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Variation Exists in Service Delivery: Similarities and Differences in the Provision of a Whole Genome Sequencing Service for Paediatric Rare Disease Patients in the National Health Service in England

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Whole genome sequencing · Implementation science · Clinical genomics · Process map

Abstract

Introduction: The National Health Service (NHS) in England is the first to offer whole genome sequencing (WGS) as part of standard care. As a high-income country with a universal healthcare system, England contributes a valuable perspective to global developments in WGS. ***Methods:*** We used an implementation science approach with mixed methods to characterise delivery of WGS for paediatric rare diseases: observations and field notes of consent appointments in clinical genetics and mainstream settings and follow-up qualitative semi-structured interviews with the clinical team. Process maps were developed for each department to identify similarities and variations between sites and thematic analysis of interview data to understand barriers and facilitators. ***Results:*** Data collection occurred in 12 departments (7 genetic, 3 neurology, 1 cardiology, and 1 general paediatric) across 7 NHS Trusts. 26 observations of 21 healthcare professionals were conducted, alongside 19 follow-up interviews. Two master maps were developed – one for clinical genetics and one for the mainstream. We identified 11 steps involved in delivering WGS, including 9 variations and 9 similarities. We identified most variation in the processes related to the "*who*," "*when*," "*how*," and "*where*" as these were aspects that could be adapted to fit into the specific set-up of the department. Barriers included reluctance to uptake in the mainstream and difficulties tracking samples. ***Conclusion:*** Recommendations include developing standard operating procedures and hiring healthcare professionals responsible for facilitating consent alongside administrative aspects. These would reduce the burden on clinical geneticists and improve turnaround times as well as contribute to streamlining and standardisation of the service.

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Introduction

The British Government has set out to create "the most advanced genomic healthcare system in the world" [1]. Since 2018, the National Health Service (NHS) in England has been offering whole genome sequencing (WGS) as part of standard care for the diagnosis and research of certain rare disease indications and cancer, as specified by the National Genomic Test Directory [2, 3]. The NHS Genomic Medicine Service (GMS) in England is structured around seven Genomic Laboratory Hubs (GLHs) and Genomic Medicine Service Alliances (GMSAs), which are responsible for genomic testing and embedding genomics into mainstream care in their geographical region [2]. These NHS GLHs and GMSAs work closely with a network of 17 Clinical Genetics Services across the country that provides specialist support to clinicians and patients in their region. Genomic tests including WGS can be ordered by specialists outside clinical genetics, thus encouraging the "mainstreaming" of genomics. Genomic associates and genetic counsellors are healthcare professionals that help with the process of genetic testing in the UK; they are often based in clinical genetics departments.

WGS as a diagnostic test for rare disease patients is set to have a profound impact. A condition is said to be rare if it affects fewer than 1 in 2,000 people; however, 1 in 17 people will be affected by a rare disease at some point in their lifetime [4]. Rare diseases are often associated with delays in obtaining a diagnosis, frequently referred to as the "diagnostic Odyssey," and can involve numerous referrals to different specialists and a battery of invasive tests until a diagnosis is reached [5, 6]. A European survey from 2004 showed that the time between early identification of symptoms and a final diagnosis for a subset of rare diseases was 5–30 years for 25% of patients [7]. NHS-funded WGS for paediatric rare diseases is primarily offered when a child has a suspected genetic disorder that remains undiagnosed despite extensive testing or a severe developmental abnormality or syndrome which would benefit from genetic insight to inform diagnosis or treatment. Although the whole genome is sequenced, analysis is focussed on areas of the genome that correlate with the patient's reported clinical and family history. Trio testing whereby both the patient and their parents' genomes are sequenced, is recommended for paediatric referrals, and has been shown to improve diagnostic yield [8]. Trio testing is the preferred form of WGS in the NHS; however, if a parental sample cannot be obtained, clinicians will go ahead with duo or singleton testing. Trio testing in WGS differs from other types of genomic testing, where testing is typically initiated just on the patient, with familial/parental testing taking place afterwards depending on the result.

WGS and analysis in the NHS GMS is facilitated by Genomics England. Patients offered WGS are also asked if they wish to contribute their samples, genome data, and ongoing collection of health data to the National Genomic Research Library (NGRL). This is a secure, national and de-identified database managed by Genomics England, which also includes research participants. It enables approved researchers to use the samples and data to study diseases, identify new diagnoses and look for new treatments. Therefore, as part of the consent process, patients who make an informed decision to have WGS also make a separate decision about whether to become a participant of the NGRL. Their choices are recorded on a standardised Record of Discussion (RoD) form.

Introduction of a novel technology into an already complex healthcare system comes with many challenges. Case studies have shown that proof-of-concept is not sufficient for adoption of a new intervention into routine usage with fewer than 50% of clinical innovations ever making it into general use [9]. Notably, collaboration across specialisms will be inherent to the successful delivery of WGS since it is a multi-stage process involving the identification of suitable patients, accurate phenotyping, supporting the consent process, obtaining blood samples, sequencing the genome, interpreting variants, delivering results, exploring treatment options, and potentially cascade testing [10].

Our research uses an implementation science approach to understand how WGS is being delivered as a clinical service across the NHS. The setting for this study, the first high-income country with a universal healthcare system to introduce genomic testing as a clinical service, makes the findings from this study an important addition to the literature. Implementation science is a research approach used to bridge the gap between validated innovations and clinical practice [9]. Existing studies show that leveraging an implementation science approach to the introduction of WGS services produces results of value to the healthcare system that was studied [11, 12]. For example, research in Australia has highlighted that barriers to the implementation of genomic testing for rare diseases included a lack of perceived value for the community, lack of knowledge among clinicians, and absence of leadership [13]. In the USA, the Implementing Genomics in Practice (IGNITE) Network has been exploring methods for effective implementation, diffusion, and sustainability of genomic testing in diverse clinical settings [14]. Findings from that initiative have shown

valuable insights into possible obstacles for the implementation of WGS including lack of integration of genomics data with electronic healthcare records and the need to strengthen clinicians' knowledge and beliefs about genomic medicine [15]. The need to get buy-in from a complex network of stakeholders is also documented in France and Quebec [16], and is high on the priority list for The France Plan Medicine Genomique 2025 which aims to develop a national framework for big-genomic data [16]. A review of literature published between 2017 and 2022, reports on implementation efforts of large-scale genomic screening or diagnostic programmes in sixteen countries with most in the pilot phases [17].

In our study conducted in England, we used process mapping, similar to that described in Antonacci et al. [18] involving data gathering, process map generation, analysis and recommendations. Process maps are one tool in the toolbox of implementation science research that is used to identify the steps and actors involved in implementing a novel technology. For example, how many appointments are involved in the WGS process and who is involved at each stage, additionally when, by whom, and where are crucial actions carried out to ensure that WGS is successfully delivered. Process maps can also help identify the barriers and facilitators at each step of WGS service delivery, which can then lead to the development of recommendations for practice. Process mapping, with data collected using an annotated interview process map, was an approach used successfully by Best et al. [12] to explore the scaling up of clinical genomics in Australia including understanding variations in service delivery. Through this approach, they were able to build a detailed understanding of the process of carrying out genomic testing, identify variation across the service as well as identify barriers to service delivery. We therefore approached this work with a similar set of aims and using a process mapping approach but in a different healthcare setting where the genomics service was delivered at a national level as opposed to one adopting both national and local approaches.

