ENDING THE WAIT

Actions to reduce the waiting list for Ireland's genomics service

Rare Diseases Ireland

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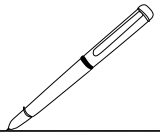
This report was commissioned by Rare Diseases Ireland with support from Roche. It was researched and written by Alistair Hodgett of Polwarth Strategy.

Preface



Among the key areas of strategic focus in the National Strategy for Accelerating Genetic and Genomic Medicine is that "meaningful partnerships with the public will be established to ensure that the public and patient voice is at the heart of implementation of the strategy and in the design and development of any new services or initiatives." This report and its recommendations are presented in response to that commitment to put the patient voice at the heart of implementation.

The development of a high-performing genetics and genomics service is of particular importance to people affected by or at risk of developing a rare disease. Genetic testing is a cornerstone of diagnosis and treatment for many rare diseases and, as we found in our Rare Reality report in 2022, in more than three-quarters of cases of people that had access to genetic testing, it was key to a definitive diagnosis.

We have further documented patients describing waiting lists as imposing a harmful delay in an already stressful situation on both themselves and their families. Delayed diagnosis exposes people to inappropriate and sometimes harmful care, treatment or surgery and denies people access to information needed to make informed reproductive choices.

Waiting lists need to be urgently reduced so that people living with rare diseases can receive an accurate diagnosis and initiate appropriate treatment and care as quickly as possible, before further deterioration in their health. The research in this report has established that substantial reductions in the waiting list are achievable by a relatively modest increase in the size of the workforce that delivers the service, coupled with technology-enabled management of the patient journey.

Our voice is loud and clear – we need immediate action to reduce risk and deliver an accurate diagnosis for people living with rare diseases. All citizens have the right to a timely diagnosis.

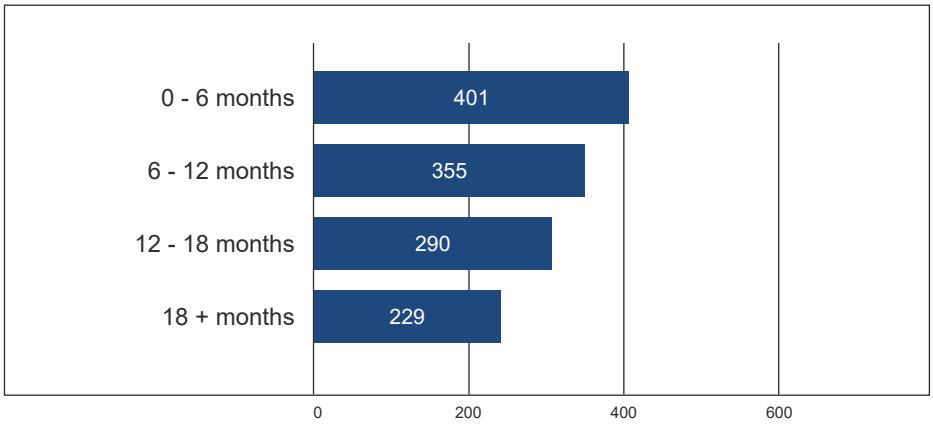
Vicky McGrath
Chief Executive, Rare Diseases Ireland
March 2023

Introduction

Realising the ambitions in the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland[i] (hereafter 'national strategy') will require addressing many issues however none is more critical or urgent than reducing the waiting list to access the Clinical (Medical) Genetics service (hereafter 'service'). It remains the most acute current pain point for both patients and clinicians and practical actions directly targeted at reducing the waiting list ought to be central to early-stage implementation of the strategy.

A lengthy waiting list for the service means that individuals experience frustration as they consult different specialists, may receive unnecessary and potentially harmful treatments, and are delayed in receiving an accurate diagnosis and beginning the most appropriate treatment. The harmful consequences of long waiting lists and delayed diagnosis also include inability to access information needed to make informed reproductive choices, inability to access life changing products such as mortgage and insurance and delaying marriage. According to recent research[iii] some patients are dying on the waiting list and as a direct consequence of their death before diagnosis, family members who themselves may be at risk also remain unaware and untested. The personal and economic costs of waiting lists for genomics services are enormous and potentially affect significantly greater numbers than the number of individuals on the waiting list.

