

Prenatal Screening for Down's Syndrome: Parent and Healthcare Practitioner Experiences

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Abstract

This article reviews research on both parent and healthcare practitioner experiences of prenatal screening for Down's syndrome. Whilst studies on Down's syndrome screening are broad in scope and diverge in theoretical and epistemological foundations, their core focus can be classified into two main and intertwining categories: 1) the decision-making practices of expectant parents and why they do/do not participate in screening; 2) the interactions between parents and practitioners, particularly the discrepancies of knowledge and the pursuit of "informed choice" and "non-directive care." To conclude, I highlight current knowledge gaps and the areas of substantial interest for future critical sociological engagements.

Introduction

It is almost one-hundred-and-fifty years since John Langdon Down, an English physician, first described a condition which was later defined as "mongolism." In 1961, however, the condition was renamed "Down's syndrome" (or Trisomy 21) after a prestigious group of biomedical researchers signed a letter objecting to the "embarrassing term" of mongolism (Allen et al. 1961, 426). Down's syndrome is one of

the most common chromosomal conditions in the world, affecting approximately one to two of every 1000 live births in England and Wales alone (Morris and Springett 2013).¹ People with Down's syndrome are likely to have a range of symptoms including learning difficulties, shortened limbs, reduced muscle tone, restricted physical growth, a flat profile of the face, and a large protruding tongue (NHS FASP 2012). However, the condition is frequently identified as compatible with life, that is, as "not lethal" (Ivry 2009, 192). This translates to people with Down's syndrome being likely to survive childbirth and enjoying a good quality of life, although symptoms and prognosis vary significantly in each respective case. Indeed, the main feature of Down's syndrome is the inconsistency of its manifestation. However, due to medical advances and better knowledge regarding treatment and care, children with the condition can survive beyond sixty years today (CARIS 2012).

In the past fifty years, Down's syndrome has occupied a central position in reproductive politics, particularly in Western healthcare systems. Prenatal screening for the condition has become a universal programme, coming into "routine use, becoming embedded in, we might say, a social matrix" (Cowan 1994, 36). Whilst predicted advances of genetic screening generally may be more modest than initially expected, the range of available screening techniques for Down's syndrome (and other conditions) has steadily expanded.² In 2011, roughly 74% (N=542,312) of all expectant parents in England and Wales – or rather those accessing NHS services – opted to be screened for Down's syndrome (NHS FASP 2012). The uptake in prenatal screening has dramatically increased annually in England and Wales since 2007 (53%). Whilst 2008 and 2009 saw uptake rates of 57% and 62% respectively, an uptake rate of 70% was recorded in 2010 (NHS FASP 2012). This differs from the Netherlands where uptake rates for Down's syndrome screening vary from 38% to 86% (van den Berg et al. 2005a) and Japan

where uptake rates are less than 2% (Nishiyama et al. 2013). The increase in England and Wales uptake rates may be attributable to the recent increase in maternal age (an increase in maternal age is the only known attribute increasing the chance of an unborn baby being diagnosed with Down's syndrome), the risk of miscarriage decreasing on account of increasingly proficient screening technologies (Budds et al. 2013; NHS FASP 2012; ONS 2011), and service supply factors since all mothers-to-be (rather than just mothers-to-be aged thirty-five and above as in earlier years) are now offered screening for the condition. The lower uptake rates in Japan, in contrast, are perhaps attributable to a lack of information on prenatal diagnosis and abortion not being legally permitted for "foetal abnormalities" (Nishiyama et al. 2013, 796).

