, indicating that general awareness of these conditions needs to be raised. Studying children with a ND CNV as a standalone group may increase the knowledge base about them and enable parents to direct professionals to information resources more easily, helping them to gain a better understanding about their support needs.

A better knowledge base could also be helpful to both clinicians and parents at the point of diagnosis. Clinicians who communicate a child's genetic diagnosis to parents are not always aware of the implications of the condition for the child and their family, and can feel uncomfortable when discussing them (Chapter 1). Parents can also be left uncertain about what the future will bring (Chapter 1). Better understanding about what later life can be like at school could improve both parents' and clinicians' experience of diagnostic appointments, and aide earlier identification of emerging difficulties for both professionals and parents.

Similarly, better knowledge could empower parents with increased awareness that might assist their 'fight' for SEN support for their child (Chapter 4) and better children's chances of being supported at the earliest opportunity. Of course, parents should not have to battle for the support their child is legally entitled to, but given the current challenges facing public services, it would seem naive to claim that this reality for families will change in the foreseeable future. This makes better knowledge amongst professionals about the struggles in accessing educational support for children even more important, as the allyship of those professionals can be instrumental in parents securing it (Chapter 4). Educators are an obvious group for which better awareness would be beneficial. However clinical professionals were found to have a great impact in this regard, and it could be helpful for clinicians to 'check in' with parents about their child's school experiences during routine appointments to identify those who might need their support. Good information for all professionals involved in children's care will hopefully encourage good multi-disciplinary working around the child and their family and again, help the chances of timely identification and assessment of need and provision of support (Colizzi et al., 2020).

Greater awareness about the risks of bullying in this group could also lead to greater vigilance to, and monitoring of, the signs of bullying by parents and educators which, again, may facilitate early identification and implementation of intervention and preventative measures. Charities, like Cerebra, have expertise in communicating research to the public and professionals and so it will be important to work with them to increase awareness.

The opportunity for early intervention is especially great for children with a ND CNV diagnosed at birth. However, to seize this opportunity, the knowledge base about what these conditions mean for children needs to be available. We will likely see many more newborns diagnosed with genetic conditions if the UK's genomics healthcare strategy (HM Government, 2020) is successfully implemented. Therefore, the number of children and families who will benefit from ND CNV specific research will likely increase over time.

2. Children's school experiences are variable and may contribute to the phenotypic heterogeneity observed amongst children with ND CNVs.

Children with a ND CNV have heterogeneous presentations. This is in part influenced by genetic factors (e.g., the impact of 'second hits' and additional common variants) (Chapter 1). However, their environment likely also contributes to the pattern of problems children develop and to what extent children are impaired.

The findings of this thesis indicate that not all children experience school in the same way. According to their caregiver, not all children were bullied, and the majority of children were adequately supported by their school. Given the existing evidence which highlights the marked influence early peer relationships and limited access to education has on a person's outcomes (Chapter 1), the finding that *some* children have negative experiences in these two domains suggests that children's experiences at school might influence the phenotypic heterogeneity of this group.

Both Chapter 2 & 3 presented cross sectional findings and so it is important to note that neither direction of effect nor causality can be inferred between the school experiences studied here and children's outcomes. Nonetheless, it is at least plausible that in children with ND CNVs, bullying at school and lack of support is causally related to their poorer outcomes compared to those who are not targeted by

their peers and supported adequately. The findings here provide a basis for future investigation into these topics. Longitudinal studies whereby data are prospectively collected at a series of different time points will be needed to help determine causal effects.

Should harmful environmental experiences influence the problems children develop, protective experiences should also mitigate against negative consequences, further highlighting the need for early interventions. Research which measures the outcomes of children who have been supported compared to those who have not been will also be needed. Given experiences were not consistent between children, approaches which target those most vulnerable to harmful experiences could be more effective versus approaches which take a blanket approach.

3. Children's school experiences seem to impact the mental health of their mothers.

The semi-structured interviews I conducted with mothers presented in Chapter 4 highlighted some of the implications of children's school experiences on mothers themselves and provided some context to empirical data which indicate mothers of children with a ND CNV are at risk for psychological adversity (Baker et al., 2021; Niarchou et al., 2022).

Mothers reported that their mental health had suffered during the process of trying to obtain SEN support for their child; a process associated with overwhelming levels of stress. Mothers were also emotionally impacted by their child's school relationships, sometimes seemingly even more so than their child.

The motivation of mothers to secure support was to ensure that the educational rights of their child were met, that they were happy at school and that they were being set up with the best chances for their future lives. It was evident that their child was their upmost priority and therefore potential measures which aim to address mothers' mental health, but which do not address their child's underlying needs will likely not be sufficient in improving their psychological wellbeing.

Following the negative conclusions of the recent SEN review, the UK Government outlined the steps they will take to improve the process of applying for SEN to make

the experience more positive for parents (explained further in Chapters 3 & 4). It will take time to determine whether these measures are effective. However, children with a ND CNV will often need support beyond education from other public services too (e.g., mental health services) which are also under resourced and struggling to keep up with demand (British Medical Association, 2019). Future research should explore pathways to other support services to delineate barriers and facilitators as it would be unsurprising to find parents are navigating high levels of stress when trying to access other sources of support too.

