THE ADELAIDE HEALTH FOUNDATION

Study of Genetic Counselling and Testing in the Republic of Ireland

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UCD DUBLIN

The Adelaide Health Foundation

The Adelaide Health Foundation (AHF) is a voluntary foundation focused on advancing and promoting equitable access to healthcare services and education in Ireland, through the pillars of Community, Research and Education.

We have a proud history of supporting and publishing evidence-based research which can influence change within the Irish healthcare system, improve the health and wellbeing of the population and inform health policy.

AHF published research studies include:

- Assessment of funding options and barriers to Universal Healthcare (UHC) and an evaluation of general public opinions on the introduction of UHC in Ireland.
- Clinician and patient stakeholder analysis of chronic disease management in Ireland.
- A Health Asset and Needs Assessment (HANA) of Tallaght.
- Study and recommendation on how best to encourage patient and family involvement in the design and delivery of health services.

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Preface

Significant advances are being made worldwide in genetics and genomics research and services and an enduring interest in these areas led to the Adelaide Health Foundation commissioning this research study.

Research is one of the three pillars of the Foundation's work and our objective under this pillar is to "follow through" by supporting the implementation of study recommendations.

Shortcomings in Ireland's genetic testing services are well documented, with limited capacity and a small clinical workforce affecting the availability of and access to quality services.

The impact of these limitations on the patient, doctor and health professional experience led to the AHF commissioning this study to explore current practices and to document advancements in genetic testing and counselling services. We sought recommendations for the optimisation of treatment and facilities to improve the patient and staff experience in Ireland.

In scoping this research study, we invited expert input. We also welcomed the 2022 publication of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland.

Catherine Mac Daid
Chair, Adelaide Health Foundation
October 2024

1 Introduction

Why did we commission a Genetic Testing & Genetic Counselling Research Study?

Significant advances are being made worldwide in genetics and genomics research and services.

Development of genetic services in Ireland has not kept pace with similar services in other countries. They require regulation and are underfunded and fragmented. These shortfalls impact the availability of and access to quality services which negatively impact the patient, doctor and health professional experience.

The AHF's enduring interest in Genetics led to the commissioning of this research study of current services and recommendations for future developments.

The study fits with the values of the AHF, which include 'Independence in opinion and action of the foundation and medical practitioners, including but not confined to the fields of ethics and genetics.'

What were the aims of the study?

The project set out to capture the gaps in genetic counselling and genetic testing in Ireland that create risks for patients by:

- reviewing genetic testing and counselling services available in Ireland and comparing them against best international practice
- providing recommendations for service delivery together with a roadmap to improve the patient experience.

Who undertook the Research Study?

Following a comprehensive tender process, the UCD team led by Principal Investigator Professor Sally Ann Lynch delivered the project over 24 months.

2 Objective

Study of 'Genetic Counselling and Testing in the Irish Republic to include scoping of current practice, international comparisons and recommendations for national practice'.

Objective

The aim of the AHF funded research project was to:

- review genetic testing and counselling services available in Ireland and compare them against best international practice
- capture the gaps in genetic counselling and genetic testing in Ireland that created risks for patients, to identify the consequences of long waits to families accessing genetic services and the impact of this on patients' and carers' personal lives and plans.
- provide recommendations on improving service design and delivery together with a roadmap to improve the patient experience.

A research team was commissioned to examine current Irish Clinical Genetic services, review the relevant international best practice, and provide recommendations for Irish service delivery to meet the needs of patients and the public.

The AHF funded project delivered four main streams of research, which included targeted surveys to patients and families using Genetic services and to Genetic Counsellors (GC), a review of genetic testing practices and a review of risks within clinical genetics.

The research yielded significant findings which have translated to 8 recommendations [see Table 6.]

3 Research Methodology

The Research team addressed each of the project objectives separately, as the methodology to generate data required different approaches.

A final analysis with integrated findings from all aspects of the project was provided at the end of the research.