In addition, a report conducted by the National Down's Syndrome Cytogenetic Register (NDSCR) claims in 2011, there were 1,873 diagnoses of the condition, 65% of which were made prenatally (N=1,211), in England and Wales (Morris and Springett 2013). According to the report, of the 1,211 prenatal diagnoses, 89% (N=931) were terminated, 8% (N=87) were live births, and 3% (N=33) were natural miscarriages or stillbirths (the outcome of 160 prenatal diagnoses is unknown). The proportion of terminations following a diagnosis of the condition in England and Wales has remained steady for over twenty years. From the first report in 1989 until 2011, the annual rates for termination after a diagnosis of Down's syndrome have ranged from 88% to 94% (the mean rate is 91%). This corresponds to the work of Boyd et al. (2008) who claim that ten out of eighteen European countries have an average termination rate of 88% after a prenatal diagnosis of Down's syndrome. Similarly, in certain areas of Australia and select US states, the termination rates following a diagnosis are reported as 95% (Collins et al. 2008) and 74% (Natoli et al. 2012) respectively. However, it is crucial to remember that many countries do not track termination rates for Down syndrome on a

national scale. This is likely to be related to local variations in laws, with termination being illegal in countries such as Northern Ireland (McNeill et al. 2009). Nonetheless, it is clear how Down's syndrome screening has been centrally located in reproductive politics and how it has subsequently been the subject of much academic attention.

I begin this article by providing a brief context of Down's syndrome screening and testing. In what follows, I identify sociological studies, inevitably sharing overlaps with other disciplines, largely focused on two core and interrelated aspects of screening for Down's syndrome: 1) the decision-making process of expectant parents and why they do or do not participate in screening; 2) the interactions between parents and practitioners, particularly regarding discrepancies in understandings and how these are negotiated, managed, and rectified by each party. This latter focus is most frequently directed at the level of the intertwining rhetoric of "informed choice" and "non-directive care." Whilst studies have examined the social significance of *diagnostic* testing for Down's syndrome (Browner and Preloran 1999; Bryant et al. 2006; Crang-Svalenius et al. 1998; Rapp 2000) or used screening/diagnostic testing interchangeably (Green and Statham 1996; Jaques et al. 2004; Kaiser et al. 2004; Marteau 1995; Press and Browner 1997), I focus exclusively on screening not least because of restricted word space and, more importantly, because screening is important enough to be lifted out of broad debates around universal screening/testing programmes. To reiterate, a screening test does not produce a diagnosis but rather provides expectant parents with information about the *chance* of their baby having a condition. In contrast, a diagnostic test can establish the presence or absence of a condition in a child, although it should be noted that diagnostic tests are not 100% accurate and are susceptible to human error (Austin et al. 2013).

The studies cited around Down's syndrome screening derive from different countries (and thus diverse social, cultural, economic, political, and medical contexts), use both qualitative and quantitative methods,³ focus on aspects of Down's syndrome screening at different periods of a pregnancy (pre-screening, mid-screening, post-screening), and stem from scholarly roots including sociology, public health, medicine (midwifery, nursing, genetics), anthropology, psychology and bioethics. The reader therefore needs to place this review within this diverse context. I conclude by highlighting knowledge gaps and future directions of sociological research. Thus, this article is not a systematic review in the narrow sense but rather highlights current work in the field, both qualitative and quantitative, and what knowledge gaps subsequently persist.

Prenatal screening for Down's syndrome: a context

Prenatal screening for Down's syndrome results in expectant parents receiving a "risk factor," a numerical probability establishing the odds of an unborn baby having the condition. The characteristics used for creating this risk factor are locally variable and a variety of risk factors can be produced including a background risk (based on age, ethnicity, pregnancy history, weight, gestation, smoking habits, the number of unborn babies, and a nuchal translucency measurement⁴, among others), a biochemistry risk (based on the measurements of biochemical markers), an adjusted risk (based on combining a background risk and biochemistry risk), and an ultrasound risk (based on the nuchal translucency size alone). As such, different probabilities are possible for the same pregnancy which might affect the higher-risk categorisation. The risk factors provided to expectant parents vary between hospitals. In NHS hospitals in Wales, for example, expectant parents usually only receive the adjusted risk factor for Down's

syndrome (in Wales, screening for Down's syndrome via ultrasound scanning is only available at privately-funded institutions).