Figure 1: Waiting List for Clinical (Medical) Genetics service, February 2023



Waiting list data reported by the National Treatment Purchase Fund includes both adult and paediatric cases at the core public genetics service at the Department of Clinical Genetics at Children's Health Ireland, Crumlin. The waiting list figures do not include individuals waiting for an appointment with a genetic counsellor or those accessing services through standalone services for cancer, ophthalmology, perinatal or other specific areas. https://www.ntpf.ie/home/pdf//2023/02/hospitals/out-patient/chi.pdf

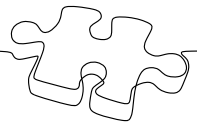
This review of services in Ireland and comparison to services in peer countries finds that there are opportunities to take actions in the short-term – over a period of 2 years or less – which will achieve meaningful reductions in the time patients spend on the waiting list for services in Ireland. Waiting lists are a persistent and acute issue – with waiting times for routine cases as long as two years - and likely to be a recurrent challenge as demand from patients and clinicians[iv] grows. Referrals have grown at 10 per cent per year since 2020 (rising from 3,230 in 2020 to 3,629 in 2021 and 3,903 in 2022) and implementation of the planned public and clinician awareness programmes included in the national strategy will potentially accelerate this rate of growth in demand. Studies have also observed a sustained trend of increasing time required for the management of both new and review patients driven by increases in the complexity of diagnostic testing and investigation[v] . Improving the ability of the service in Ireland to meet rising levels of demand and complexity is therefore an imperative in both the short and long-term.

A genomics service that seeks to realise it's clinical potential, provide a quality experience for patients and pursue an efficient allocation of resources should have waiting time as a key measure of performance. The services of peer countries reviewed in this report have achieved wait times of between 3 and 12 months and on the basis of the insights from these services, Ireland should consider a wait time of 12 months or less as achievable in the near term. A further reduction in wait time to 6 months or less, considered optimal on clinical, safety and efficiency grounds[vi], will be achievable through sustained prioritisation and resourcing.

The recommended actions to achieve this significantly reduced wait time are:

- Address under-resourcing by accelerating recruitment and expanding the target workforce size to add the additional staff needed based on benchmarks.
- Eliminate further bottlenecks and inefficiencies in the patient journey through supporting front-line team innovation, including providing a budget earmarked for solving known bottlenecks and inefficiencies such as duplicate referrals.
- Reduce the burden of paper-dominated administration by providing front-line teams with access to interim and long-term technology solutions that improve management of patient referrals and other routine processes.
- Develop cross-border collaboration with Northern Ireland with a view to providing an all island genomics service.

Implementation of the National Strategy

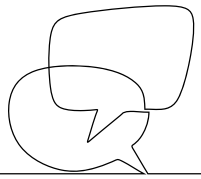


The National Strategy for Accelerating Genetic and Genomic Medicine in Ireland sets out a "*vision for Ireland's future genetic and genomic service*" and includes a number of commitments to implement the strategy. The implementation will be driven by a national office for genetics and genomics to be established in 2023 which will work to develop a five year plan that will "*set out the actions, deliverables, and responsibilities to progress this strategy's implementation. It will define short, medium, and long term goals that can be initiated and implemented within the strategy lifecycle while working towards the achievement of the longer term vision*."

The urgency of short-term actions to improve the current service is acknowledged in describing the role of the national office as "*deliver(ing) service improvements in the short term through the implementation of the funding commitment in the National Service Plan 2023 to address urgent service deficiencies*", adding that "*the initial funding of €2.7 million will be directed towards the establishment of the new national office with the associated recruitment of roles, as well as towards addressing gaps in the frontline workforce in relation to clinical geneticists, genetic counsellors, and genomic resource associates.*"

Implementation of the strategy will need to overcome barriers common to other health strategies as well as those specific to genomics. In a contribution to the strategy document from the patient representative Karen Morgan, she notes "*My hopes for patients and families when the strategy is implemented are…that we have easier access to genetic services in Ireland … and a reduction in test waiting times*." This report seeks to provide practical recommendations for specific actions which can be taken in the short-term which are i) capable of achieving meaningful reductions in waiting times; ii) derived from the insights of clinical geneticists in other genomics services; and iii) consistent with the long-term development of the service in Ireland. It is intended as advisory to the leadership of the national office and other key stakeholders, providing a basis for engagement with patient groups and others in pursuit of addressing this aspect of service performance.

Approach to research

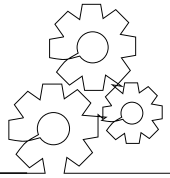


In order to examine the core public genetics and genomics service, the scope of the research excluded standalone services dedicated to cancer, ophthalmology, perinatal or other specific areas. As referral volumes in a therapeutic area increase and diagnostic pathways are defined there is a natural evolution of distinct but linked services for cancer and, depending on the stage of development and configuration choices in a particular healthcare system, for other therapeutic areas. The core service, as examined in this research, remains occupied with the substantial volume of rare and undiagnosed diseases.

A series of interviews were conducted with clinical geneticists in six countries, each of whom was either active in their respective genomics service or had direct experience of the development of the service[vii]. Semi-structured interviews were conducted over the period October to December 2022 in parallel with a review of relevant documentation provided by interviewees. All interviews were conducted on the basis of non-attribution in order to allow interviewees to speak candidly about their respective services. The expert contribution of these clinical geneticists is greatly appreciated; their insightful contributions on the topic of the practical implementation of a national genomics service exceeded the scope of this report.

