Genomic Health Literacy Among Patients in Ireland

FINAL REPORT

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Prepared for

The National Genetics and Genomics Office

Health Service Executive

By Amárach Research

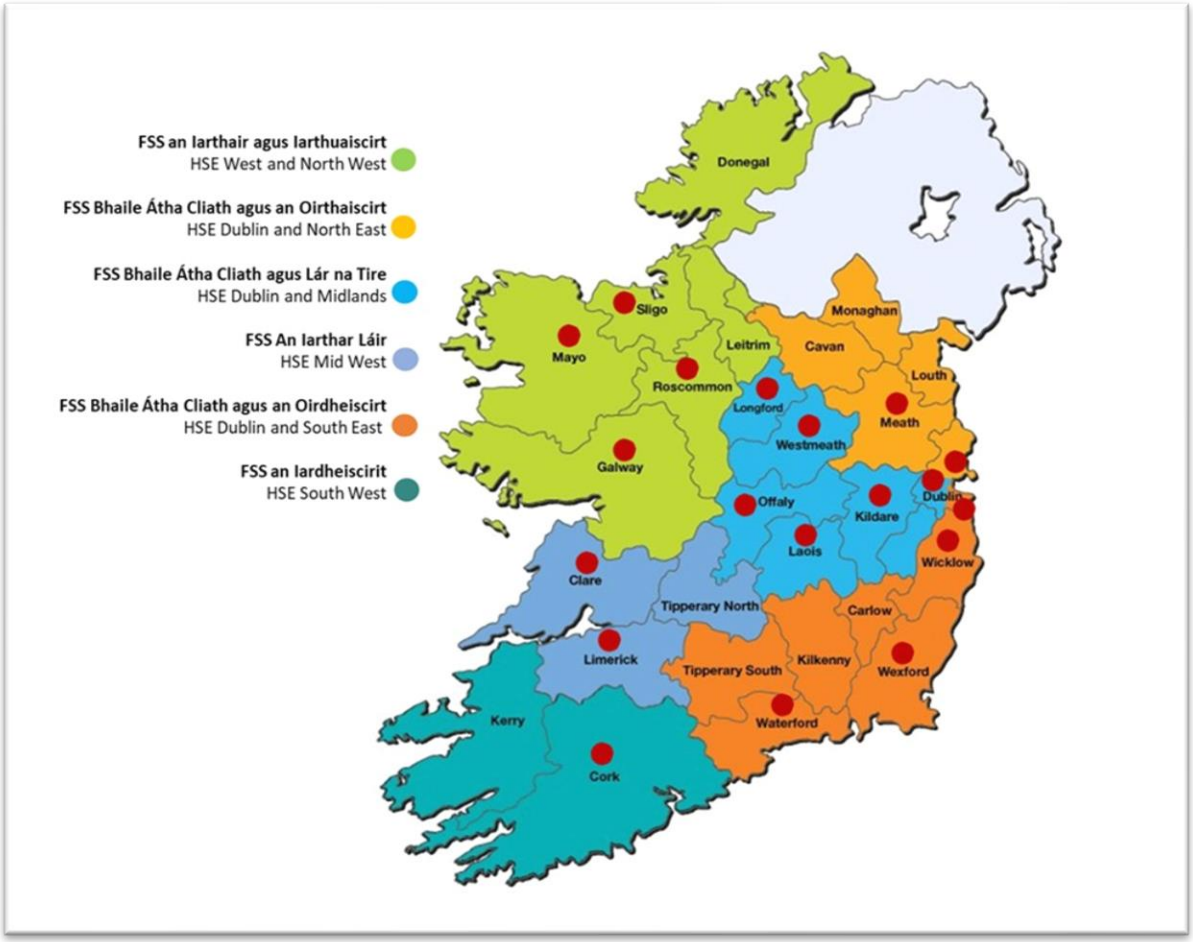
Private & Confidential

Contents

1 Methodology, Sampling & Aims/Objectives

1.1 Participants overview

- **37 research sessions** took place, 8 of which were focus groups of 80-90 minutes duration, and 29 were hour long in-depth interviews. Interviews and focus groups were conducted between 13th February and 7th March 2024.
- The fieldwork has generated more than **40 hours** of discussion with **63 individuals.**
- Participants were screened and recruited through a variety of approaches and engagements. Amárach managed this centrally using online surveys and telephone recruitment / confirmation calls to qualify prospective participants.
- All participants were resident in the Republic of Ireland.
- Each red circle indicates a participant(s) county of residence.