The Research team executed:

- Part 1 – Public perception of Irish genetic services and international comparisons
- Part 2 - Targeted survey of Genetic Counsellors
- Part 3 - Review of Genetic testing practices
- Part 4 - Review of risk within Clinical genetics

The following addresses each of these approaches.

Part 1 Public perception of Irish genetic services and international comparisons

To develop a full review of Genetic services within the Republic of Ireland, the Research team conducted a public survey and collaborated with service providers in other locations to complete a review and comparison in relation to international services.

Part 1.a Public Survey on Genetic Services

With the help of the Rare Diseases Ireland [RDI] patient advocacy group, the Research team created a survey to capture Irish people's experience of accessing genetic testing and clinical genetics. The survey was disseminated by RDI social media and open to Rare Disease families.

The survey asked about:

- waiting times
- the professionals who ordered testing and gave results
- impacts on families.

The survey was:

- available online for a month, through RDI over February/March 2022
- in the Republic of Ireland, open to anyone over 18 years of age

An adapted version of the survey was given to adults with Inherited Metabolic disease through the clinic at the Mater Hospital.

The team found that Irish patients experience long waiting times to access clinical genetic services. Patients self-report anxiety and stress. They also reported strained family relationships due to delays in diagnosis and lack of knowledge about whether the condition might affect another family member.

These delays negatively impact decisions around family planning, education and employment and affect family members wanting to find out about their own risk.
Reference Table 1: *'Reported Impact of Waiting Times on personal life.'*

Reported Impact of waiting time on personal life (n=142)	N	%
Placed tension on relationships with partner, family members or friends	45	32%
Wider impact on relative's family planning/relationships/education/ employment plans	33	23%
Delayed plans to have more children	25	17.6%
Changed/delayed education	11	8%
Delayed plans to start a family	9	6.3%
Changed/delayed employment	8	6%
Delayed plans to marry/settle down/commit to a relationship	6	4.2%
Delayed plans for mortgage or insurance	2	1.4%

Table 1: *'Reported Impact of Waiting Times on personal life.'*

Mainstream genetic testing activity is evident – with testing being organised by healthcare professionals who are not working in genetics. Some families expressed concern with the competency of health care professionals arranging tests and delivering genetic results. They were also concerned about delays in accessing clinical genetics expertise once they have received a genetic diagnosis from another healthcare professional.

'The delay in my results gravely impacted our ability to think about starting a family'

77% of adult Metabolic patients surveyed had never met a genetic counsellor and 36% could not accurately recall whether they had genetic testing (despite all having had genetic testing done).

'Extremely stressful time as my husband's diagnosis has a profound effect on our adult children who are all of childbearing age'

'We need understanding and don't have it'

Patients and families surveyed also had an opportunity to give open ended comments on their experience and frustrations with genetic services in Ireland.

As part of the genetic services public survey, a question was asked about genetic counsellors: 'What do you believe is included in the role of Genetic Counsellor' [multiple choice]. 171 participants responded. Participants correctly identified some roles of genetic counsellors, but some also expected that genetic counsellors would provide long term psychological counselling and paternity testing which is not the remit of a GC role.

What do you believe is included in the role of a Genetic Counsellor?

Part 1.b International comparison of Genetic Services

The researchers collaborated with the national rare disease patient advocacy organisation Rare Diseases Ireland and Polwarth Strategy Ltd to look at genetic services in seven countries: Ireland, Northern Ireland, England, Norway, Finland, Austria and Australia. This was part of a wider study commissioned by Rare Diseases Ireland to identify international best practices to help reduce waiting lists. Their study report is on Rare Diseases Ireland website 'Ending the wait' Ending the Wait – Rare Diseases Ireland (rdi.ie).

The genetic service provision was examined, through interviews with Consultant Clinical Geneticists. The interview transcripts were reviewed to look for possible solutions to improve Irish Clinical Genetic services.