Interviewee insights were reviewed to identify a set of practical early-stage actions that impact on patient access. While services develop in the context of their respective wider health systems, and different choices have been made in their configuration in the health system context, all have sought to ensure prompt access. This need is not limited to one stage of the development of the service; as a genomics service evolves and the volume of annual referrals increases, it requires sustained action to maintain good access and mitigate against increases in waiting times. Such actions remain relevant to the ongoing management of the service over the long-term.

The drivers of access to a genomics service



The capacity of a genomics service to serve the population at any given level of demand depends on its resourcing and configuration[viii]. The three elements most directly related to improving patient experience and access - the patient journey, the workforce, and information management - have been reviewed here in order to provide context for the recommended actions. Selected examples from other genomics services have been cited to illustrate the practicality of implementation and, in some cases, the extent of the impact on waiting times.

Patient journey

The patient journey through a genomics service consists of the stages from referral and assessment through sample collection and testing to diagnosis. At each stage, there are opportunities to introduce small efficiencies and minimise re-work, reducing the average time required for the management of each referral. With approximately 50 identified sub-tasks for the service across different stages[ix] , some of the tasks can be executed together in order to reduce the total number of interactions – such as repeat appointments - required between individuals and the service. For example, information collection and consenting can be combined into a single appointment.

Interviewees described active management of the patient journey as time-intensive but yielding measurable benefits in reduced waiting times.

Some of the approaches used by other health care systems were targeted at continuous improvement of the triage process and others involved larger system design decisions.

In **Northern Ireland**, a dramatic reduction in waiting list time – from over 60 weeks to an average of approximately 12 weeks – was achieved in the midst of the COVID pandemic. This was primarily achieved by enabling improved visibility – a transparent patient journey – for the team delivering the service; this visibility of the patient journey enabled the team to develop and implement targeted interventions at ‘bottlenecks’. These interventions include employing virtual consultations and re-triage as noted in other sections of this report. This was achieved without additional staff resources, albeit starting with a higher level of staffing than is presently the case in Ireland.

In **Australia**, Queensland’s use of remote counselling via telehealth video conferencing allows patients to attend their initial assessment, complete the consent process and initial information gathering without significant travel time to a major hospital centre. Patients welcomed the non-clinical interactions being managed by telehealth. Appropriate use of telehealth was balanced with clinical interactions remaining in-person, with these equating to 20-30% of interactions with patients. Clinicians reduced the travel burden on patients by conducting in-person appointments in 9 satellite locations.

In **England**, the NE Thames service uses referral criteria to ensure that certain conditions (such as sickle cell disease) are promptly re-referred to specialist services, while others (conditions such as Alpha-1 Antitrypsin Deficiency or familial hypercholesterolemia) are re-referred to the GP to order the initial test. These referral criteria also contribute to the mainstreaming of genomics in the healthcare system and the development of an accurate picture of the nature of demand by therapeutic area, aiding planning and resourcing of different related services.

Workforce

The workforce directly engaged in providing a genomics service is a mix of consultants, counsellors and support or administrative staff.

Workforces which have acquired a higher ratio of counsellors and administrative staff to consultants are able to achieve a more efficient mix of capacity. A larger team of counsellors and administrative staff enables the tasks required for the service to be executed at an appropriate level of expertise. Where counsellors and administrative staff predominantly undertake more routine and time intensive tasks such as initial patient contact, appointment logistics, and liaison with test providers, this enables consultants to dedicate increased time to medical examination, management of complex cases, making diagnoses and providing clinical leadership to the team and colleagues.

The impact of a larger non-consultant workforce includes direct improvements in the quality of the service. A 2020 review of the composition and capacity of workforces also found that "genetic counsellors have been shown to play an important role in utilization management through patient identification and triage and through reviewing genetic test requests in a laboratory setting, resulting in a reduction in inappropriate testing."[x]

In **England**, the NE Thames service has sought to ensure that a range of tasks are managed by managerial and administrative staff, notably consenting and the paperwork surrounding whole genome sequencing. Family history coordinators and genomic associate posts are also in place to assist in this delegation of tasks from consultants, and there is believed to be further potential to expand these roles.

In **Australia**, a 2019 review of workload and workflow studies[xi] found that "clinical throughput is limited by the time taken to complete direct patient/provider interactions and patient-related activities (PRA), a measure of indirect clinical and administrative requirements" with patient-related activities representing a large majority of total workload. The review recommended that services adopt a number of metrics and highlighted the value of metric collection and benchmarking to adequately resource services to match growing clinical demand. A key recommendation was to capture and benchmark the number of full-time equivalent clinical geneticist and genetic counsellors involved in direct patient care per million catchment.