Materials and Methods

This study was part of a larger research programme examining the implementation of WGS for paediatric rare disease in the NHS GMS as detailed in our protocol paper [19]. The overarching aims of the programme are to evaluate the implementation of the GMS during its early years, identify barriers and facilitators to successful implementation, and provide recommendations for practice.

Study Design

The study uses an implementation science approach with mixed methods. Our methods combined observations along with detailed field notes and follow-up qualitative interviews. Using both observations as well as interviews enabled us to build a more complete picture of how WGS was being delivered in the NHS in England. Interviews and observations are key methods in implementation research [20]. This was a cross-sectional, abductive (a "hybrid" of inductive and deductive analysis), qualitative study. The principles of abductive research were followed as outlined in Thompson [21]. Data were collected at 7 NHS Trusts through observations of clinic appointments for WGS consent and follow-up interviews with the HCPs involved.

Ethics

Ethical approval for the study was approved on 16th July 2021 by the London Bloomsbury Research Ethics Committee (21/PR/0678). All participants, including health professionals conducting consent appointments as well as patients and families being observed, received a Participant Information Sheet prior to deciding whether to participate. Age appropriate Participant Information Sheets were also designed for younger patients. Written informed consent to participate in the study was obtained from all adult participants and all underaged participants' parents. A separate consent form was used for the follow-up interviews.

Participants and Recruitment

HCPs, paediatric patients with undiagnosed rare conditions, and the patients' parents were recruited to take part in observations of clinic appointments and follow-up semi-structured interviews. HCPs were approached via purposive sampling, who then selected appointments for observation via convenience sampling of patients. The inclusion criteria for the families were that the paediatric patient had a rare condition and that the parents provide informed consent to participate in the study for themselves and on behalf of the paediatric patient. If consent was provided, CL observed the appointment and afterwards CL interviewed the responsible HCPs about the appointment.

We aimed for three observations at each of the seven sites including a mix in terms of clinical specialities (genetics, neurology, paediatrics, etc.) as well as type of HCP conducting the appointment (clinician, genetic counsellor, etc.) (Table 1). This number (n = 21) was

Table 1. Summary of participant characteristics (healthcare professionals)

Participant characteristics	*n* = 21
Age	
21–30, years	5
31–40, years	3
41–50, years	9
51–60, years	2
Unknown	2
Gender	
Female	17
Male	4
Department	
Clinical genetics	7 (16 observations)
Paediatric neurology	3 (8 observations)
Paediatric cardiology	1 (1 observation)
General paediatrics	1 (1 observation)
Role	
Consultant	11
Genomics associate	4
Clinical fellow	2
Speciality doctor	1
Speciality registrar	1
Pre-reg genetic counsellor	1
Specialist nurse	1
Years in role	
>1 year	6
1–5 years	6
6–10 years	2
10+ years	5
Unknown	2
Genomics experience	
Lots	13
Some	6
None	2
Consent patients into 100,000 genomes project	
Yes	9
No	11
Unknown	1

determined pragmatically and considered sufficient to give a good overview of how the service was being delivered, as well as being manageable for the research team at each site who were contacting patients and families about the study. The seven sites were located in London, the North of England, and the South of England to enhance participant diversity and reduce geographical bias (Table 1). Principal investigators based in clinical genetics departments at each of the seven NHS Trusts, were asked to identify HCPs from both clinical genetics and mainstream services within their Trust who consented parents for WGS. CL then contacted them via email to explain the study and send a Participant Information Sheet, consent form and participant demographics form. At one site, CL also promoted the research in a departmental meeting, and 1 HCP agreed to take part following this.

Participating HCPs were asked to identify upcoming clinical appointments where a patient/family was likely to be offered WGS. Once a family had been identified, they were either (a) sent a participant information sheet and cover letter prior to their clinic appointment with a follow-up phone call from a member of the administrative team or the clinician to assess interest in participating or (b) CL approached the family in the waiting area prior to their appointment to explain the study, give them the participant information sheet and assess interest in participating. Those families that were interested in participating were asked to complete a consent form and a participant demographic form prior to the clinic appointment beginning. All formats of appointment were eligible including face-to-face, video-call, and telephone. CL conducted all observations of clinic appointments. In some cases, more than one clinic appointment was observed involving the same HCP.

Data Collection and Procedure

Observations of clinic appointments and semi-structured interviews were conducted between November 2021 and October 2022. A field note template was developed in collaboration with stakeholders specifically for the study. Data captured included clinical speciality, whether patient/parents has been seen previously by clinician, type of appointment, e.g., routine clinic appointment or whether appointment set up specifically to discuss WGS, type of HCP conducting appointment, information sent prior to appointment, topics discussed during appointment, how bloods are collected, how consent is recorded, what other administrative tasks are conducted during appointment, what information patients receive or will receive after appointment, length of appointment, and any other notable observations. Field notes were completed by CL during and immediately after observing the appointment. HCPs were subsequently invited by CL to take part in a follow-up interview face-to-face (n = 7) or using virtual video conference software (n = 12). Interviews were on average 35.56 min long, and the range was 20.15–57.10 min.

To support process mapping and to maximise the information gained from the follow-up interviews, the interview topic guide was structured according to the framework of the phases involved in WGS. The questions aimed to address each stage of the process including how patients are selected for WGS, the different ways WGS

appointments occur, who can obtain consent from the patient, where and how blood samples are sent, and when and how results are delivered. The topic guide also included questions to elicit views on the current WGS process including perceived barriers and facilitators. As the focus was on HCP's experiences, specific laboratory procedures were not investigated in detail.

Data Analysis

Field notes from observations were reviewed and follow-up HCP interviews were transcribed verbatim for data analysis. A two-stage process was taken during data analysis to (1) build process maps and (2) thematically analyse interviews for information on barriers and facilitators. Data analysis was organised according to a predefined framework delineating the four sequential stages of the WGS process that we created using the investigators' existing knowledge of WGS delivery as well as prior research [12]. The four steps in the framework were (1) selecting patients for WGS, (2) the consent process, (3) the testing procedure, and (4) delivering results to the patients. We then conducted a thematic analysis of qualitative data from detailed field notes recorded during observations of clinic appointments and follow-up interviews with HCPs to populate the framework and identify similarities and differences across departments and clinical specialities (Stage 1 – deductive). We also inductively coded the data to look specifically at the barriers and facilitators to service delivery (Stage 2 – inductive).

The first stage of data analysis involved two elements in building process maps, with the first (Stage 1a) visually drawing out the steps and HCPs involved in WGS and how the tasks are divided among different actors, e.g., "clinician decides if the patient is eligible for WGS." This involved iteratively reading interviews with HCPs from each department involved in the study. We built one process map per department to understand how they worked as a unit. As the interviews were read, boxes representing steps were added onto the map canvas for the department. Maps were reviewed iteratively after each interview for a department was analysed and duplicate boxes within a map were merged or deleted for clarity and coherence. In the first instance, both C.L. and N.M.L. independently analysed the interviews/field notes for two departments and populated the process maps. The maps were compared, and any discrepancies discussed and resolved. As there was high concordance, the remainder of the process maps were built by one researcher (N.M.L.) with ongoing regular meetings (C.L. and N.M.L.) to discuss findings. Once the process maps were completed, we compared them visually one-by-one across each of the four sequential stages and made a list of all the approaches to identify similarities and differences between departments. At this stage we were able to break down the four stages into a more granular sequence of steps. The second element of data analysis (Stage 1b) involved developing master maps. Initially, our aim was to develop one overarching map from the twelve individual maps. However, because of the significant amount of variation across the maps, we decided to construct two maps: one summarising the process for clinical genetics departments and one summarising the process for mainstream departments. We presented the process maps in interim meetings with the study advisory team and a participating department to verify their accuracy.