After screening, expectant parents are categorised as "lower-risk" or "higher-risk." In NHS hospitals, for example, the recommended national cut-off level for this categorisation is 1:150 (a 1 in 150 "risk" of having a baby with Down's syndrome). However, this varies between countries and depends on political/economic factors together with whether national screening policies are established (Boyd et al. 2008). According to Boyd et al., for example, diagnostic testing is routinely offered to expectant mothers aged thirty-five and above in Germany and Spain independent of a risk factor. Taking England and Wales once more as an example, if expectant parents receive a risk factor numerically higher than 1:150 (e.g. 1:300), they are "lower-risk" and not offered diagnostic testing (amniocentesis or chorionic villus sampling/CVS). In contrast, if they receive a risk factor numerically lower than 1:150 (e.g. 1:50), they are "higher-risk" and diagnostic testing can be arranged to prove or refute a suspected diagnosis. For Gross and Shuval (2008, 550), this reflects a shift in medicine "from the actual to the potential." It should be clarified that each screening method is considered as having a "false-positive rate," meaning expectant parents receive a positive (higher-risk) result but do not have an unborn baby with Down's syndrome. The false-positive rate is crucial (as well as controversial) since it indicates the number of unaffected pregnancies subjected to diagnostic tests which have a one to two percent chance of causing a miscarriage.

If expectant parents decide against diagnostic testing, no further screening or testing for Down's syndrome is typically offered. If expectant parents consent to testing and a diagnosis is established, counselling is offered by a healthcare practitioner – who this is and from what speciality is locally specific – before a decision must be made

about continuing or terminating a pregnancy. Around three to five percent of pregnant women consenting to screening receive a higher-risk result (NHS FASP 2012) and according to Buckley and Buckley (2008), between 1 in 20 and 1 in 30 higher-risk (screen-positive) results detect an unborn baby with Down's syndrome. Uptake rates for diagnostic testing following a higher risk result for Down's could not be identified, nor could statistics about the number of foetal losses after a diagnostic test be located. The main objective of prenatal screening is to identify women in whom a risk factor is deemed high enough to warrant offering diagnostic testing. More information on, and the socio-histories of, screening/testing for Down's syndrome, as well as the condition itself, are offered elsewhere (Cowan 1994; Nancollas 2012; Rapp 2000; Selikowitz 2008; Wright 2011).

Existing insights

From here, I review research on both parent and healthcare practitioner experiences of prenatal screening for Down's syndrome. Taken together, these studies recognise how screening is a social as well as medical intervention (Armstrong 2012; Armstrong and Eborall 2012; Boardman 2010; Buchbinder and Timmermans 2013; Cowan 2008; Cox and McKellin 1999; Faulkner 2012; Gillespie 2012; Hogarth et al. 2012; Kehr 2012; Lock et al. 2007). Whilst several studies have explored parent and practitioner experiences of prenatal screening for Down's syndrome, this has predominantly focused on two core concerns: 1) the decision-making processes of expectant parents; 2) the interactions between parents and practitioners. In what follows, I turn my attention to these studies and their contribution to the field.