Some interviewees attributed lower than desired counsellor staffing levels in their services to constraints on supply, such as a lack of domestic training of genetic counsellors, or regulatory failures such as lack of approval for the counsellor profession. Both the training and regulatory issues are cited for action in Ireland's national strategy.

Relative staffing levels across the workforces of some of the reviewed

services are examined in the next chapter of this report.

Information management

Information management encompasses the wide variety of information captured and used to deliver the service. Key to improving access and reducing wait times is the maintenance of accurate and up-to-date records on each patient as they move along the patient journey in a system accessible to those staff who provide the service. Systems in which information is manually transferred such as forwarded emails, re-typed reports or printed letters lose significant resources in the form of the time of clinicians and other staff that gets diverted into these processes. In Ireland, the 2021 paper "Duplication of referral, a tsunami of paper: how much does it cost the Irish health services?" found that re-referrals, in which a referring clinician submits duplicate referrals to the genomics service, diverts consultant and administration time away from other patients.[xii]

In **Queensland**, improvements were achieved though solutions – both platforms and digitalisation - implemented ahead of the rollout of comprehensive solutions. Queensland implemented Trakgene[xiii] enabling the collection and access to family data and appointment booking and management; this precursor system, capable of integration with other hospital systems, also allowed the benefits of electronic health records (EHRs) to be achieved more rapidly once in place. The service also ensures that information that arrives in non-digital format (such as referrals via fax) is digitised at the point of arrival and all internal information storage and management is digital.

In **England** the NE Thames service described the hospital-wide electronic patient record system as among the "the best investments" in support of the genomics service; it includes giving patients direct electronic access to letters, appointments and test information (Epic was introduced in Great Ormond Street - which houses the NE Thames service - as the electronic patient record system in 2019). The service also benefits from a requirement for all GP referrals to be made electronically using the NHS e-Referral Service[xiv] with all non-GP referrals made via a dedicated email.

In **Northern Ireland** the NIECR (Electronic Care Record) brings together patient data including letters, appointments and lab results and allows clinical decisions to be made at the point of first triage, for example the proactive scheduling of sample collection to coincide with pre-existing appointments.

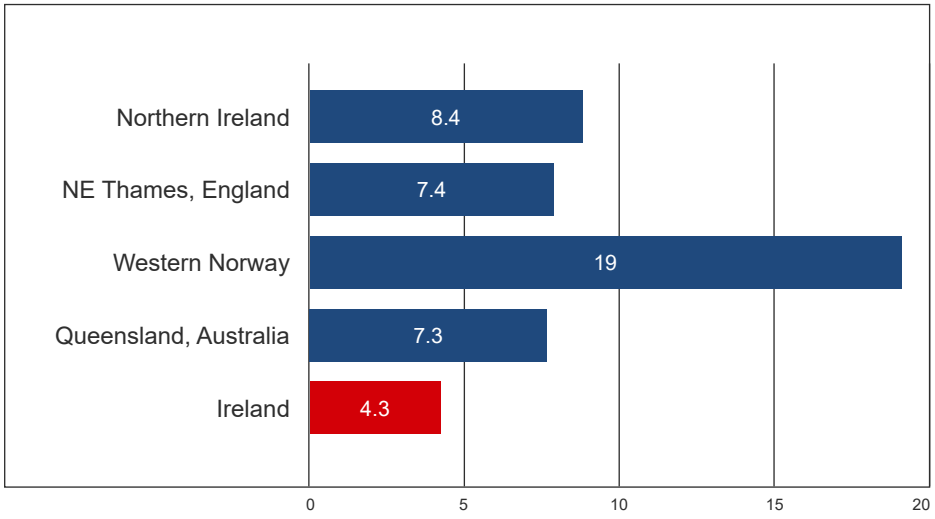
Management of the flow of information to and from test providers and external laboratories can also generate delays and rework that contributes to inefficiencies and increased wait times. Managing multiple test labs with variable reporting (for example, omitting required detail) creates a large administrative burden on the genomics workforce. The development of a panel of consolidated testing providers who conform to a common standard of reporting can aid in eliminating the need to regularly request additional information from providers. The management of laboratories can also be designed to achieve efficiencies that translate into reduced waiting time.

Staffing levels

To determine the minimum staffing level needed in Ireland's core genomics service, data was gathered on the workforces of several of the reviewed services. As an indicator of performance on patient access, interviewees also provided an estimate of average waiting time for the primary clinical consultation (see data table in Endnotes).