The next stage of data analysis (Stage 2) involved thematic analysis of the interview transcripts following the guidance of Braun and Clarke [22]. We chose thematic analysis as it facilitates the development of themes, enables the capturing of semantic meaning, orientates the data inductively and is theoretically flexible [23]. This round of analysing the transcripts involved explicit examination for barriers and facilitators in the implementation of WGS. While this stage was predominantly inductive, the predefined framework of stages involved in the WGS process was used to categorise codes arising from the transcripts. NVivo12 software was used to facilitate coding and data analysis. Initially, both C.L. and N.M.L. independently coded two transcripts and compared their coding for discrepancies. As there was a high concordance, coding of the remainder of the transcripts was completed by one researcher (N.M.L.) with ongoing regular meetings (C.L. and N.M.L.) to discuss findings. Finally, the themes and sub-themes were incorporated into a thematic map.

C.L. and N.M.L. identified key barriers and associated facilitators from the process maps and qualitative interviews. We tabulated these, translating the facilitators into recommendations for practice. These were then discussed among the co-authors (comprising genetic counsellors, a genetic consultant and a social scientist) and minor changes were made to ensure recommendations were feasible.

Results

Sample Demographics

We observed 26 WGS appointments conducted by 21 HCPs in 12 departments – 7 clinical genetics departments (16 observations), 3 paediatric neurology departments (8 observations), 1 paediatric cardiology department (1 observation), and 1 general paediatrics department (1 observation). These were located across 7 NHS Trusts in England located in London ($n = 3$), the South East ($n = 1$), and the North of England ($n = 3$). HCPs that took part in the observation study included consultants ($n = 11$),

genomics associates (n = 4), clinical fellows (n = 2), a speciality doctor (n = 1), a speciality registrar (n = 1), a pre-registration genetic counsellor (n = 1), and a specialist nurse (n = 1) (Table 1). Observations were of appointments of children with a range of conditions including intellectual disability (n = 15), congenital malformations (n = 9), epilepsy (n = 3), hereditary spastic paraplegia (n = 2), neuropathy (n = 2), cerebral malformation (n = 1), ultra-rare and atypical monogenic disorders (n = 1), hereditary ataxia (n = 1), cerebellar anomalies (n = 1), and cardiomyopathy (n = 1). In total, 54 parents were observed who were aged between 19 and 36 years, 25 of whom were female, 25 educated to GCSE level or lower, 28 self-reporting as White ethnicity and 14 as Asian ethnicity. Follow-up interviews were conducted with 19 HCPs, 18 of whom took part in the observation study including consultants (n = 10), genomics associates (n = 3), clinical fellows (n = 1), a speciality doctor (n = 1), a speciality registrar (n = 1), a pre-registration genetic counsellor (n = 1), and a specialist nurse (n = 1), and 1 WGS co-ordinator, who had an administrative role in one of the clinical genetics departments.

Process Maps

At the time of conducting the study, very few participants had returned WGS results since its implementation into the GMS. Therefore, most of the detail in the maps relates to the first three stages and not returned results, although participants did reflect on what they were most likely to do at this stage. N.M.L. colour coded the maps according to the phases of the WGS process. Different shapes were used to represent the start of the patient journey (arrow), key decision points (diamond), and standard process steps (rectangle). For example, decision points regarding whether the patient should be handled by a clinician or other HCP are denoted by the diamond. Textured boxes signify that a step can occur at different time points, e.g., blood draw. Arrows denote whether steps are standard (single line) or optional (dotted).

Variations and Similarities between Departments

We identified 11 steps in the WGS process of which there were 9 variations between departments and 9 similarities (Table 2). Variations included (a) whether consent takes place in a mainstream or genetic clinic (b) whether, how and what information parent(s) receive prior to appointment, (c) professional background of person obtaining consent, (d) whether the consent discussion is embedded or separate from clinic appointment, (e) format for recording consent, (f) which professional completes required paperwork and when, (g) procedure for chasing samples and/or consent forms, (h) when and where bloods are taken. Similarities included (a) specialities referring for WGS, (b) content of consent discussion, (c) required paperwork for submission to laboratory, (d) communication of results.

Variations and similarities were not mutually exclusive as some steps of the process had both variations and similarities between departments. When analysing the data, it was apparent that each step of the process would have a "*who*" and "*when*" associated with it and, if appropriate, a "*how*," "*where*," and "*what*." Notably, we found that most of the variation related to the "*who*," "*when*," "*how*," and "*where*." For example, we found that *how* the patient entered the WGS pathway varied across department – in some cases, the patient was referred from a mainstream specialist to clinical genetics; in other cases, the mainstream clinician consented the patient/parents for WGS themselves. Some patients had been seen in that service previously, but before WGS was available routinely, and in other cases, they were new patients. Another example where the *who* and *when* varied between departments was the consent discussion; consent could be conducted by a clinical geneticist, genomics associate, genetic counsellor, or specialist nurse, and it might take place at a routine clinic appointment or at a separate consent appointment. An example of variation for the *where* related to where bloods were taken; this could be at the same appointment where WGS consent occurred, a separate phlebotomy appointment, at a local hospital or a GP surgery. In comparison, we identified most similarities in the process related to the "*what*." For example, *what* was included in the consent discussion as the discussion was guided by standardised forms that are used across England for WGS consent, referred to as the RoD.

Comparison between Clinical Genetics and Mainstream Maps

Maps that represented a clinical genetics model of delivery were more likely to contain a variety of HCPs in the WGS process including genetic counsellors and genomics associates (a new cadre of HCP who supports the WGS consent process including conducting consent discussions and associated administrative aspects) (Fig. 1). Genetic counsellors and genomics associates were predominantly involved in facilitating the consent process and organising paperwork for test ordering. This enabled clinical genetics departments to separate the clinical appointment from an additional appointment to discuss in-depth and record consent, sometimes allocating a full 40 min to the consent discussion. Provided that enough resources were available, this consent

Table 2. Variations and similarities in the WGS process between 12 departments in the NHS GMS