Decision-making process of expectant parents

The development and diffusion of prenatal screening techniques has triggered critical debates around the seemingly contradictory aspects of offering reproductive choice to – and prompting social, legal, and ethical dilemmas for – expectant parents. As such, there has been a proliferation of studies focusing on reproductive practices more generally and how expectant parents must make serious life decisions frequently on the basis of partial knowledge (Ettorre 2000; Franklin and Roberts 2006; Glover 2006; Parens and Asch 2000; Rapp 2000; Rothman 1986; Rothschild 2005; Schwennesen et al. 2010; Thompson 2005). In relation to Down’s syndrome screening, much of the literature centres on the decision-making processes of expectant parents and how they justify certain choices. Common justifications for consenting to screening include, but are not limited to, offering reassurance about an unborn baby being unlikely to have Down’s syndrome (García et al. 2008; McNeill et al. 2009; Santalahti et al. 1998), satisfying curiosity about an unborn baby (Skirton and Barr 2007; van den Berg et al. 2005a), fulfilling demands of a partner (Jaques et al. 2004), and a fear of supporting and parenting a child with a disability (Chiang et al. 2006; Pilnick and Zayts 2012; Remennick 2006). This trepidation or “tentativeness” (Rothman 1986) was most often felt by expectant mothers who shouldered a moral duty to prevent what is expected to be the burden of a disabled child on their family (Chiang et al. 2006; García et al. 2011; Gross 2010; Ivry 2006; Remennick 2006). A similar fear is reported by expectant parents with previous pregnancy experiences and/or a family history of chromosomal conditions, citing this when accounting for their decision to have screening (McNeill et al. 2009; Spencer 2002). One of the most common justifications for participating in screening, however, is that expectant mothers perceive themselves as being at an “advanced maternal age”, translating to women aged thirty-five and above, since this is the only known attribute increasing the risk of an unborn baby being diagnosed with

Down's syndrome (Kaiser et al. 2004; McNeill et al. 2009). It should be noted that decision-making processes are immensely complex and expectant parents are likely to account for their decision with reference to several of these explanations. Indeed, expectant parents are frequently conflicted about whether *to know* or *not to know* (Aune and Möller 2012; Liamputtong et al. 2003).

Several of these studies appear to understand consenting to Down's syndrome screening as a result of rational decision-making processes. However, others identify how Down's syndrome screening is an instance of conformity rather than an expression of choice (Chiang et al. 2006; Gottfreðsdóttir et al. 2009b; Heyman et al. 2006; Markens et al. 1999; Marteau 1995; Pilnick 2004; Pilnick et al. 2004; Press and Browner 1997; Remennick 2006; Santalahti et al. 1998; Skirton and Barr 2007; Sooben 2010; Williams et al. 2005). The title of an article by Gottfreðsdóttir et al. (2009a) – “this is just what you do when you are pregnant” – seems apt for highlighting this conformity and routine nature of screening. Hunt et al. (2005) likewise claim that expectant parents in their study did not have a clear understanding of the nature of screening since they viewed it as a standard and recommended part of routine care. For Gottfreðsdóttir et al. (2009b), this interpretation of Down's syndrome screening as a “normal part of the surveillance of ordinary pregnancies” (Vassy 2006, 2042) reflects how some expectant parents do not discuss screening and testing prior to consenting to a procedure. The routine nature of screening, according to other research, is also borne out of practitioners seemingly endorsing its acceptance (Heyman et al. 2006; McNeill et al. 2009; Remennick 2006) and how ultrasound scans can be viewed, first and foremost, as offering a chance for meeting the baby, and for making a pregnancy seem more real, rather than for detecting potential medical concerns (Draper 2002; Gammeltoft and Nyugen 2007; Heyman et al. 2006; Lupton 2013; Mitchell and Georges 1998; Williams et al. 2005). In a similar vein,

Tsouroufli (2011) claims that expectant parents opt for screening as a routine aspect of antenatal care because of the prompt processing of expectant parents in the hospital, because practitioners endorse it as a safe test (no chance of a miscarriage), and because practitioners expect that expectant parents will opt for the procedure.