1.2 The sample

- This sample includes people who have:
  - Experienced genetic testing because of being diagnosed or **at risk of a 'rare disease' genetic condition** (which includes those with an inherited cancer predisposition condition) – adults and parents of children affected.
  - and / or **cancer, not related to a cancer predisposition condition** – adults.
- It also includes people who **previously accessed genetic and/or genomic testing in pregnancy**, including for some in conjunction with the above.

No. participants	***Circumstances represented:*** *(note some participants experienced testing for multiple reasons)*			
	Adult rare disease	Adult cancer	Adult pre-natal	Parent rare disease
63	**16**	**22**	**5**	**25**

1.3 Research aim

The aim of this research is to provide insights into the information needs of patients using genetic and genomic services at different stages of their journey and to identify how best to address these needs.

1.4 Specific objectives

Two key questions are posed:

- How can the HSE increase health literacy of patients accessing genetic and genomic information and services?
- What do those who have been offered a genetic or genomic test and their carers need to know before having a test and how can the HSE address their information needs to help them better navigate and have an improved experience of using genetic and genomic services?

2 Research Findings

2.1 Context - route in to testing

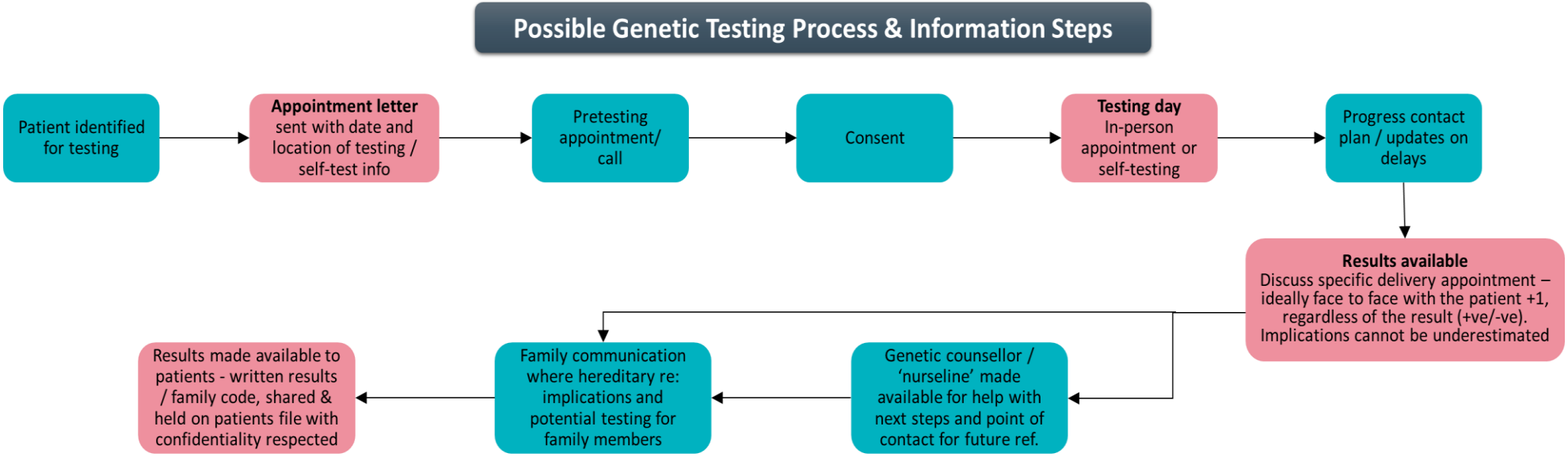
- **Importance of patient journey/route into genetic testing**
  - It was evident at an early stage of the fieldwork that the nature of the patient journey and route to testing was of importance in discovering and categorising patient/carer information needs and identifying the ways in which these are currently addressed, or not, and suggestions for potential solutions.
- **Routes into genetic testing**
  - The research heard how patients entered their genetic testing journey through a variety of routes. Patients shared the background on their routes into testing, including but not limited to:
    - **Consultant / medical team / other medical professional referral**, such as G.P. **into genetics counsellor / geneticist for** pre-testing explanation, consent and testing process.
    - Testing and associated explanations, consent and paperwork completed **directly via a specialist consultant/clinician's office.**
    - **Self-referral** into the service when hereditary information comes to light – some may have emerging symptoms of concern, others without symptoms but **concerned about the potential to inherit** a condition / take preventative action.
    - **Self-referral linked to concerns around symptoms**, as yet undiagnosed condition, and potentially after undertaking their own research and investigations and reflecting on family history or contacting an advocacy group - piecing together a jigsaw independently.