Phase	Step number and description	Variations between departments	Similarities between departments
I. Selecting patients	1. Source of referrals	In some cases, patients would be referred to clinical genetics for WGS. In other cases, the family would be consented for WGS in the mainstream setting. Clinical genetics could reject the referral if it was felt that consent could be carried out in the mainstream setting	There were similarities in where patient referrals to clinical genetics came from (mainly neurology and paediatrics)
	2. Suitability of patient for WGS	The suitability of WGS for some patients could be discussed either at a wider MDT meeting or one-to-one with a consultant or member of the local clinical genetics department	
II. Facilitating informed consent	3. WGS information materials	There was variation in whether and what information families received prior to and after their appointment. Some families received no information about WGS beforehand, other received an NHSE leaflet. Similarly, not all families were given/sent the NHSE leaflet about WGS after their appointment. Some HCPs provided links in letters to additional materials (e.g., YouTube video)	HCPs said that if they were not already providing WGS information materials, they planned to do so in the future
	4. Facilitating consent	The consent discussion was either embedded inside a routine clinic appointment during which a medical history, family history and a physical examination might be carried out (if the patient had not been seen before) or, might be scheduled as a separate appointment with a clinician, genomic associate, genetic counsellor, or nurse	Field notes highlighted that the content of the consent discussion was similar across appointments, as guided by the standardised sections of the RoD forms
	5. Capturing consent on the RoD	There was variation in the way that consent was captured. Some departments printed the RoD form and asked parents to sign with a wet-ink signature. In other departments, HCPs electronically recorded remote consent	All patients are asked to sign the RoD form indicating their consent for WGS. Discussion and consent to participate in the NGRL occurred in the same appointment as WGS was discussed and consented for
III. Testing	6. Taking bloods	There was variation in approaches to taking bloods, if not already stored, including at different time points and locations. These included (1) within the WGS consent appointment if the HCP taking consent was phlebotomy trained, (2) in the phlebotomy department of the hospital where the WGS appointment was being conducted, (3) the parents were asked to get bloods done at their local hospital, (4) at the family's GP	All departments would carry out at least one follow-up call to try to get hold of a missing forms or blood samples from parents

Table 2 (continued)

Phase	Step number and description	Variations between departments	Similarities between departments
	7. Missing consent forms/bloods	Departments varied on how many times they chased a RoD form from the family and/or blood samples. For example, in one department missing forms/bloods were only chased up once and in another up to three times. Chasing of incomplete forms and/or bloods could be conducted by a clinician, genomic associate, genetic counsellor, or nurse	All departments would offer WGS even if they only had a single or duo sample, if a trio was not possible
	8. Completing paperwork and sending to laboratory	There was variation in who completed paperwork and sent it to the laboratory. Paperwork could be completed by a clinician, genomic associate, genetic counsellor, or nurse. In some cases, the paperwork was pre-populated with, e.g., name, NHS number, DoB, etc. prior to the appointment. In other cases, this was done during the appointment itself, and in other cases the form was completed after the appointment	Selection of what panel to use (denoted as a "R number" in the National Test Directory for the NHS GMS) and HPO terms on the test order form were always completed by the clinician
	9. Identifying variants	The degree to which the requesting consultant was involved in variant interpretation differed between departments. Some consultants specified genes of interest or liaised with the laboratory	
	10. Delivering results to HCP		The laboratory would always send the WGS report to the requesting clinician named on the test order form
IV. Delivering results	11. Communicating results to the patient		Diagnostic results and variants of uncertain significance would typically be delivered in-person (face-to-face or virtually) by the referring clinician. No findings results would typically be communicated via letter only (in 1 department by telephone)

HPO, human phenotype ontology; MDT, multi-disciplinary team; NHSE, NHS England; RoD, record of discussion; WGS, whole genome sequencing. Coloring of phases reflects coloring seen in the process maps. Blue = selecting patients, green = facilitating consent, purple = testing, orange = delivering results.

discussion sometimes occurred on the same day as the clinical appointment. Otherwise, it occurred at a later date via telephone, preventing patients from having to travel back and forth. Maps that represented a mainstream model of service delivery were more likely to centre the WGS process in the hands of the clinician who assessed the suitability of patients for testing, facilitated the consent process, as well as arranged bloods and completed/uploaded paperwork for test ordering (Fig. 2). This was found to significantly increase the workload of an individual clinician, although it also enabled all steps to be carried out on the same day.

Barriers and Facilitators

We identified several barriers and facilitators across the different stages of the WGS process (Fig. 3). Quotes supporting themes and sub-themes are provided in Table 3.

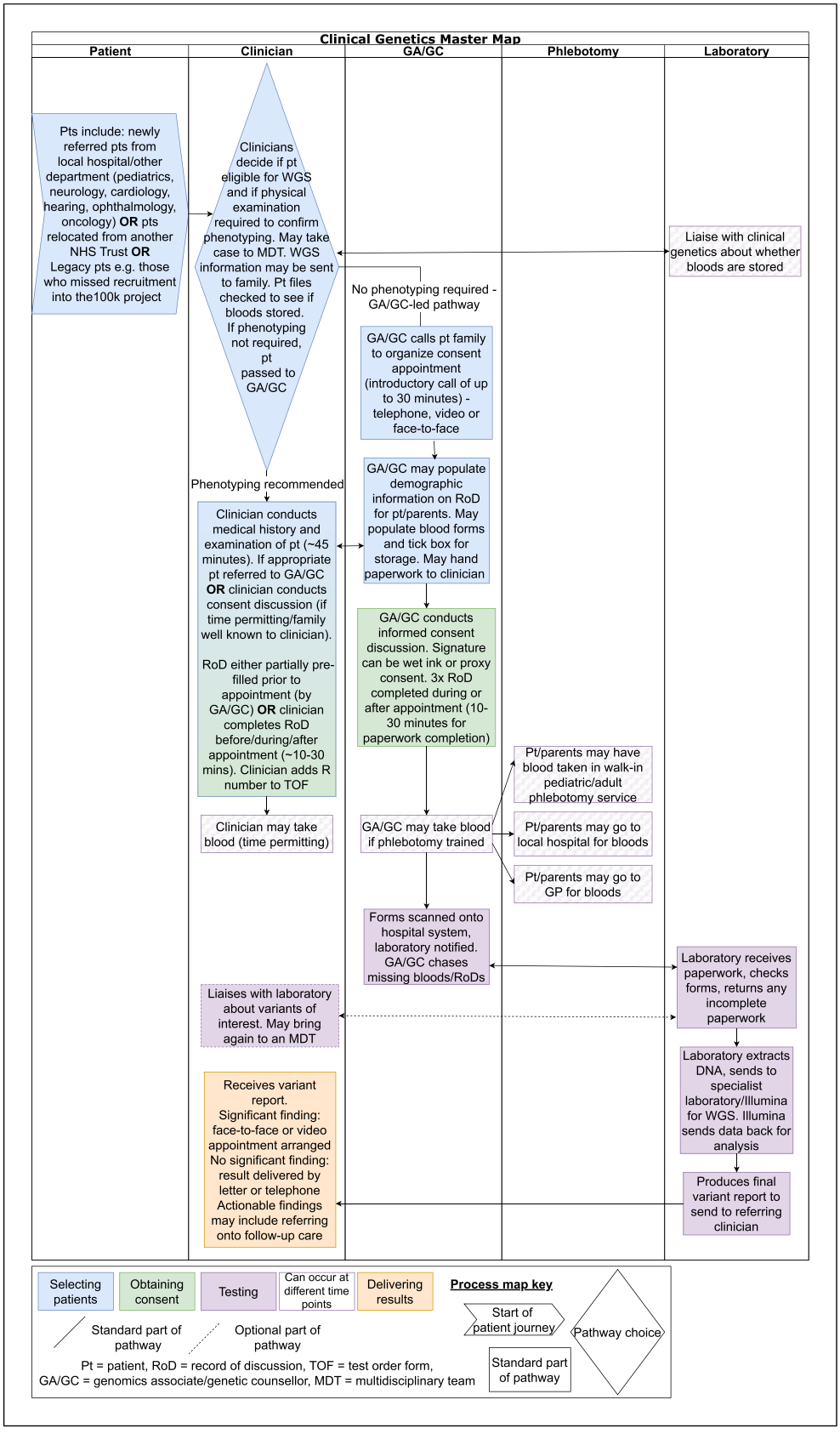

Fig. 1. Clinical genetics process map (master map) depicting the steps and professionals involved in the delivery of WGS in the clinical genetics setting.