The following observations were made;

Area of Service	Country	Ideas for improvement
Improving access to Clinical Genetic services	Australia Northern Ireland	Satellite, telemedicine, and telephone clinics allow patients and families to be seen closer to home and can have a positive effect on waiting lists as less clinic space is needed
Mainstreaming	England	For some common genetic conditions, genetic testing can be arranged by the GP with support and advice from Clinical Genetic services. A National Genetic test directory is a useful support tool. However, encouraging genetic testing by mainstream clinicians requires a supportive framework e.g. training in consent
Genetic Counselling profession	Norway Austria Finland	As in Ireland, Genetic Counselling is not universally recognised as a Health and Social Care Profession and Genetic Counsellor is not a protected title. Genetic Counselling is valued by Clinical Geneticists as an important part of Clinical Genetic services. Steps have been taken to introduce formal Genetic counselling training programmes
		The importance of Genetic Counsellor role obtaining professional registration was noted.
Workforce Planning	Australia	Workforce planning is important, however this needs to be properly implemented to make sure sufficient numbers of staff are trained.
		The introduction of support roles e.g., Clinical Assistants or Genomic Resource Associates can help with the early steps in the patient pathway e.g. preparing the charts for clinics.
Genetic Testing	Australia Finland England Northern Ireland	While development of Irish genomics laboratory services is welcomed, this remains highly specialized testing and unlikely to be delivered fully in-house due to the rarity and complexity of tests. Other centres continue to use some accredited external laboratories, both public and commercial, and the expectation is that Ireland will also need some reliance on external testing

The improvements emerging from this review have been incorporated into the overall integrated recommendations of the entire study.

Part 2 Survey of Genetic Counsellors in the Republic of Ireland

The project team surveyed Irish genetic counsellors in two separate online surveys, covering the areas of:

1. Workforce and practice
2. Wellbeing

The Research team created a Work practice survey for Irish genetic counsellors, using published surveys from the United Kingdom, American and Canadian Genetic Counsellors as guidance.

There have been very few dedicated surveys on Wellbeing in Genetic Counsellors (GC). Because of this the Research team used the recognised ProQOL (Professional Quality of Life) health measure for professional quality of life in health care workers (https://proqol.org/proqol-health-1) for the second survey.

For confidentiality the surveys were anonymous. To maintain confidentiality, it was not possible to link results to information obtained from both surveys.

Survey links were emailed to all genetic counsellors and advertisements were sent to genetics departments. With an estimated 18 GCs working in the Republic of Ireland there was an 83% survey response rate.

- 93.3% of GCs are in public sector employment and 33.3% in mainstreamed GC roles (employed outside of genetics clinics).
- 47% of Genetic counsellors work in the field of general genetics and/or cancer genetics.
- 53% of genetic counsellors have at least some of their work in a specialist field, such as preimplantation diagnosis, rare diseases or prenatal diagnosis.

Genetic Counsellors perform a variety of tasks, as shown in Table 2.

For confidentiality the surveys were anonymous. To maintain confidentiality, it was not possible to link results to information obtained from both surveys.

Which tasks are a regular part of your job?	N	%
Production of information for patients or healthcare providers	14	93.3%
Evaluation of family history and advice by letter	13	86.7%
Telephone patient appointments	13	86.7%
Liaise with laboratories abroad	13	86.7%
Liaise with local clinical services	13	86.7%
In-person patient appointments	12	80.0%
Attend multidisciplinary meetings outside of primary department	12	80.0%
Liaise with local laboratories	12	80.0%
Teaching	11	73.3%
Triage of referrals	10	66.7%
Policy development	9	60.0%
Mentor genetics counselling trainees	7	46.7%
Liaise with clinicians abroad	6	40.0%
Management/supervision of other staff	5	33.3%
Research	5	33.3%
Advocacy	5	33.3%

Table 2: *'Which tasks are a regular part of your job?'*

The specialised workforce is highly trained - 93.3% are professionally registered with the UK and/or the EU genetic counselling boards. Continuing professional development (CPD) and counselling supervision which are mandatory for registration are frequently self-funded and undertaken in personal time.