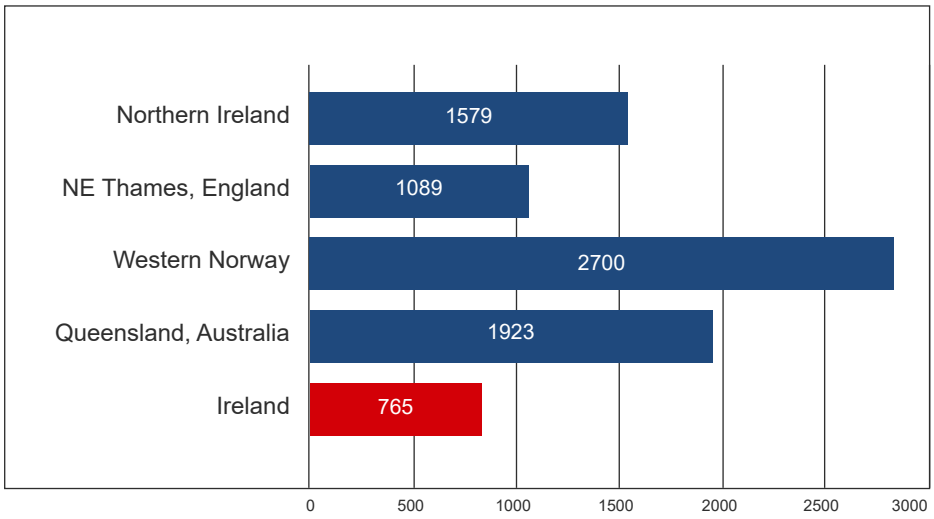
At the current level of 4.3 staff per million population, Ireland's service is staffed well below the level of other services (see Figure 2). While all of the reviewed systems had significantly higher levels of staff relative to their populations, several interviewees stated that they considered their own services to be under-staffed a current levels despite these services benefitting from fully electronic records and other resources not available to the service in Ireland. Western Norway is notable for a significantly higher level of staffing than the other reviewed services which has contributed to a capacity for a high volume of annual referrals (see Figure 3).

Figure 2: Staff per million population



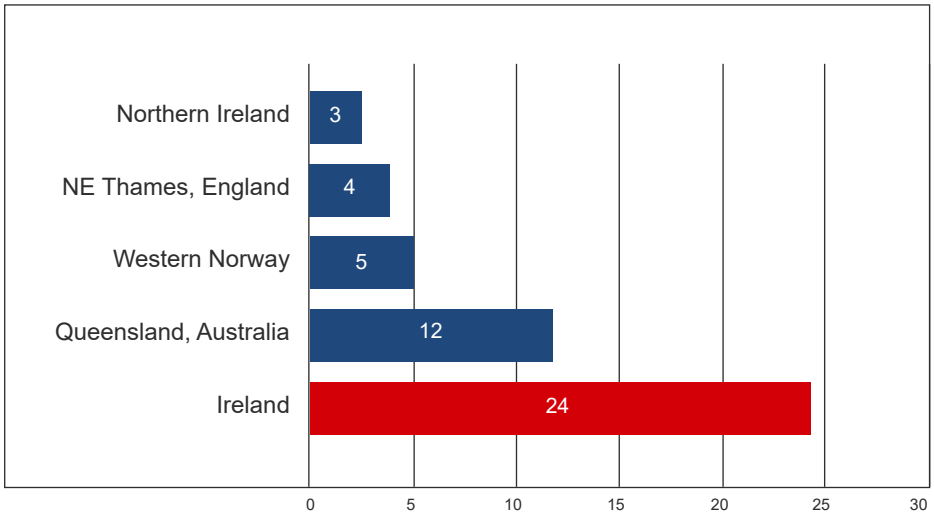
Ireland's service is receiving relatively low levels of referrals at 765 per year per million population (see Figure 3), which may be indicative of i) a reluctance by referring clinicians to add an individual onto an already long waiting list; ii) a larger latent demand which is not being met; and iii) clinicians electing to bypass delays in central genetic testing services and send tests directly to international genetics laboratories. The latter may inadvertently contribute to duplicate referrals and waste, and fail to provide adequate expert support for the referring clinician when the test result is received.

Figure 3: Annual referrals per million population



Currently the waiting time for a first clinical appointment is 24 months (see Figure 4): twice as long as the closest reviewed service. The impact of an under-resourced workforce is that without the capacity to see patients, they are added to a waiting list. Over the years, the shortfall in the workforce has resulted in a waitlist as long as 36 months.

Figure 4: Wait for first appointment in months



In public comments[xv], the HSE has stated that recruitment is planned for nine roles comprising five genetic counsellors, two genetic coordinators and two consultant geneticists; when completed, this would bring total staff to 31 and improve the ratio of staff per million population to 6.1; this would however still leave Ireland with a staffing level below any of the assessed systems.

Notes on workforce data

Consistent with the scope of this report, this data excludes staff involved in distinct services (for example in cancer) as well as staff in laboratory services: this ensures that the data, as far as possible, is comparable across systems. Variations in the different roles in different systems[xvi] mean that titles may not reflect those used in this report. The workforce numbers are reflective of the systems at time of publication, and roles currently vacant are not included in the count.