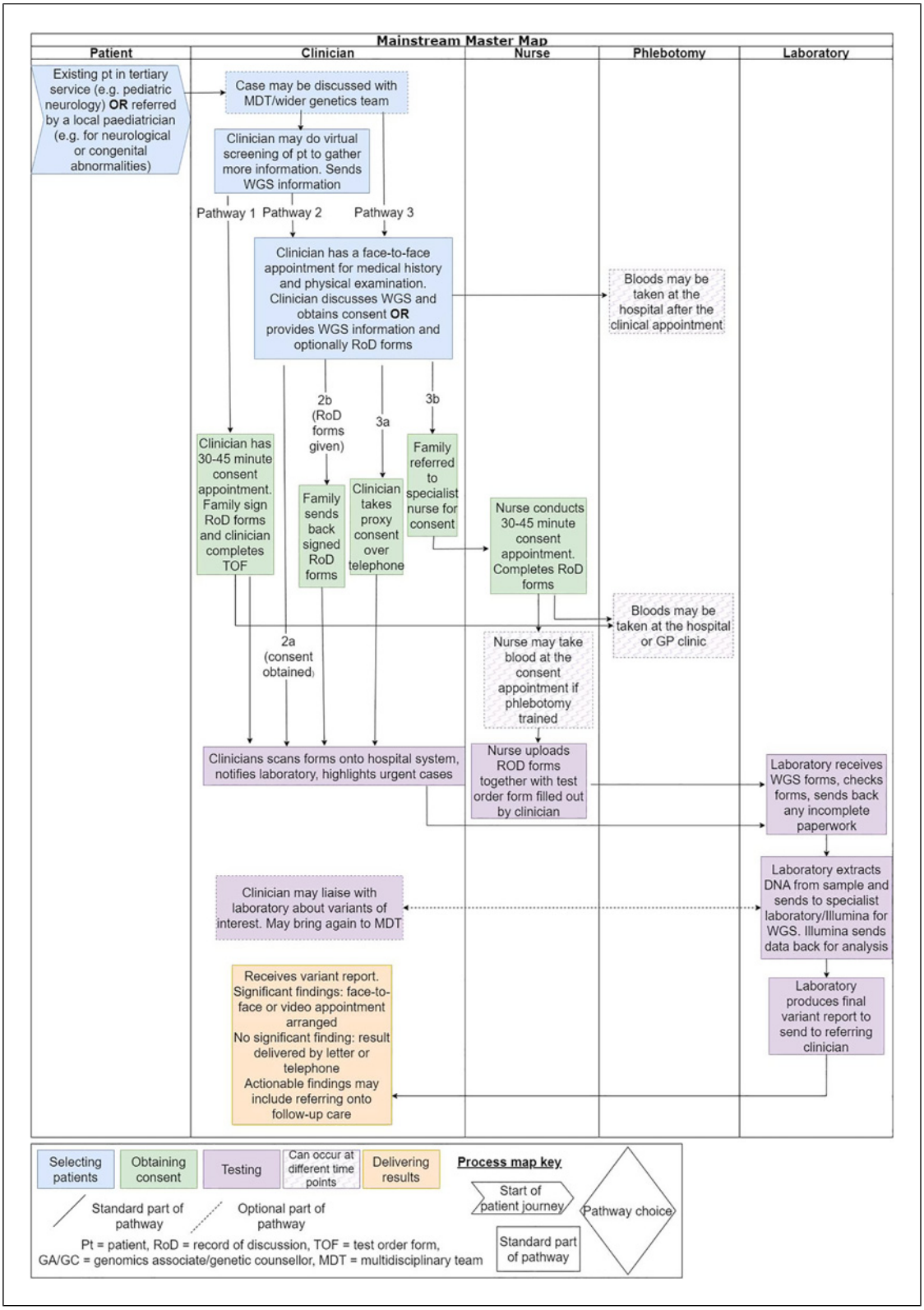

Fig. 2. Mainstream process map (master map) depicting the steps and professionals involved in the delivery of WGS in the mainstream setting.

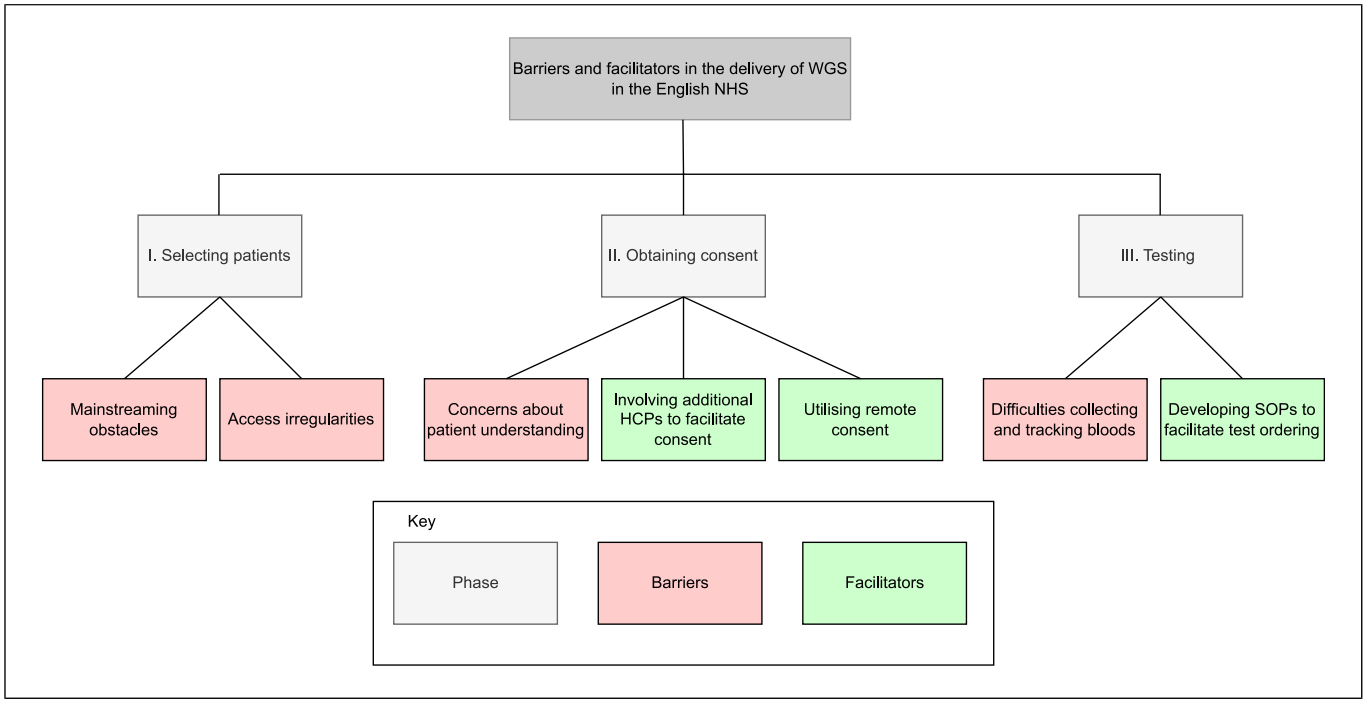

Fig. 3. Thematic map of themes and sub-themes relating to the delivery of WGS in the English NHS (light grey = overarching phase of the WGS process, red = barrier, green = facilitator).