For the likes of Baillie et al. (2000) and Åhman et al. (2010), the naturalisation of Down's syndrome screening as a "normal" part of pregnancy means expectant parents are not always aware of, or prepared for, the complex information and heavy choices associated with a screening result. Many studies, indeed, have reported how screening prompts feelings of fear and anxiety among expectant parents before, during, and/or after receiving a lower-risk or higher-risk result for Down's syndrome (Aune and Möller 2012; Burton-Jeangros et al. 2013; Green and Statham 1996; Ivry 2006; Markens et al. 1999; Marteau 1995; Pilnick et al. 2004; Remennick 2006; Williams et al. 2005). For Heyman et al. (2006) and Hunt et al. (2005), this angst, sadness, and grief frequently emerge following a higher-risk result and a decision needing to be made about diagnostic testing. In their study of doctors and pregnant women, Burton-Jeangros et al. (2013) employ Giddens' (1991: 70) conception of "manufactured uncertainty" to identify how tensions exist between the pursuit of control by surrendering to medical surveillance processes, the permanence of uncertainty in life processes, and the limitations of probabilistic data (i.e. the difficulty of interpreting a "risk factor"). Strategies of managing and negotiating this higher-risk result include developing interpretations via metaphors (Burton-Jeangros et al. 2013) or remaining emotionally detached from an unborn baby in the event of decisions needing to be made around a termination of pregnancy (Remennick 2006; Williams et al. 2005). This work shows that whilst expectant parents may engage with screening to abate anxiety and receive reassurance, they can simultaneously become anxious about waiting for results, be

confronted with distressing decisions about diagnostic testing, and can be apprehensive and fearful even after a lower-risk result (Burton-Jeangros et al. 2013; Marteau 1995; Pilnick et al. 2004).

Studies have also reported on why expectant parents do not take up prenatal screening for Down's syndrome. Justifications include avoiding the adverse health risks associated with potential diagnostic testing such as causing a miscarriage (Liamputtong et al. 2003; Markens et al. 1999; Pilnick et al. 2004), the inaccuracies and unreliability of procedures (Gottfreðsdóttir et al. 2009a; van den Berg et al. 2008), a lack of information on procedures being provided (García et al. 2008; Gross and Shuval 2008), concerns among expectant parents regarding the emotional impact of intervention (Markens et al. 1999; McNeill et al. 2009; van den Berg 2005a), information not being provided to certain ethnic and socio-economic groups (Dormandy et al. 2005), a fear and mistrust of medicine and its values (Gross and Shuval 2008), the complexity and inconclusiveness of risk information (Heyman et al. 2006; Markens et al. 1999), expectant parents ruling out the possibility of diagnostic testing or terminating a pregnancy (McNeill et al. 2009; van den Berg et al. 2005a), and personal and/or religious values about the worth of children with Down's syndrome and disabilities more generally (Gottfreðsdóttir et al. 2009a; Liamputtong et al. 2003; Remennick 2006). Once more, these studies identify how decision-making processes are complex, fluid, and intricate, with expectant parents likely to justify not screening for Down's syndrome with reference to any number of these rationalisations.

Interactions between parents and practitioners

Research on Down's syndrome screening, as well as concentrating largely on decision-making processes of expectant parents, also explores their interactions with healthcare

practitioners. Previous studies identify how Down's syndrome screening is carried out by midwives (Dormandy et al. 2006; Ekelin and Crang-Svalenius 2004; Samwill 2002), doctors (Burton-Jeangros et al. 2013; Driscoll et al. 2009; Marteau et al. 1993), or midwives and doctors interchangeably (Pilnick 2004; Smith et al. 1994; Williams et al. 2002a) rather than genetic counsellors and clinical geneticists. Notwithstanding their specialism, practitioners are often discussed in relation to "informed choice" and how practitioners are expected to provide "non-directive care," each translating to tendering medically-accurate information to expectant parents detached from any personal biases (Schwennesen and Koch 2012; Sooben 2010; van den Berg 2005b). Notions of choice and autonomy are at the heart of current discourses in Western healthcare systems (Williams et al. 2002a), placing an emphasis on looking and listening to "grant patients their life as well as knowing them as if they were dead" (Mol and Law 2004, 44). In this context, reproductive technologies are heralded as a route to liberation since they offer expectant parents information about, and control over, their offspring (García et al. 2008; Seavilleklein 2009). However, others show that this is not always empowering (Lippman 1994; Pilnick 2008; Williams et al. 2002b).