Recommended Actions

The recommended actions to reduce the waiting list for genomic services in Ireland were selected based on their relative ease of implementation and their near-term positive impact on access to the service. All the reviewed services had higher workforce ratios, more resources for patient journey management and better access to digital information tools. The current momentum for change in genomics services in Ireland provides a favourable context for implementation of these actions, and the large number of people on the waiting list for genomics services requires an urgent response.

1: Address under-resourcing by expanding the target workforce size and accelerating recruitment

Increasing the number of staff is essential to achieving improved access for patients. To serve the Irish population, the workforce needs to be approximately doubled. This would bring us on a par with Northern Ireland staffing levels of 8.4 staff per million population. Even if we target the lower staffing levels similar to those of England (NE Thames), then in addition to the nine roles already committed to we need an additional seven roles.

Given the well-known challenges in recruitment across all sectors, and the specific constraints on the supply of staff with the requisite experience and training in genomics, delay with recruitment is the greatest risk to the development of the service. Additional measures will therefore be needed to ensure that the size of the workforce increases in the near term. These measures could include the pursuit of a number of parallel initiatives such as targeted recruitment of qualified Genetic Counsellors from abroad, specification of the role and recruitment for the new genomic resource associate (GRA) positions and the recruitment of additional non-clinical administration staff. The pool of applicants should also be widened as much as possible by avoiding writing overly-restrictive role descriptions, for example requiring specific disease experience for roles. Administrative roles that address key issues and bottlenecks in the patient journey should be particularly considered as candidates may be more numerous. As an example, the management of test providers and external laboratories could be a dedicated administrative role within the genomics service.

2: Eliminate further bottlenecks and inefficiencies in the patient journey through supporting front-line team innovation

Individuals delivering a service at any point in its development are best-equipped to identify areas of service development and improvement, particularly with respect to patient journey management, but often lack both the time and authority to secure these improvements. This results in a 'leak' of resources from providing patient-facing services.

Active patient journey management is central to managing the service both in the short-term and as part of a continuing balancing of resource against demand: a paper in the European Journal of Human Genetics noted that "*appropriate triage of referrals is a critical factor in trying to address demands on the service in the face of limited resources*."[xvii] Ensuring teams have the capacity for regular re-triage is an effective means of both reducing variations in triage observed across and within different services, and of matching different patient cohorts to currently available resources. Mapping and analysing the current patient journey in flow chart format can visualise the bottlenecks and allow teams to identify and, when authorised to do so, implement changes to address these bottlenecks. Discussion of case studies and information sharing with colleagues both within the service, within the healthcare system and with international colleagues on patient journey management approaches will enhance these reviews.

3: Reduce the burden of paper-dominated administration by providing front-line teams with access to interim technology solutions

A range of efficiencies are made possible by improvements in the technology that underpins patient journey management. The importance of data and analytical infrastructure is identified in the national strategy with a commitment for the national office to "*review the existing data generation, storage, processing, and analytical capacity and capabilities available locally to develop an approach to standardisation at a national level*." This stands in contrast to a current situation described as "poor IT systems preventing innovation and more prompt appointments."[xviii]

The review planned in the national strategy does not explicitly include the day-to-day operational management of the service, referrals and tests. Ahead of the planned review and the implementation of its recommendations, clinical teams should be empowered to implement interim technology solutions that specifically improve the management of the patient journey within the service and reduce wait times. For example, where clinical teams within the service have identified platforms or other tools that will improve data management across the patient journey, procurement processes and specific IT resources need to be accelerated to provide these in the short-term.

Similarly, current approved and/or

installed platforms should be reviewed to determine if they can serve as interim solutions that bridge to longer-term solutions. If current technology can be adapted to address issues in waiting list management, then this can avoid the need to procure and install new platforms. Chief among these issues is the current inability to electronically triage referrals: paper-based referral and triage is an 18th century approach to prioritising access to a 21st century technology.

The capacity of the team should allow – through addition of administrative staff with this in their role - for the digitisation of data such as referrals which arrive on paper. Currently only GPs can use e-referrals with consultant referrals overwhelmingly on paper and many lab results are accessible only as printed emails.

Digitisation of the operational data needed for an efficient service has multiple benefits. There is a clear opportunity to provide referring clinicians and patients with access to confirmation that they are on a waiting list and where they are in the journey. As noted in a recent paper on costs to the Irish health services of duplication of referrals, at present a "*referring clinician cannot confirm their patient is on a Genetics waiting list*[xix]" leading to a level of duplicated referrals and patient anxiety. The paper notes that the "*barriers are individual hospital IT systems and the need to adhere to GDPR regulations which prevent non-Genetic staff electronic access to waiting lists*." This ought to be addressable as "*other specialists clinics are openly available within (Children's Health Ireland) to a referring consultant from within the directorate*" and indeed mandated on the grounds of the rights of patients to have access to their own health information including their position within the waiting list and journey to treatment. The paper notes that "*changes to traditional referral pathways, such as online referral pathway, have been shown to improve referral processes in other subspeciality disciplines with long waiting lists*." Additional benefits in the form of reduced cancellation rates are also achievable, as well as semi-automated requests for additional information needed from referrers such as missing reports.