Selecting Patients

Mainstreaming Obstacles

Identifying patients and ensuring that WGS is appropriate starts with processing referrals. Consultants in both the clinical genetics and the mainstream setting identified the additional time and capacity required for the WGS process as a barrier to uptake. Clinical geneticists felt overwhelmed by the volume of referrals and felt they would be unable to take on all cases of patients potentially suitable for WGS. HCPs highlighted the importance of mainstreaming of genomics to tackle this issue, although also acknowledged the resource constraints being experienced by mainstream colleagues and their potential discomfort with discussing it (Quote 1). Clinical geneticists also acknowledged the change in mindset that was required by mainstream colleagues to view genomics as part of their clinical practice and perceived this to be key to reducing the long waiting list for genetic testing that currently existed (Quote 2). Nevertheless, all HCPs noted challenges for mainstream colleagues in consenting patients during routine appointments that were generally shorter (20–30 min) than standard clinical genetics appointments (40 min) (Quote 3).

Access Irregularities

Participants reported a lack of consensus between departments about how to evaluate referrals for patients with intellectual disability. They felt that the threshold to accept a patient for WGS was not clearly defined (Quote 4). Participants also reported difficulties in discussing and sharing information about what WGS entails with patients whose first language was not English.

Overall, in the phase of referring and selecting patients for WGS (sub-divided into mainstreaming obstacles and access irregularities in our results), we report a tension between HCPs in clinical genetics and in the mainstream regarding how to share the responsibility of consenting patients for WGS. Participants in both settings felt too under-resourced and time constrained to make WGS decisions and initiate the process.

Facilitating Informed Consent

Concerns about Patient Understanding

There were concerns among some participants around lack of patient understanding and the potential for routinisation of testing, in particular if the clinician had recommended WGS (Quote 5). Some HCPs raised concerns around parents giving consent for taking part in the NGRL at the same time as consent for diagnostic

Table 3. Summary of themes, sub-themes, and quotes extracted through thematic analysis of qualitative semi-structured interviews with HCPs delivering WGS in the English NHS GMS

Theme	Sub-theme
I. Selecting patients	Obstacles with mainstreaming ***Quote 1:*** *"I don't think we'd be able to do every single record of discussion or every single referral ever in the area. The waiting list would be huge, so I think definitely part of it is teaching and integrating this into other [clinical] practices, which is difficult in itself with capacity and things, so we'll see how it goes."* ***HP18, Genomic Associate*** ***Quote 2****"They [mainstream clinicians] can request the testing and organise it all. So, ophthalmology, paediatric neurology – I'm not saying it's easy, and there's been some fairly robust letters back to the department saying this is what you're here for. It's like, well no actually. The whole point of this is that we're not the bottle neck anymore and having a patient sitting on our waiting list for a year, which is what's happening at the moment, doesn't help anyone. But then of course they'll say we don't have the set-up for this, we can't get details of parents. . ."* ***HP6, Consultant Clinical Geneticist*** ***Quote 3*** *"They [mainstream clinicians] don't have a 40-min appointment like the genetics department do, they have 20 to 30 min for their patients. And even with those 40 min, some of those patients need to be examined. . . so it just doesn't work with the paperwork involved. And you could do it afterwards. . . But then after their clinic they're doing clinic letters so it's just like, it's where do they fit in the time."* ***HP20, Lead Nurse***
	Irregularities in access ***Quote 4:*** *"I suppose the most challenging bit is deciding for the referrals, that either reach us or we gate keep before they ever actually become a referral through MDTs, it's that level of intellectual disability, what's the threshold. And I think it may be that different centres will pitch that slightly differently."* ***HP6, Consultant Clinical Geneticist***
II. Facilitating informed consent	Concerns around patient understanding ***Quote 5*** *"I think what can happen with a lot of genetic testing is that it becomes very routinised and because the doctor has said we need to do this test, it's a matter of yes we'll sign this document because doctor has said so, rather than because we've had a discussion, we actually understand and we want this for ourselves, or we don't want this for ourselves or for our child."* ***HP10, Pre-registration Genetic Counsellor*** ***Quote 6*** *"And in my opinion I don't really agree with including a research consent with a clinical consent because it should be separate because I think patients feel obliged to sign up for research, because they, although I say literally that "If you decide to not take part in the research you'll still get the same clinical test" I think for them it's quite difficult to see the difference if the doctor who's requesting the test asks at the same time to participate in research."* ***HP21, Consultant Clinical Geneticist***
	Hiring additional HCPs to facilitate consent process ***Quote 7*** *". . .so I'd say that that is the way we're aiming to do it [hiring genomics associates for consent] . . .Because that's felt to be the most efficient use of time and to get through our waiting list, and to get the patients seen by the right people at the right time."* ***HP4, Clinical Fellow in Genetics*** ***Quote 8*** *"Doing the initial phone call has actually been really, really helpful because then you get to engage with patients and you're not restricted by time where you've got a 45 min slot for the consenting appointment. . . so I feel that setting it up as a two appointment process has really helped with that and helped people to actually at the end of the first phone call say 'actually you know what, I have a bit to think about and that's been really helpful putting that in context, I'm going to discuss it with my partner' and then I'll say 'well let's book in an appointment, we can discuss it together and there's no obligation to say yes or no at the end of it, let's just have a chat about what's concerning you and what information you need'."* ***HP10, Pre-registration Genetic Counsellor*** ***Quote 9*** *"So actually having the genomics associates has cut down my prep time for, with the paperwork, from about half an hour – well that's being dramatic – 20 min to about four or 5 min. So, that's working really well having them on board and having them doing things"* ***HP4, Clinical Fellow***
	Utilising remote consent ***Quote 10*** *"if both parents are available but one of them can't come then the way we get around it if at all possible is then getting them on loudspeaker during the appointment, so we'll phone them on the mobile and they just listen in to the 10, 15 min to cover WGS and NGRL chat. If they're happy with that then just the remote consent box is ticked and that's them sorted."* ***HP6, Consultant Clinical Geneticist***

Table 3 (continued)

Theme	Sub-theme
III. Testing	Difficulties collecting and tracking bloods ***Quote 11*** *"It's just the practicalities of getting parental blood and the convenience of it, so just filling out forms, sending forms, or asking GPs or asking other services, I'm not sure how smoothly that's going to work. . . often very difficult to get the GP practices to do it. . .One of the things I think we've identified which is not in place is any kind of way of tracking so I'm very mindful that the onus is upon us as the consultant or the clinician to say, to actually check things get to the lab"* ***HP13, Consultant Paediatric Neurologist***
	Developing SOPs to facilitate test ordering ***Quote 12*** *"It's easier to have a look at. . .standard operating procedures. If I can't get in touch with a patient, I'll call them twice and then send a letter so we've kind of got it a bit more organised when we do the same for each patient, try and contact them a certain amount of times if it's not gone through. So, I feel like it's becoming more organised, yeah. There seems to be a system in place."* ***HP11, Whole Genome Sequencing Co-ordinator***

testing because they felt parents might "feel obliged to sign up" to take part in research and may not truly "see the difference" between signing up for diagnostic testing and research if these conversations happen during the same discussion (Quote 6).