Several studies capture how expectant parents do not perceive their care as non-directive; the stating of options, in turn, does not always amount to the neutral provision of advice since some options have the force of an instruction (Browner et al. 1996; Hunt et al. 2005; Lippman 1991; Pilnick 2008; Williams et al. 2002b). Indeed, some have censured practitioners for providing biased information, their conduct being viewed as coercive and directive rather than passive and facilitating (Marteau et al. 1993; Tsouroufli 2011). Since knowledge is reproduced during interactions, the likes of Gothard (2011, 68) suggest that what we regard as a choice is never a value-free activity and is always made "within the context of social, cultural and community influences,"

few of which are simple or transparent. Whilst some are sympathetic for practitioners who struggle with balancing professional and private values (Anderson 1999; Farsides et al. 2004; Williams et al. 2002a) which can compromise informed choice (Marteau et al., 1992), others doubt whether fully autonomous and informed decision-making within the context of Down's syndrome screening can ever exist (Pilnick 2004).

Practitioners encountering difficulty in communicating screening information whilst remaining non-directive and ensuring informed choice is reported elsewhere (García et al. 2008; Heyman et al. 2006; Pilnick et al. 2004). This stems from the conflict between the time practitioners have available to explain screening and the time needed to discuss the procedure (Sooben 2010; Vassy 2006; Williams et al. 2002a), the trouble of conveying (risk) information and the practical/ethical aspects of screening (Burton-Jeangros et al. 2013; Ekelin and Crang-Svalenius 2004; Heyman et al. 2006; Hunt et al. 2005), communication breakdowns when expectant parents' first language is not the native language (Hey and Hurst 2003), and the different definitions between expectant parents and practitioners of what constitutes a "normal result" and/or "normal child" (Hunt et al. 2005; Vassy 2006; Williams 2006). Additionally, research shows how some practitioners do not know how to best support higher-risk expectant parents (Getz and Kirkengen 2003; Williams et al. 2002a, 2002c), how some expectant parents are not fully aware of the key features of Down's syndrome prior to screening and testing (Williams et al. 2002c), and how expectant parents may not fully understand or have realistic expectations of screening (Burton-Jeangros et al. 2013; Gammons et al. 2010; van den Berg 2005b). Such difficulties cast doubt on whether the ethical principles of "choice" and "autonomy" are realistic in modern healthcare systems.

Some studies have explored how practitioners can respond to such difficulties. Burton-Jeangros et al. (2013) show how practitioners commonly draw on subjective

interpretations and cultural meanings of scientific productions (i.e. risk factors) to erect an understanding. In addition, Burton-Jeangros et al. (2013) and Ivry (2006) reveal how practitioners can withhold some information from expectant parents around possible pregnancy outcomes to avoid causing unnecessary anxiety. Whilst this care is directive, this may not *always* be defined as bad practice. During interviews with practitioners, Schwennesen and Koch (2012: 283) show how their conduct, whilst perhaps not always immediately compatible with the non-directive ethos, can constitute “good care.” By answering expectant parents’ appeal for some direction, for instance, practitioners move closer to promising “informed choice” by supporting them to make meaningful decisions largely on the basis of uncertain knowledge. Rather than this being described as a serious problem of oppressive power resulting in coercive moments of decision-making, such actions are categorised as ensuring informed choice since non-directive care may not always be the most suitable response (Schwennesen and Koch 2012; Williams et al. 2002b).

Nonetheless, many of these studies show how Down’s syndrome screening – particularly when involving interactions between expectant parents and practitioners – presents a number of perhaps irresolvable problems for each respective party. Others have reported further concerns around how practitioners, whilst sometimes critical of screening techniques for inducing unnecessary anxiety and *limiting* expectant parents’ decision-making capacity (Burton-Jeangros et al. 2013; Williams et al. 2002c), are both undertrained in communicating screening information (Cleary-Goodman et al. 2006; Sandall et al. 2001; Skirton and Barr 2007) and hold limited knowledge of Down’s syndrome (Dormandy et al. 2006; Skirton and Barr 2010) and screening for the condition (Farsides et al. 2004; Hey and Hurst 2003; Samwill 2002; Smith et al. 1994; Williams et al. 2002c). This involves having little direct contact during medical training

with people who have developmental disabilities (Cleary-Goldman et al. 2006; Driscoll et al. 2009). Many such studies conclude, thus, that Down's syndrome screening is a problematic practice and further training/policy intervention is recommended.