There are further opportunities for waiting list reduction once teams have the autonomy to implement them, such as use of 'off the shelf' videoconferencing to enable tele-health consultations where appropriate. The use of video-conferencing needs to be formally approved and encouraged in order for teams to utilise its potential. Adopting videoconferencing and other measures in the short-term can also be seen as pilots or scoping exercises for the development of long-term and scalable solutions. These solutions will be needed to enable access for remote populations as well as cohorts of patients who may have chronic disabling disorders which further complicate travel to appointments. This is particularly the case where consultations are all centralised in Dublin, as is the situation at this time.

4: Investigation of all-island collaborations

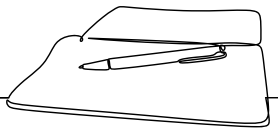
The value of serving populations across borders and of increasing the population catchment for services, was cited by a

number of interviewees. The national strategy notes that "*in line with the objectives outlined in Ireland's Shared Island Initiative, initial meetings were held between members of the Steering Group and representatives from Genomics Northern Ireland to share information and promote collaboration*." Research conducted under the Shared Island initiative called out rare disease as an area of particular opportunity, noting that "*working as a single island with a population of approximately seven million people would allow the safe and sustainable development of services that were currently not viable in separate jurisdictions. Areas identified as having potential for future collaboration were generally high-volume interventions such as orthopaedics, or niche areas that required specialist skills, such as rare diseases*".

Cross-border north-south genomic services collaboration has the potential to increase total capacity on the island, increase total population catchment and assist in reaching border populations, cross-border families and hard-to-reach groups. A combined population catchment of seven million also enables efficiencies of scale and investments on par with services in other jurisdictions such as the NHS England genomics geographies which each serve an average population of 7.4 million.[xx]

Genetics knows no borders, and with many families living on both sides of the border increased collaboration would improve care for those families. Additionally, collaboration would enable certain amounts of sub-specialisation, ensuring that the most complicated cases are accessing the best expertise available across a large pool of expert genetic consultants and genetic councillors.

Conclusion



A reduction in waiting times for genomics services in Ireland to under one year, as is demonstrated by the systems reviewed, is achievable through actions that focus on the key enablers of a more efficient service. A further reduction in wait time to six months as recommended in the Rare2030 Foresight Study is desirable and capable of being secured over time.

The recommended actions are interdependent, and ideally would be implemented together. However, improved management of the patient journey and reductions in waiting time will only be achieved with additional staff, and at a level above that currently planned.

A 2020 review of workforces in genomics captured the limitation of only addressing staff numbers in noting that "policies aimed at increasing the size of the genetics workforce are on their own unlikely to succeed in boosting system capacity enough to meet current, let alone future, demand."[xxi] However if staff numbers are increased to a level commensurate with other services and combined with other actions they will multiply each other's impact. A larger workforce with improved visibility of the waiting list and autonomy to act to address it through technology-enabled triage is a necessary foundation for rapid implementation of the national strategy.

In summary, this review recommends:

1. **Address under-resourcing by accelerating planned recruitment and expanding the target workforce size.**

a. Accelerate planned recruitment through clearly defining the role of the new Genomics Resource Associate positions and launching recruitment, and by widening the number of candidates for Genetic Counsellor positions through flexibility in the requirements of applicants.

b. Recruit administrative staff to manage activities such as digitisation of data across the complete patient journey and management of test providers and external laboratories.

2. **Eliminate further bottlenecks and inefficiencies in the patient journey through supporting front-line team innovation.**

a. Institute regular reviews to map and analyse the current patient journey to find the bottlenecks and inefficiencies.

b. Delegate authority to teams to identify and implement opportunities to address bottlenecks and inefficiencies in the patient journey.

c. Allocate a recurring budget to the core clinical team to resource the interventions needed for continuous improvement of the patient journey administrative process.

3. **Reduce the burden of paper-dominated administration by providing front-line teams with access to interim and long-term technology solutions.**

a. Reduce paper-based administration through digitisation of data that arrives on paper.

b. 'White list' a set of technologies essential to service improvements so that they can be implemented without delay by the genomics service.

c. Allocate dedicated IT resources to the service and trial new long-term IT solutions within the service.

d. Approve and encourage the use of video-conferencing within the service.