'Highly demanding clinical workloads can make it hard to make time for CPD, reflection etc'

The job title 'Genetic Counsellor' is not a protected title which creates a risk for the patient/public and the Healthcare Professional.

'Proper planning is needed to ensure high standards are kept, proper clinical oversight is established, CORU registration is made a priority, career structures are developed and a professional organisation founded'

The surveys were undertaken in February 2023. The research team are aware that since then there has been an increase in Genetic Counsellors entering the field, currently operating at a junior level.

The ProQoL Health survey measures Compassion Satisfaction and Compassion Fatigue across 5 themes. Genetic Counsellor respondents scored high for Compassion Satisfaction (ie. positive consequence of caring for others), indicating that despite challenging work environments GCs feel they are positively contributing to clinical genetics services. Perceived support was reported as moderate. Compassion Fatigue measures a combination of how respondents rate Moral Distress, Secondary Trauma and Burnout. Although Moral Distress is currently reported as low, Burnout and Secondary Trauma ratings were moderate with a threat to individual GC wellbeing and a risk of developing Compassion Fatigue as the workforce faces ongoing challenges to deliver the service their patients require. **Reference Table 3:** Average ProQOL (health) measures

Average ProQOL (Health) measures	
Category	**Rating**
Compassion Satisfaction	High
Burnout	Moderate
Moral Distress	Low
Perceived Support	Moderate
Secondary Trauma Stress	Moderate

Table 3: *Average ProQOL (health) measures*

As part of the questionnaire, genetic counsellors were able to give open-ended comments on their reflections of the profession of genetic counselling in Ireland.

If there is going to be a demand for more GCs training /pathways to developing GCs are needed

Proper planning is needed to ensure high standards are kept, proper clinical oversight is established, CORU registration is made a priority, career structures are developed and a professional organisation founded

Genetic services in Ireland are chronically under funded and under staffed in comparison to departments I have worked in, in the UK

Often find myself apologising for wait times & given there are so many patients to see there is not much time for service or indeed my own professional development

I was unable to find work in Ireland as a GC because I do not live near Dublin

Very positive to see the profession growing and evolving

I would love the opportunity to do some research alongside clinical work

It's like working with your hands tied behind your back. I can present all the reproductive options to families & then families can explain to me why they can't afford any of the options

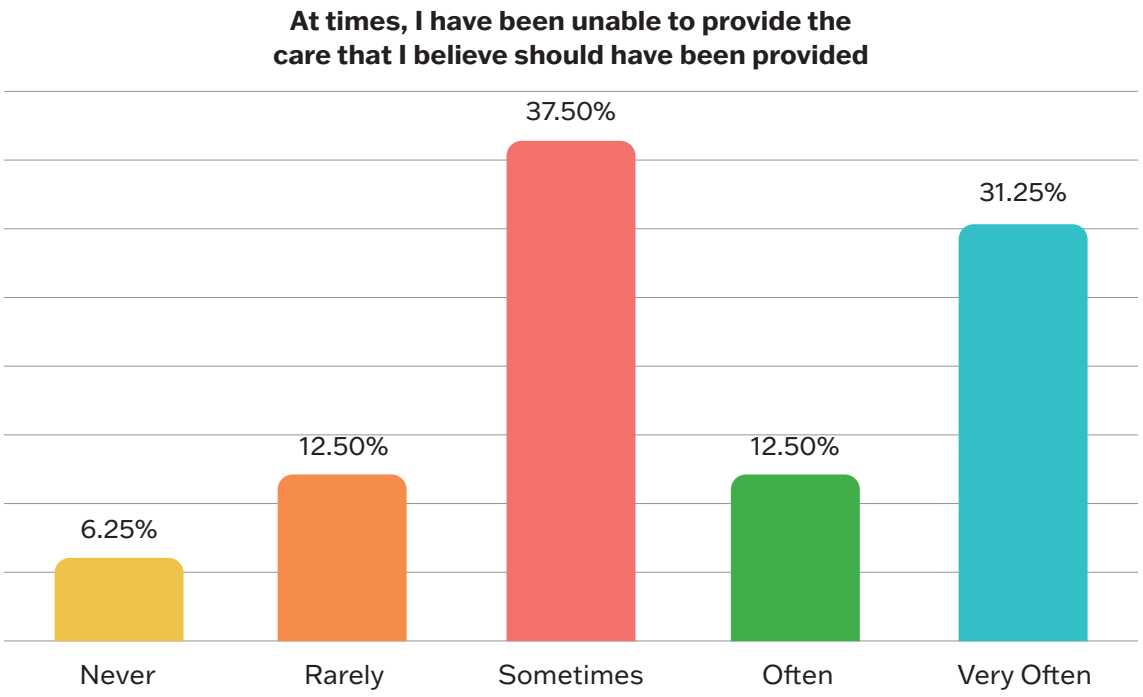
There will be more GCs in mainstream roles so clinical governance needs to be worked out, clear roles and scope of practice