2.2 Context – journey stages

- **Headline steps in a journey may include:**
    - **Genetic / genomic testing first arises / becomes relevant** i.e. the first point at which the need / relevance of genetic / genomic testing is raised or connected to the circumstances. This may be in conjunction with an **appointment** for further discussion and/or testing, or whilst waiting for these.

- **Reflection & consideration** - background information needed about genetic testing in general, and in relation to individual circumstances, and information to help prepare patients for the journey ahead. This may be prior to an initial appointment and/or during a period when the patient is not yet connected/fully connected with the services.
- **Initial connection with services** - preliminary discussions or meetings and/or information provision in relation to forthcoming genetic testing.
- **The testing appointment / procedure itself.**
- **The analysis/lab testing process** and **waiting period**, including delays to results.
- **The results phase** – delivery, implications, information & understanding.
- **The next steps** – further testing (loop back to the above) / treatment paths / information sharing with family / support routes / no further action:
    - For family sharing how is this communicated, who is there to help with testing for other family members, what are the triggers that would suggest family testing is required, with children is there an age limit and what is this?
- **Future implications** for patient and / or family members

- **In terms of HSE / genetic services touchpoints**, generically this creates several steps, some of which may take place independently, others together within one patient visit / interaction. Some are by-passed, depending on circumstances and routes into the services.



2.3 Additional Layers

- Several additional considerations play a role in communications requirements:
  - Nature of symptoms / condition – rare disease, cancer, treatment, life limiting, terminal, age - adult, baby, child.
  - Testing recommended by health care provider or requested by patient.
  - Implications of results for the individual and / or the wider family.
  - Experiences, relationships and information within the family prior to genetic testing.
  - Information available on family history – known & documented or pieced together.

2.4 Patient literacy prior to genetic/genomic testing

- The research observed **two main cohorts in respect of patient literacy levels** coming into the genetic testing services:
  - **Very limited, or non-existent literacy, knowledge, understanding** – primarily those referrals to genetic testing where an unexpected health condition diagnosis has been received or symptoms are under investigation / diagnosis is inconclusive. Research participants were very clear that they had entered the service totally unprepared for what they described as information which was extremely complex and new to them.
  - **Some knowledge about certain aspects of testing and terminology** because of other family members having been through testing or where a hereditary health condition has come to light and the patient has undertaken their own preliminary research.
  - Once testing became a part of their healthcare journey, **patients / carers had an immediate need for general information** on the process, specifically:
    - *what would be involved?*
    - *where would they need to go for the test?*
    - *who would they see during the testing appointment?*
    - *what would be involved and how long would it take to be tested?*
    - *how long would the wait be to receive results, and how would they be provided?*
    - *what will happen when they get the results?*

- *what might the results mean for their family?*
- *who could they reach out to when questions came to mind?*
- Nature and format of information prior to testing:
- Most people wanted to read-up on genetic testing in a general sense as soon as the need for an appointment was introduced – ***'What is genetic testing/What are genetic services?'.***

2.5 Testing Stage Information & Resources

- The next level of information at an early stage in the genetic testing experience is for **greater detail to be made available for patients/carers to refer to and explore in their own time.**
  - Patients need this information and guidance at different points in their journey, so flexibility in accessing is important.
  - Patients described anxiety over their health / health of loved ones and how they are under pressure of time to rapidly expand their knowledge and vocabulary.
- **Huge appreciation of the support of some excellent genetics counsellors.**
  - They described how the counsellor held their hand, guided them through the process and walked with them through the journey.
  - There appeared to be an element of confusion over the nature of this role so clarity pre-appointment/contact would help to set realistic expectations.