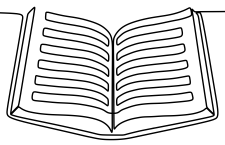
4. **Investigate opportunities for all-island genomic services**

a. Establish cross-border pathways for genomic services using resources of the National Treatment Purchase Fund

b. Conduct an evaluation of the steps required to establish an all-island genomics service that would help meet the goals of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland.

These actions are not only consistent with the lessons from other services which are further along in their development, but directly contribute to achieving the long-term ambition of the national strategy. As one clinical geneticist interviewed for this report stated: "Find your quick wins because they're very often the things needed for your big strategies anyway."

Endnotes



	Ireland	Queensland, Australia	Western Norway	NE Thames, England	Northern Ireland
Staff in core service per million population	4.3	7.3	19.0	7.4	8.4
Annual referrals per million population	765	1,923	2,700	1,089	1,579
Total staff in core service	22	38	19	33	16
Clinical geneticists	4	8	12	12	6
Genetic counsellors	11	16	5	10	6
Administrative roles	7	14	2	11	4
Time to first clinical appt after referral (months, avg)	24	12	5	4	3
Annual new referrals received	3,900	10,000	2,700	4,900	3,000
Population (m)	5.1	5.2	1.0	4.5	1.9

Endnotes

[i] https://www.hse.ie/eng/about/who/strategic-programmes-office-overview/national-strategy-for-accelerating-genetic-and-genomic-medicine-in-ireland/

[ii] Waiting List, Clinical (Medical) Genetics https://www.ntpf.ie/home/outpatient.htm

[iii] Dying to see you? Deaths on a clinical genetics waiting list in the Republic of Ireland; what are the consequences? J Community Genet DOI 10.1007/s12687-020-00491-3

[iv] "…increasing referrals, greater demand for inpatient consultations, and increasing investigational complexity, all of which contribute to greater service demand." The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review, Genetics in Medicine (2020) 22:210–218; https://doi.org/10.1038/s41436-019-0602-2

[v] The changing face of clinical genetics service delivery in the era of genomics: a framework for monitoring service delivery and data from a comprehensive metropolitan general genetics service, Genetics in Medicine (2020) 22:210–218; https://doi.org/10.1038/s41436-019-0602-2

[vi] Recommendations from the Rare 2030 Foresight Study, https://download2.eurordis.org/rare2030/Rare2030_recommendations.pdf

[vii] Interviewees were from systems in Northern Ireland, England, Australia, Norway, Austria and Finland.

[viii] "…opportunities for increased efficiency through task-sharing, delegation, alternative service delivery models, and augmentation of services through the use of technology."

[ix] Table 2, The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review, Genetics in Medicine (2020) 22:210–218; https://doi.org/10.1038/s41436-019-0602-2

[x] The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review, Genetics in Medicine (2020) 22:1437–1449; https://doi.org/10.1038/s41436-020-0825-2

[xi] The changing face of clinical genetics service delivery in the era of genomics: a framework for monitoring service delivery and data from a comprehensive metropolitan general genetics service, Genetics in Medicine (2020) 22:210–218; https://doi.org/10.1038/s41436-019-0602-2

[xii] Duplication of referral, a tsunami of paper: how much does it cost the Irish health services?, Irish Journal of Medical Science (1971 -) https://doi.org/10.1007/s11845-021-02866-y

[xiii] https://www.trakgene.com/about-our-clinical-genetics-software/

[xiv] https://www.gosh.nhs.uk/wards-and-departments/departments/clinical-specialties/clinical-genetics-information-parents-and-visitors/refer-patient-genetics-department/

[xv] HSE to establish two perinatal genetics testing hubs next year, Irish Examiner, 28 December 2022

[xvi] "Internationally, there is no consensus on the appropriate workforce of clinical geneticists pMC." Genetics in Medicine (2020) 22:210–218; https://doi.org/10.1038/s41436-019-0602-2

[xvii] Towards establishing consistency in triage in a tertiary specialty, European Journal of Human Genetics (2019) 27:547–555 https://doi.org/10.1038/s41431-018-0322-0

[xviii] Duplication of referral, a tsunami of paper: how much does it cost the Irish health services?, Irish Journal of Medical Science (1971 -) https://doi.org/10.1007/s11845-021-02866-y

[xix] Duplication of referral, a tsunami of paper: how much does it cost the Irish health services?, Irish Journal of Medical Science (1971 -) https://doi.org/10.1007/s11845-021-02866-y

[xx] NHS Genomic Medicine Service: Geographies, https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/

[xxi] The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review, Genetics in Medicine (2020) 22:1437–1449; https://doi.org/10.1038/s41436-020-0825-2

Additional References

Review of the Clinical Genetics Medical Workforce in Ireland, HSE (2019) https://www.hse.ie/eng/staff/leadership-education-development/met/plan/specialty-specific-reviews/clinical-genetics-2019.pdf