Highly demanding clinical workloads can make it hard to make time for CPD, reflection etc

Variants are being upgraded & downgraded all the time, but there is very little training and support for Genetic Counsellors in managing this with patients and families

It is difficult as a newly qualified GC trying to become registered...Not being able to move around to get exposure to different specialities

➜ The questionnaire also asked Genetic Counsellors if they felt they could not deliver care required. Of those surveyed, over 80% felt that, at some time, they had been unable to provide the care that they believed should have been provided.

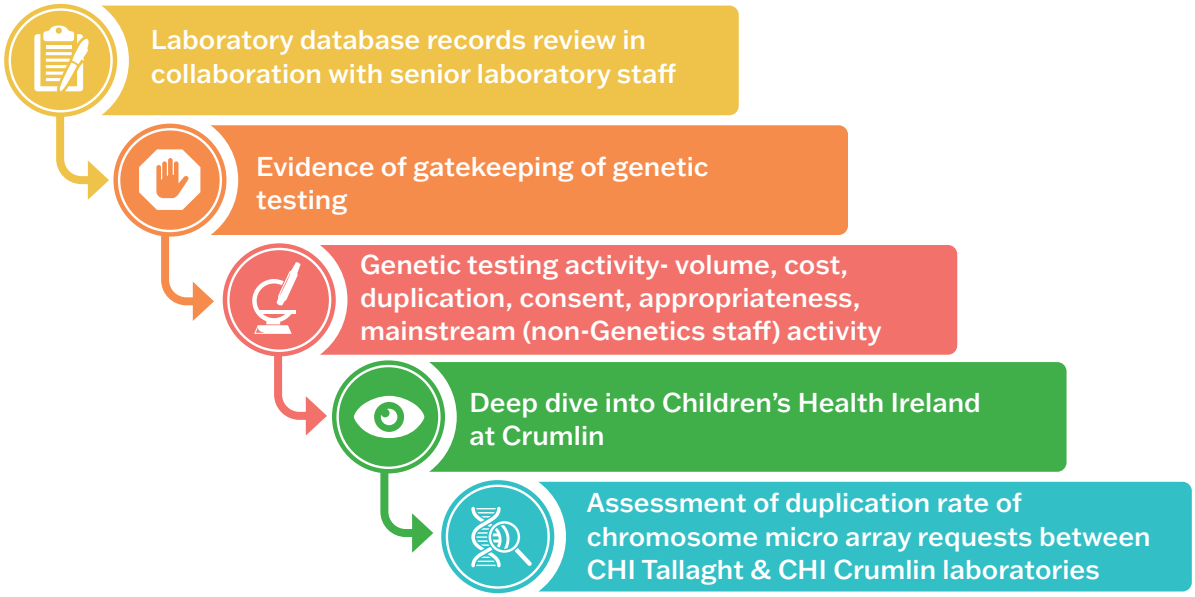


'Variants are being upgraded & downgraded all the time, but there is very little training and support for Genetic Counsellors in managing this with patients and families'

Part 3 Review of Genetic testing practices

A review of records from the Genetics Laboratory was carried out at Children's Health Ireland (CHI), Crumlin (CHI@Crumlin). Separately the team collaborated with CHI@ Tallaght in Tallaght University Hospital laboratory to estimate duplication rate of one specific test, (chromosome microarray) as a marker for wastage as this is a once in a lifetime test.