### 5.3 Methodological issues and future work

As noted within individual chapters, power is an issue for some of my analyses. The rarity of known ND CNVs within the population means that recruiting samples as large as other cohort studies studying general populations is difficult. Whilst the ECHO, IMAGINE and DiGEN studies collectively offer one of the largest cohort studies of individuals with a ND CNV, findings and conclusions should still be taken cautiously. Data continue to be collected from families of children with a ND CNV by ongoing IMAGINE-2 and DiGEN studies which will allow future work to investigate whether the findings of this thesis are replicated in a larger sample. There could also be future opportunity to link data with health and education records (e.g., the National Pupil Database) to enrich samples.

Analyses were cross-sectional which restricts the inferences that can be made about the causes and outcomes of bullying and access to SEN provision. Longitudinal studies are needed to help determine causal effects between variables. The IMAGINE-2 study is revisiting children who took part in IMAGINE-1 and therefore could facilitate longitudinal investigation of this thesis' aims in the future.

This thesis presents the school experiences of children with a ND CNV as reported by their caregiver only (primarily their mother). The initial research design of Chapter 2 & 4 sought to hear from teachers and children, however, due to challenges posed by COVID-19 restrictions (which have been expanded on in more detail within the respective chapters), I had to alter my research design accordingly. Our knowledge about the school experiences of children and the challenges associated with obtaining SEN support could be improved if future work sought to hear from

additional informants. Given the reported difference between mothers and fathers involvement in securing SEN support for their children, it would be particularly interesting to hear from fathers, and compare how their perceptions about their child's experiences compares to not only their child's but also the child's mother.

### 5.4 Future work

In addition to the work that should be conducted to address the limitations of this thesis as noted above (e.g., longitudinal analysis), additional areas of future work have been identified.

Firstly, school experiences were chosen as the specific environmental factor of interest for this thesis because school represents such a major part in all children's lives and has known impacts on an individual's later life outcomes. The project was in part inspired by a conversation Professor Marianne van den Bree had with someone diagnosed with 22q11 DS and their mother about their experiences at school and the hypothesised consequence of this period on their mental health. Furthermore, members of the Cerebra Steering Group and parents taking part in the research conducted by the Rare Genetic Research Group at Cardiff University had frequently shared insights into the challenges associated with their child's schooling. Therefore, school experiences seemed an important issue to investigate. However, there are many more environmental exposures that could impact the outcomes of children with a ND CNV, for example, their family environment. Additionally, there are many other school related experiences which could be explored.

This thesis did not explore the experiences of children with individual ND CNVs but instead grouped children with different ND CNVs together to study their experiences collectively. When sample sizes permit, it would be useful to differentiate children by their specific ND CNV, as has been done in other studies (Bozhilova et al., 2023; Chawner et al., 2019), in order to clarify whether exposure to school experiences is associated with the same outcomes regardless of the specific ND CNV carried.

I am particularly interested in the social factors which impact access to support, and I would be interested to explore whether some of the factors identified in Chapter 4 (e.g., socioeconomic status) reflect the experiences of a larger sample using quantitative methods. Although this has been suggested elsewhere (Wolstencroft et

al., 2022), I am not aware of a study which has investigated this topic specifically. The differences between access to educational support between children with a *de novo* and inherited ND CNV were also of interest, and I would like to investigate this observation further, as well as any additional differences between these two groups.

I was also interested to hear about the varied level of insight children had into bullying as a concept and their confusion about the motivations of their peers' behaviour, which appeared to their mothers to be deliberately malicious in nature. Given that intent to cause harm is needed for a behaviour to qualify as bullying (Olweus, 1993), I would be interested in exploring whether the level of insight children have into the intentions of their peers influences the impact of these experiences. Are children who have less awareness about the intentions of others less impacted by the malicious acts perpetrated towards them compared to children who have more insight? The relative impact of bullying by siblings was also not explicitly considered in this work but would be of interest in future.

Lastly, better understanding of the environmental and additional genetic influences on the outcomes of children with a ND CNV will facilitate future research into the gene-environment interactions between such variables and how these relationships impact phenotype.

## 5.5 Conclusion

The findings of this thesis contribute to our understanding of environmental impacts on the phenotype of individuals with a ND CNV. They also suggest that investigation into children's school experiences, a previously understudied topic in this group, is worth pursuing.

Some children with ND CNVs may benefit from increased social and SEN support at school to help them cope with the potentially harmful consequences of peer bullying and to help them access their school's curriculum. Professionals should be made aware of these issues as this could help to ensure the needed support is implemented at the earliest opportunity and provide children with the best opportunities to reach their potential. This could also help to relieve mothers of significant levels of stress which might see their risk for mental health adversity decrease somewhat. If the findings of this work and the causal relationships

proposed are confirmed in studies which address the methodological issues noted above, its reach will become more widely felt as genomic medicine becomes more embedded into healthcare practice and the rate of children diagnosed with a ND CNV increases.

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