Knowledge gaps

The substance and contribution of the reviewed studies is irrefutable. However, existing insights can be subjected to a number of criticisms. I highlight these knowledge gaps for the remainder of this article. First, some important voices are missing, or at least relatively silent, in the literature. This includes expectant fathers who, whilst vocal in some studies (Draper 2002), are often neglected in favour of studying pregnant women. In addition, there is a scarcity of research on expectant parents who decide to terminate a pregnancy, though I suspect practical and ethical concerns explain this absence. Nonetheless, exceptions are available (Helm et al. 1998; Korenromp et al. 2007; Olarte Sierra 2010; Reist 2006; Skotko 2005; Tymstra et al. 2004) and it would be valuable for researchers to further explore the views of the minority of expectant parents continuing a pregnancy after a Down's syndrome diagnosis. Research also frequently disregards expectant mothers at an "advanced maternal age". Whilst research has attended to how expectant mothers cite their age when accounting for their decision-making processes, very few (if any) have explored the implications the label of "advanced maternal age" has for their identity-work. This becomes more significant when considering how mothers are frequently categorised as morally responsible for containing risks and preventing harm to born/unborn children (Gottfreðsdóttir et al. 2009b; García et al. 2011; Lupton 2013; Wolf 2011). Additionally, this reflects how pregnant women are often presented as a homogenous group within such research, with little attention being paid to how attributes such as age, class, race, and social/economic background play a

role in screening practices. This also relates to dismissing the importance of the cultural context on expectant parents' attitudes toward screening, although Ivry (2006, 2009) provides excellent exceptions. Furthermore, as noted by Pilnick and Zayts (2012), expectant parents who receive a higher-risk for Down's syndrome have been the subject of little research. Whilst such research exists (Burton-Jeangros et al. 2013; Heyman et al. 2006), it is by no means abundant. Finally, whilst research on practitioners exists, this likewise generates a relatively undersized literature (McCourt 2002; Tsouroufli 2011; Williams et al. 2002a, 2002c). The important findings of these small but limited collections of research indicate that these groups deserve more attention beyond the brevity they are currently afforded.

A second criticism is that research is regularly based on retrospective accounts of expectant parents and practitioners (Aune and Möller 2012; Bryant et al. 2010; Burton-Jeangros et al. 2013; García et al. 2008; Gottfreðsdóttir et al. 2009a; Heyman 2010; McNeill et al. 2009; Press and Browner 1998; Santalahti et al. 1998; Williams et al. 2005). Focusing solely on practitioners, studies commonly use questionnaire and/or interview data or, more problematically, ground arguments in post-hoc accounts of expectant parents (Gammons et al. 2010; Skotko 2005; Sooben 2010). Whilst a key locus of enquiry, methodological choices in many studies of expectant parents and practitioners, predominantly questionnaires (Jaques et al. 2004; Samwill 2002; Smith et al. 1994; van den Berg 2005a) and interviews (Burton-Jeangros et al. 2013; Farsides et al. 2004; Williams 2002b, 2000c), make invisible the mundane aspects of, and meaning-making practices in, medical encounters. By de-contextualising context-specific situations and relying on romantic conceptions of a rational experiencing individual, such research is rarely grounded in ethnographic data which would capture how screening is enacted, managed, and negotiated between practitioners and expectant

parents in everyday routines. Indeed, as Jerolmack and Khan (2013, 13) claim, what people say is frequently a poor predictor of what they do; “to escape [this] attitudinal fallacy, we must study interaction.” Ethnographic exceptions are evident (Heyman and Henriksen 2001; Hunt et al. 2005; Ivry 2006, 2009; McCourt 2002; Pilnick 2004, 2008; Pilnick and Zayts 2012; Schwennesen and Koch 2012) but they are far from abundant and, more critically, often focus on consultations of Down’s syndrome screening alone. This is a grave sampling error since it mistakes a part for the whole; consultations, indeed, are only a microcosm for all aspects of medical work around prenatal screening for Down’s syndrome.