The deep dive review in CHI@Crumlin looked for volume of genetic test requests and evidence of mainstream activity. Test requests were compared to the UK's Genetic Test Directory and EU ethics standards to see if doctors are ordering the most appropriate tests in a correct and ethical way.



Laboratory results show that clinicians and laboratories are working without a genetic testing framework. This means that there is no guidance for which tests are ordered, and who orders them. Some tests are analysed in CHI@Crumlin but many are sent abroad.

The deep dive revealed that there is a 6% duplication rate of one specific genetic test, a chromosome microarray. This is a 'once in a lifetime' test and should not be repeated. Any repetition of testing is a waste of resources, diverts money from elsewhere in the health service and is perceived as a clinical risk.

The team feels this is a marker for widespread duplication throughout Ireland as there is no centralised service nor IT inter-operability, so no one knows who has ordered what or where it was ordered.

	Gatekeeping	Cost Saved	Cost Wasted
Single gene & chromosome duplicate test orders	Yes	€197,700	€0
Microarray duplication comparison between CHI@Crumlin & CHI@Tallaght 73/1213 tests were duplicated (6%)	No	€0	€21,720

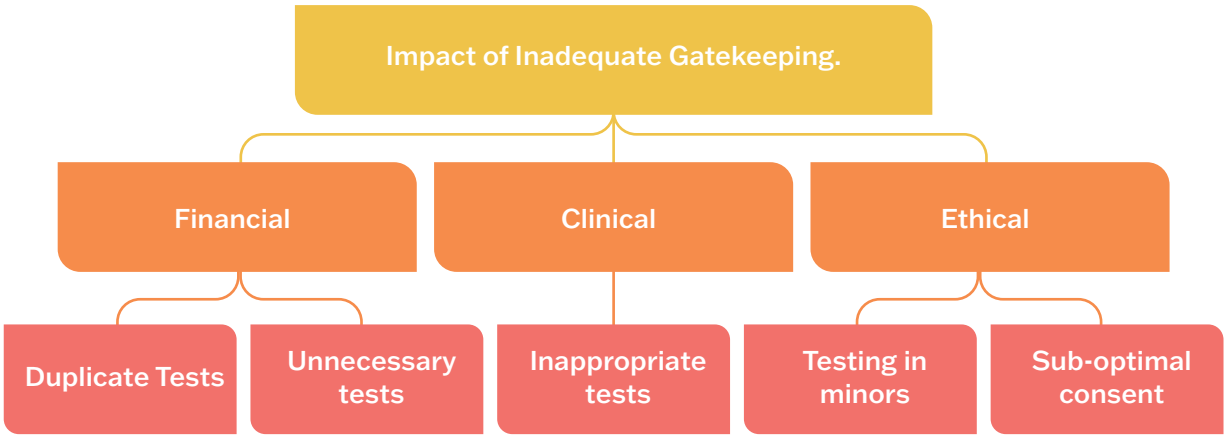
Duplication test orders in CHI@ Crumlin with gatekeeping in place	Single gene testing	Chromosome testing
Same clinician requesting duplicate test	55/111 (49.5%)	101/234 (43.2%)
Same hospital requesting duplicate test	64/111 (57.7%)	158/234 (67.5%)

The following maps illustrate the difference a testing sample journey makes between testing services within the NHS, Northern Ireland and the HSE Republic of Ireland. This also illustrates the risks within the current HSE processes.