Involving Additional HCPs to Facilitate Consent

Having additional HCPs such as genetic counsellors and genomics associates available to discuss and take consent, helped facilitate the consent process. This support structure was mostly found to occur in clinical genetics departments where the workload related to consenting patients and ordering tests was shared between medical consultants and other HCPs. This allowed consultants to use their time more efficiently by focussing on routine clinical genetics appointments, and perhaps introducing parents to WGS, while other HCPs (genetic counsellors and genomic associates) could focus on WGS consent discussion, taking consent as well as associated tasks (form filling, uploading documents, chasing blood samples etc.). This system was felt to be the most efficient way to tackle existing waiting lists (Quote 7), but also built in additional time for families to consider whether to consent for WGS, in particular if the consent discussion was predicated by an initial phone call (Quote 8). Consultants commented on how this system had cut down the time they spent on WGS paperwork (Quote 9).

Utilising Remote Consent

Some interviewees had been using remote consent in those instances where the consent appointment was being conducted virtually or by telephone. This was found to streamline the WGS process and removed the need for paper consent forms to be sent back and forth (including chasing forms that had not been returned) (Quote 10).

Testing

Difficulties Collecting and Tracking Bloods

There were several roadblocks to obtaining blood samples and ensuring that they reached the laboratories responsible for extracting DNA and sequencing the sample. If bloods were not already stored, bloods could not always be taken on the same day as the WGS consent discussion because not all hospitals had a walk-in paediatric and/or adult phlebotomy service. HCPs reported difficulty advising patients on how to book bloods at their local hospital due to localised procedures. It was also reported that some GP surgeries would not arrange blood draws. These logistical roadblocks caused delays in getting blood samples to the laboratory. Another related barrier was tracking whether blood samples had been received by the laboratory. HCPs were frustrated by the current system, whereby they had to remember to email the laboratory to check if a sample had been received, rather than having an automated notification system in place (Quote 11). Consultants felt it was not an efficient use of their time to track and chase blood samples and consent forms. This is especially complicated and time-consuming in trio testing, where the patient and both parents need to provide blood samples.

Developing Standard Operating Procedures to Facilitate Test Ordering

Some departments had hired additional HCPs such as genomic associates or administrators so that consultants did not have to spend time chasing forms and blood

samples. In addition, we identified that some departments had developed standard operating procedures (SOPs) which detailed procedures for consenting families and ordering WGS including guidelines around chasing missing samples and signed RoD forms, which was felt to provide clarity and efficiency (Quote 12).

Discussion

The NHS in England adopted a top-down approach of implementing genomic medicine into routine care, that contrasts to approaches used in other countries, such as the bottom-up, organic approach seen in Australia [24]. In Australia, genomic initiatives are at varying stages of implementation, and have differing resource, structural and cultural characteristics, meaning that it can be difficult to develop a generalisable model of implementation. Implementing genomic medicine on a national scale at the same time across NHS Trusts and individual departments is likely to confer benefits such as clear and consistent communication from leadership, faster implementation, clear accountability, avoidance of duplication, as well as consistency across sites regarding for whom and how the service is delivered [25]. To our knowledge, this is the first study to detail the various stages in the workflow for consenting, ordering and returning paediatric WGS results to parents in the newly established NHS GMS. Through this work we have been able to understand the variations and similarities that occur between departments as well as between clinical genetics and mainstream settings. Notably, and perhaps surprisingly, given that consistency might be expected from a top-down approach to implementation, our results show that at the time of conducting this study, variation existed in service delivery. This variation may be because of the flexibility embedded within the WGS service specification which allows sites to adapt the service to their particular contexts, or it may be because the study was conducted relatively soon after implementation into the NHS as sites were still "finding their feet." The next questions will be to look at whether these variations across the service still exist, whether they impact patient outcomes, and therefore how much variation, and at which touchpoints, should variation be tolerated. This is important as variation between settings might result in disparities in services available to patients or variations around informed consent processes, data reanalysis, and data sharing [15].

Many of the similarities we identified in our study were those that related to the specific guidance provided by NHS England around the content of the consent discussion and the specific forms that need to be completed (which we have referred to as the "*what*"). In comparison, where we identified most variation in the process related to the "*who*," "*when*," "*how*," and "*where*" as these were aspects that could be adapted to fit into the specific set-up of the department, i.e., the clinical context. A recurring example from our data was whether departments had hired additional HCPs such as genetic counsellors and genomic associates to take on tasks including consenting patients and families, paperwork completion and chasing blood samples and/or paperwork. Genomic associates represent a relatively new role in the genomics workforce which is often part of the genetic counsellor career structure and has a clinical role that is different from a secretary [26]. Notably, we found that genomic associates were consenting patients for genomic testing, despite their being uncertainty as to whether this should form part of their scope for practice [26]. Further research is required to understand the competencies of this cadre of health professionals to understand whether this is in fact appropriate.

We found that SOPs could help mitigate inconsistent care and health disparities. Specifically in the case of intellectual disability, where severity is highly variable, they could provide a consensus on when to offer WGS. In the case of following-up on consent and blood samples, SOPs would ensure equity in that all patients would be contacted the same number of times. In the case of SOPs, a top-down approach may be the most appropriate solution. However, a fine balance needs to be struck between allowing for flexibility across sites which have differing capacity, e.g., in terms of chasing samples, and ensuring equity of access and a standardised approach for patients. The expanded recommendations for practice identified through this study are presented in Table 4. While some of these recommendations are not exclusively relevant to WGS, all of them were identified as potentially improving the current WGS consent process and testing pathway.

The Genome UK 2022 report sets out the three strategic pillars for implementation of genomic healthcare: (1) diagnosis and personalised medicine in routine healthcare, (2) prevention through screening, and (3) supporting research [1]. Through this work we have been able to identify several barriers and facilitators to service delivery, which may directly impact pillar 1 of

Table 4. Recommendations for improving the delivery of WGS in the NHS in English developed from the barriers and facilitators identified in observations and interviews with HCPs