- For some patients, **coming into the testing appointment armed with detail on the process would help to reassure and inform their questions** to the medical professionals they meet.
  - Time with the consultants is often short and occasions are limited.
  - For **others, it may be overwhelming in advance, but they would potentially need a place of reference immediately or soon after the testing appointment / procedure.**
  - Patients said there can be a lot to take in and it is often only later that questions form.
  - For all, at a variety of stages:
  - An **overview of terminology** – referencing information such as purpose of and type of testing, and for some circumstances, test types (names).

- **Locations where testing takes place**, and where labs are located, **and why** these may include labs overseas and are relatively limited in their number. Stating facts, rather than as an 'apology'.
- **Typical time frames involved**, and explanations associated with any typically extended or known lengthy time frames.
- **Typical job roles or personnel involved in testing process** – primarily 'what is the role of a genetics counsellor', and if this is not for emotional / psychological support, where would that come from if needed?
    - **Signposting to advocacy/support groups** and charities where / when it becomes appropriate within a diagnosis process.
- **Clarification around consent** forms, explanations of their contents, what is required of the patient/carer, what assurances are in place with regard to confidentiality?

2.6 Consent, Confidentiality and Data Storage

- Whilst in the main participants did not raise these matters spontaneously, when the topics were introduced to them, the most frequently occurring response related to a vague recollection of signing papers and very little else.
- The participants gave the researchers the impression that whilst these are clearly important issues, there was an element of trust in the system and that forms would be 'standard'.
- For most people, the importance of the test being carried out urgently and the heightened emotions, worry and stress of trying to take in all the appointment information and their future implications superseded the importance of forms within their recollection of experiences.

2.7 Results Stage Information and Resources

- There were **very mixed experiences of receiving results**, or in some case, not receiving results.
- Taking the absence of results first.
    - Some participants, those living with very rare conditions, and/or over a long period, explained how genetic testing had been undertaken on the advice that it was 'part of a bigger research project', so in these cases patients didn't actually receive their results.

- Others reported receiving information suggesting that they had a choice whether or not to receive their results (fears over impact on insurances etc.).
- Other examples where results had been lost, or where updates on delays, or new data related to their testing had not been provided.
- For some, after a long wait, tests had to be repeated when data could not be located, adding another 6-8 weeks to the waiting times.

- The critical point people make in discussions is the **lack of information about why the wait is so long**, and the **absence of a central system to check on progress or hear about delays.**
  - Overall the lack of communication during this process, coupled with limited explanations about the process and the paths the testing may take, create and increase worry, stress, anger and for some, a lack of faith in the system.
- Developing a **consistent**, circumstance appropriate, respectful, **empathetic** system for the delivery of results is something participants requested on multiple occasions.
- Whilst some people described positively their consultant or a genetic counsellor **meeting with them, taking time, explaining, providing referral routes, materials and resources and advice**, in the various research sessions many examples were given of results being delivered in ways patients felt on reflection that were inappropriate:
- The **risk of providing results by post or phone call**, or delegating to admin staff:
  - Open to misunderstandings and misinterpretations of technical information, no obvious route given for subsequent Q&A, referral to 'Dr. Google'.
  - Delivery of results in clinical settings were reported to be inconsistent:
  - Whilst **face to face delivery appears to be the most appropriate route** from the patient's perspective, the approach to this **requires consideration** – time, travel, costs, accompaniment and overall the **implications of the result.**
  - Although a lot of those receiving results were able to **get answers to their questions at the test results appointment**, some were left unsure of what the results really meant. Not **having the right people 'in the room'** to help them answer their questions meant they went away with queries and were unsure where to go next with these.
- We learned **'do not always assume a result which did not detect a genetic issue is a positive.'**
- Difference between consultants/clinical medical staff vs geneticist / genetics counsellor.