A third criticism is that many studies on Down’s syndrome screening fail to fully discuss the condition and subscribe to medical definitions of Down’s syndrome or disability more generally. Exceptions have been identified; Sooben (2010), for instance, claims practitioners provide a functional and brief description of Down’s syndrome during screening which conforms to a biomedical problem orientation. Bryant et al. (2006) similarly argue that antenatal settings currently provide little opportunity for people to discuss and explore their beliefs about disability. In an analysis of descriptive information about Down’s syndrome in antenatal leaflets, Bryant et al. (2001) and Murray et al. (2001) suggest this literature contains false, misleading, and inconsistent information and fashions a negative image of the condition. However, studies have not conducted extensive empirical research, particularly through in-depth ethnographic observations of clinical settings, on how Down’s syndrome is discussed within screening consultations and how accusations of screening fostering a belief that Down’s syndrome should be prevented (Alderson 2001; Sooben 2010; Vassy 2006) play out in the clinic. Specifically, few focus on its definition as a “risk,” a discourse which implies impending danger which should be avoided (Douglas 1992; Shakespeare 1999), and how this

produces effects for parents and practitioners. A valuable contribution, for instance, could be an exploration of how risk communication homogenises Down's syndrome and masks the sizeable physiological and intellectual variation of people with the condition. Nonetheless, the small number of studies discussing Down's syndrome in antenatal care often fail to recognise how disability more generally emerges through cultural ideas of "the normal" and how it is shaped by a complex interplay of social, cultural, material, biological, economic, and political factors (Author forthcoming [2014]; Barnes and Mercer 2010; Davis 1995; Ginsburg and Rapp 2013; Oliver 1990; Shakespeare 1999).

Conclusion

In this article, I have reviewed research on parent and practitioner experiences of screening for Down's syndrome. It is a practice which has revolutionised reproductive medicine and changed our conceptions of pregnancy, parenthood, the family, and ideas surrounding the "normal." I have shown how current research on Down's syndrome screening focuses on either the decision-making practices of expectant parents or the interactions between parents and practitioners. I conclude by acknowledging the gaps in existing insights and the fruitful avenues for future exploration. Rather than repeating earlier expositions, that is, of pouring old wine into new wineskins, I suggest that sociologists should take account of these openings. I personally press, as a matter of urgency, carrying out a context-privy analysis of clinical practice regarding Down's syndrome screening. A close ethnographic reading of everyday affairs will unmask the ongoing, complex, and diverse ways in which knowledge, meanings, and positions are produced and reproduced in the hospital. Through ethnographic fieldwork, we become attentive to ordinary information usually invisible and taken-for-granted rather than

allowing implicit assumptions to govern understandings. In doing so, we may well fill in the remaining gaps of knowledge outlined above.

Notes

1. I often cite statistics for Down's syndrome screening from England and Wales. This is because the statistics are easily accessible (since England and Wales has a standardised healthcare system) and because this is where I conducted my research.
2. Other conditions including Edward's syndrome (Trisomy 18) and Patau's syndrome (Trisomy 13) may be suspected via prenatal screening, and confirmed via diagnostic testing, for Down's syndrome.
3. Meta-syntheses were excluded from the article. See Reid et al. (2009) for an excellent meta-synthesis on pregnant women's decision-making processes around screening for Down syndrome.
4. A nuchal translucency is the fluid thickness in the nape of an unborn baby's neck. Enlarged fluid is associated with chromosomal conditions including Down's syndrome.

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