Point in the process	Finding	Recommendation
Qualitative interviews		
Access irregularities	Reluctance of mainstream staff to consent patients for genomic testing	Place genetic specialists in mainstream centres to help mainstream HCPs and facilitate implementation (fixed term roles or secondments from clinical genetics)
	Lack of consensus about threshold for referral of patients with intellectual disability for WGS	Clearer national guidelines to support consistency around which patients with intellectual disability are suitable for WGS to ensure equity of access
Process mapping		
WGS information materials	Not all families received information about WGS prior to appointment	Where possible, appointment letters should include either a link or hardcopy to resources about WGS and the NGRL, e.g., those developed by NHSE
Qualitative interviews		
Involving additional HCPs to facilitate consent	Having GCs and GAs available to discuss and take consent helped facilitate the consent process and allow consultants to use their time more efficiently	Establish role of GCs/GAs to support the WGS consent and test ordering process. Develop clear guidelines for their roles
	Having an initial phone call prior to the consent appointment gives families time to think through whether WGS would be suitable and any questions or concerns they may have prior to the appointment	Recommend where possible an initial phone call with family prior to consent appointment or at a minimum send families information about WGS and NGRL beforehand
Qualitative interviews		
Concerns about patient understanding	Concern around routinisation of testing and lack of distinction between consent for diagnostic testing and NGRL	Consenters to check patient understanding and explore motivations for consenting to NGRL. Previous research has highlighted the importance of asking open questions to explore attitudes and understanding, e.g., how do you feel about consenting to the NGRL?
Process mapping		
Capturing consent on the RoD	There was variation in whether sites routinely used wet-ink or remote consent	Further research to compare time saving and other potential benefits or routinely using remote consent
	Remote consent helped streamline the WGS process and removed the need for paper consent forms to be sent back and forth	Further research to assess patient and HCP preferences around wet-ink versus remote consent
Qualitative interviews		
Utilise remote consent appointments	HCPs were flexible and accommodating where both parents could not attend in-person, e.g., one parent joined virtually/telephone	Flexibility around how families attend consent appointments. Provide option of joining via telephone or virtual software where both parents cannot attend in-person
Process mapping		
Taking bloods	Only some HCPs taking WGS consent were also able to take blood at the same appointment	Explore whether upskilling WGS consenters to also take blood would be cost effective and/or streamline the test ordering process
	Roadblocks to obtaining blood samples if not already stored	As above
Qualitative interviews		
Difficulties collecting and tracking bloods	No automated system to track whether blood samples had arrived at the laboratory	IT solutions to digitise test ordering and blood sample-tracking to reduce laboratory-staff time spent manually entering data and tracking samples

Table 4 (continued)

Point in the process	Finding	Recommendation
Process mapping		
Missing consent forms/bloods	Some sites had developed and implemented SOPs for consenting families and ordering WGS including guidelines around chasing missing samples and signed RoD forms	Standardise the process for chasing blood samples and consent forms by encouraging departments to develop standard operating procedures (SOP) so that staff have clear processes in place for chasing samples and forms/bloods
Qualitative interviews		
Developing SOPs to facilitate test ordering	SOPs were felt to provide clarity and efficiency around processes	As above

the Genome UK strategy. Our findings across multiple stages selecting patients for WGS, facilitating consent and test ordering are valuable as they can inform the development of recommendations to optimise the delivery of WGS within the NHS (Table 4). Our finding that clinicians who are not specialised in clinical genetics show reluctance to offer WGS to their patients, is in-line with findings from another sub-study conducted as part of this research programme [27]. In an interview study with those stakeholders involved in designing or implementing the GMS, the authors identified that the "mainstreaming agenda" encountered reluctance to become engaged from those who did not see it as a priority or viewed it as being political rather than clinically driven [27]. Further, both this study and that of Friedrich et al. [27] found that the onerous administrative aspects required to consent patients and order WGS were a deterrent to adoption of WGS. Time and resource constraints were related to the number of staff available to aid the process and both studies illustrated that hiring HCPs to aid the process is favourable.

Workforce development and digital revolution are both pillars of the recently published UK strategy for "Accelerating genomic medicine in the NHS" [28]. Regarding digital revolution, previous studies have focussed on the need for genomic report integration into the Electronic Healthcare Record (EHR) [29, 30]. Our study adds further to the literature on digitisation in genomic testing, highlighting the need for automated tracking of blood samples earlier in the genomic testing process, before a genomic report is even created. This is also supported by Friedrich et al. [27] and Pearce et al. [30], who found that lack of digital, coordinated infrastructure was a challenge to service delivery. Other countries are also struggling with this aspect, for example, both the IGNITE network and the Melbourne Genomics demonstration project identified lack of data interoperability in EHR systems as a barrier to genomic testing [15, 31].

Concern around patient understanding when discussing the benefits, risks and limitations of WGS has existed since the introduction of this technology into medical research [32]. Our study demonstrates that such concerns still exist among HCPs, over a decade later, despite the development of patient resources surrounding WGS. Shifting the paradigm will take time, especially as the emphasis on undergraduate training in genomics currently differs across UK medical schools (Seed et al., in press), and has begun with efforts including the NHS GMS website and Master's programmes from NHS England Genomic Education for HCPs and academics.

Our recommendations for improving the quality of the WGS process include placing genetic specialists in mainstream centres to help train the mainstream HCPs and facilitate implementation of genomic medicine in non-genetics specialisms. These could be fixed term roles or secondments from clinical genetics. Such roles are already being advertised on the NHS Jobs search, for example, a genetic counsellor role to support the National Amyloidosis clinical team, and a genetic counsellor role to support mainstream cancer care across the East of England [33, 34]. In both cases, these roles were funded centrally through NHS England, but such roles could also potentially be funded directly through clinical departments.

Strengths and Limitations

A strength of our study was that we were able to capture the experiences of clinical departments consenting families for WGS across a range of clinical

specialities across England over a 12-month period. In turn, we acknowledge that we have only covered a sample of the NHS Trusts that exist in England and processes in other Trusts may differ. We have been able to capture a snapshot of a particular moment in time during the early implementation of WGS in the NHS. Most of our findings relate to WGS referral, consent, and test ordering and not on return of WGS results as very few results had been returned at the time of conducting this study. Therefore, our findings related to this phase of the workflow mainly reflected what participants anticipated they would do. Practices may have changed since this study was conducted as this is a rapidly developing field, notably we are aware that more staff such as genomic associates are being employed to support mainstream clinicians with WGS. A significant finding was that patients in mainstream specialities have a very different experience than patients in clinical genetics. In the mainstream, the entire process tends to be handled by the clinician. This avoids potential delays of referring to other HCPs but the number of patients who are offered WGS is limited by the clinician's time constraints for consent and paperwork. For this reason, further data would be needed to evaluate patient outcomes (e.g., number of patients offered WGS per department, time taken from phenotyping to test result, etc.). This is being investigated in another sub-study from our programme of work [19]. We were reliant on principal investigators in clinical genetics to identify and approach mainstream clinicians for recruitment into the study – this was limiting. We were also unable to present all the process maps to all the participants of the study due to time constraints.

Conclusion

This study has helped us to understand the processes as well as similarities and variations in practice taking place as WGS becomes established within the NHS. Future research could look at whether and how this has evolved over time as the service embeds across the NHS. This research contributes valuable insights that can guide policy and practice, providing strategies for improved integration of WGS into routine clinical care.

Statement of Ethics

This study protocol was reviewed and approved by the London Bloomsbury Research Ethics Committee, Approval No. 21/PR/0678. Written informed consent to participate in the study was obtained from all adult participants and all underaged participants' parents.

Conflict of Interest Statement

The authors have no conflicts of interest to declare.

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Author Contributions

C.L. conceived the study and collected the data; N.M.L. and C.L. designed the analysis; N.M.L. performed the analysis and wrote the first draft of the manuscript; and A.C., C.P., A.P., M.H., S.W., and C.L. supported interpretation of the analysis, read and revised early drafts of the manuscript, and approved the final draft for submission.

Data Availability Statement

The data that support the findings of this study are not publicly available due to their containing information that could compromise the privacy of research participants, but are available from the principle investigator C.L. (celine.lewis@ucl.ac.uk) upon reasonable request.

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