- There were mixed experiences across all clinical results deliveries – some very positive, some less so. The gaps appeared to sit within:

    1. **Understanding the role of a genetics counsellor.**

    - Some inconsistency in the approach to counselling – one person had what she was told was a counselling session, but she said she felt it was more of an admin person, others expected psychological support, and some hoped for a genetics explanation, as described above.

    2. **The 'next steps' aspect of the results delivery.**

    - Being told what can be devastating news, without further discussion of a plan or follow up appointments, or **giving time to digest information** and ask questions, within the same visit, can be extremely difficult.
    - **A staged plan or multidisciplinary team**, each with different roles to play and information to convey could be what the results session and follow up looks like in an ideal world.
    - Being given a **'next steps' plan** once results are received is important. Patients were left not knowing what the next steps are and what it means for their families.

- **Follow up opportunities for Q&A.**
    - Once a patient returns home and considers their results and wider implication, conducts their own research or reviews materials provided at the results appointments, **opportunities to refer back for advice and information** are equally important.
    - **GP records updated.**
    - Linking back to their own GP to have results information added to notes is another key step. For some, especially when needing children to be tested at a later time, patients / parents have to go to GP to start the process and experiences were that the GPs are not always aware of previous testing.
    - **Explanations where tests are returned with inconclusive results.**
    - If tests come back with an 'inconclusive' result what may happen? Within the results process patients felt greater explanation could be provided where results are inconclusive.
- **Positive patient / consultant relationship.**
    - The research heard of examples where the patient felt their relationship with their consultant was mutually beneficial, particularly where the patient or their child had a very rare condition and there was an exchange of data and research from both sides.

2.8 Differing Needs

- There are some differences in needs according to the route into genetic testing, however standard material hosted centrally and shared within letters / at appointments would reassure and inform all entering and engaging with the services, at the stage they need to refer to it:
  - Patients with very limited or non-existent genetics literacy.
  - Patients with some knowledge of genetic services.
  - Patients opting into the services, due to family history.
  - Parents in family planning stage where existing children receive hereditary diagnosis.
  - Parents with young children / toddlers in critical stages of their development.
  - Testing / hereditary gene diagnosis in adults who are also parents.
  - Those requiring pre-natal testing.
  - Adults with hereditary conditions where results have implications for family members.

2.9 Summary of the range of Information Patients Requested

- **Prior to testing:**
  - Introduction to genetics, **what is genetic testing and why it may be helpful**. Definitions - genetics language, types of tests, rare diseases, hereditary conditions.
  - Before the appointment arrange a **pre-meeting or a video call** with someone qualified to explain the testing and what will be involved. **Outline the testing and what will be involved** and explain the timings and next steps.
- **Testing & Results:**
  - **The process** - steps involved, locations, what the tests involve i.e. simple blood test or other elements, self-testing routes, types of tests, what each looks for, where they will be analysed, what's involved and typical time lines and why it may take some time. **Updates on progress / results available.**
  - **Access to results** – for own records, for evidence linked to support routes, for a second opinion including outside of Ireland or privately – some not able to get hold of results to start this process, others have full disclosure with explanations.

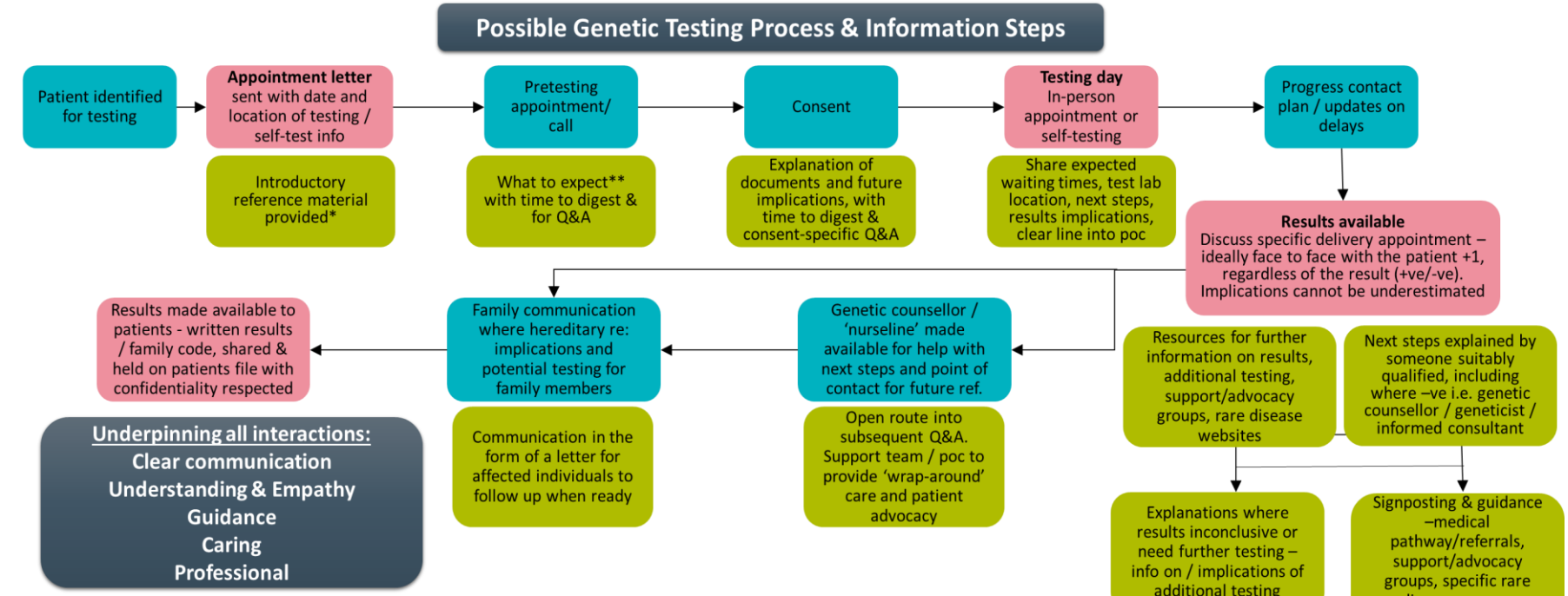
- **Waiting for results** – **named point of contact** for updates / questions – someone to be able to call for support and info – practical / technical / emotional. **Explanations around time taken** for testing to results. Role of the labs and differences, time frames, processes.
- **Who will answer questions** - practical / technical / emotional support. **What can be expected as follow up / what next steps are available.** Importance of face-to-face delivery / situation results are delivered in, support / accompaniment / post-appointment stress and worry implications.
- **Consideration of a negative or inconclusive result – may still have implications.** May still need to be delivered face to face and result in questions – not by phone/letter/consultant's secretary.

- **Role of genetics counsellor, geneticist, other health care professionals:** clarification on role, who to expect or reach out to when, functions, expertise, processes. Allocated time with these roles.
- **Opportunities to speak to someone after the results have been digested** - nurse specialists, geneticist. Helpline. Within the results appointments and / or afterwards. People are not always able to ask all the relevant questions when results are delivered. Time with professionals to ask questions in a healthcare setting, have information explained and receive support.
- **Signposting to advocacy & support groups, nationally / internationally.** Resources - emotional and financial support as well as care, and family related. For some, a diagnosis relates to the next steps in managing a long-terms condition and accessing support, e.g. documentation to declare someone 'registered blind', or for special educational needs support, or to access financial supports or seek **global support.**
- **Resources for those with family planning questions** – specialised information ref genetics and options available to parents, headline science - ivf, excluding 'problem' gene, for current and for future generations.
- **Genetics specific 'headline science' education and information** – relevance and implications within the info from a consultant / specialist regarding a health condition.
- Acknowledging that some people will access private testing: **Information about any implications of private testing** – there is a perception that, following private testing, it may be difficult to return to the HSE for care / preventative surgery.
- **Information on disclosures / implications** for family members where hereditary, for future health insurance, life cover etc.

3 Recommendations

3.1 Suggested Roadmap:

Taking the generic steps within the process, and considering the most critical information requirements:



* **Introductory reference material – may include:**

- Overview of testing stages/process – leaflet / website.
  - ❖ Terminology commonly used in genetic testing.
  - ❖ Types of tests & reasons for use.
  - ❖ Roles involved e.g. of genetic counsellor.
  - ❖ Points of contact.

** **What to expect at testing appointment and results stage:**

- Test appointment day - what will happen, who will be involved, what are their roles, how long will it take?
- Results - how will results be delivered, by whom, what are the potential routes for results & their implications?
- Next steps - provision of subsequent 'wrap-around' support, referrals, information, advocacy/support groups.
- What not to do and why (i.e. advisory *"avoid 'Dr Google' – use this site instead…."*).

Note: In some cases, patients had been asked to undertake their own testing and return the sample to the lab by courier. These scenarios require precise and reassuring instructions and ideally a named contact / helpline option to assist with the testing process/queries.

3.2 Platforms/ Sources/ Considerations

- **Overall, there is a call for a consistent process,** to be followed across the HSE, in all healthcare settings.
  - References were made to the genetics services creating a process as per standardised materials in place for surgery preparation or cancer care pathways.
  - **Communication touch points ought to align with the patient journey** and can be grouped:
  - **Hard copy materials**
    - appointment letters – details to include who, where, how
    - intro leaflets/pack – what is genetic testing, process, terminology
    - what to expect info, what routes might testing take
    - where to find out more leaflets
    - infographics/process flows/road maps
    - FAQ, who to contact
  - **Online resources - hard copy materials to be online also / use of QR codes**
    - easily accessible and digestible webinars
    - videos of professionals providing guidance
    - recordings of patients sharing their perspective and experiences
    - HSE website – clear routes to definitions and overview of services
    - links to additional resources – links to other sites, support/advocacy groups, research for genetic conditions and rare diseases – an Irish focus and global resources, with 'guidance' for use
  - **Interpersonal connections** – there are a number of highly critical points in the journey where a face-to-face conversation and guidance on next steps with a single point of contact would be the ideal scenario.

- **In addition:**
  - **genetics services training** across all levels of HSE staff, from GP surgery/community staff to hospital wards to consultants, so all aware of the available resources to support and signpost. Patients have Q&A requirements throughout their journey.

- **support groups ROI** – identify and promote groups – charities, advocacy groups
    - in some cases, particularly for certain rare diseases, point out scientific research programmes, and potentially patient/parent Facebook groups
- **global support and research communities** – with clear guidance not ROI
- **Accessible Resources**

– Consider when designing materials there may be a range of:
    - **Technical abilities / resources** – not all patients have digital skills, familiarity with online access or suitable hardware at their disposal, and people may not be in a position/ready to discuss their circumstances with others who may normally assist them with online access.
    - **Additional needs** among this community. Some participants highlighted their condition specifically impacts on their sight, hearing, ability to process information, increased need for someone to accompany them etc.
    - **Recognition of the sheer impact of genetic testing and results on individuals and families:**
– Many research participants shared considerable detail with far reaching emotional and physical consequences. There was an emphasis throughout that this ought to be recognised in any communication around genetic services.

3.3 A Sample of Patient/Carer Sentiments

'Felt like treated with kids gloves due to being pregnant, seen as a priority, communicated face to face with all results explained which was excellent and helped with the worrying. Always had time for questions and where could not answer would come back to me later.'

The Genetics Services has to step up and be a leader in this space: open up conversations.'

'But if they don't do the testing, how do they know? Even when tests come back we need to have an understanding from the person delivering the results what this condition means for us and our child. I had been fighting to be tested .... my child was over 2 before the condition was seen.'

'Within the HSE system I was just left – if you don't fight you will be left.'

'Very positive experience with the genetics counsellor - really knowledgeable, supportive, professional, kind, empathetic and service very responsive when we sought the result. It isn't easy work and I felt really supported. The specific counsellor was excellent.'

'Genetics work, including testing is 'delicate', political, there are strong views and emotions - it all requires care to open up conversations and avoid unhelpful dichotomies and division. Perhaps recognising that the diagnosis of a genetic illness/carrier involves some type of loss (at various levels) and there is a type of grief at a diagnosis. Regardless of decisions people subsequently make, there is often a type of loss.'

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