The research showed that gatekeeping by trained and experienced clinical scientists (where test requests go through quality checks before analysis) leads to cost saving, avoids unnecessary tests and promotes ethical testing (which includes appropriate consent procedures).

However, very limited gatekeeping leads to duplicate, unnecessary, and inappropriate genetic testing. The research found evidence of limited clinical details on test request forms and poor test selection by the clinical teams. For example, requesting **multiple gene testing** (a panel test) and an **exome/genome** which would include the same genes, and is not cost effective.

There is limited oversight of financial costs, test requests and sample tracking. There is also evidence of potential ethical issues, for example inappropriate testing in children for later onset diseases, and sub-optimal patient consent.



Genetic Testing - what happens?

HSE, Republic of Ireland

Multiple dispatch labs within Dublin and Cork, Cavan, Drogheda, Sligo, Mayo, Limerick, Galway, Wexford, Waterford, and Kilkenny all dispatch genetic tests.

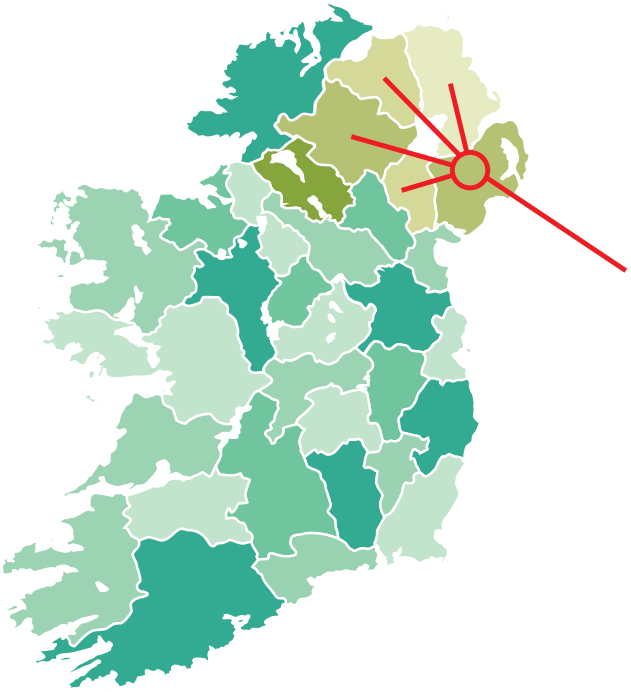
Sending out to multiple external foreign labs, both diagnostic (UK & EU) and commercial labs (Germany, Finland & USA).

The fetal medicine units in the Republic of Ireland are largely dependent on one UK based diagnostic laboratory to provide antenatal tests that require culture techniques (e.g. chorionic villus biopsies and aminocenteses).

Multiple dispatch labs means:

- ➜ No communication between labs
- ➜ No audit trail
- ➜ No check for unnecessary duplication
- ➜ Creation of unsafe working environment
- ➜ Significant risk to patient experience
- ➜ Risk to staff

NHS, Northern Ireland



In Northern Ireland, no matter where a baby is born all parental samples go to Belfast.

All Parental Samples automatically follow Childs sample

In Northern Ireland all genetic testing is centralised to one lab, which is supported by;

- Upskilling scientist
- Capital funding
- Equipment kept up to date

Central dispatch lab means:

- ➜ Full audit trail
- ➜ Full transparency
- ➜ Controls are implemented
- ➜ Waste minimised
- ➜ No Duplication of tests

Part 4

Review of Risk within Clinical Genetics

One of the aims of the study was to provide recommendations on improving service design and delivery, together with a roadmap to improve the patient experience.

The team also reviewed how best to develop a clinical genetic specific risk assessment tool with HSE clinical risk staff and State Claims Agency for expert input as they have full understanding of the risk matrix and how it is used in practice. Their input guided the team as to how best to adapt it for our purposes.

To meet this objective, a patient pathway was developed to map out key elements of the Clinical Genetics patient journey.

A 22-step Process Map spans 5 stages:

- patient and family history assessment
- clinical management of genetic testing
- sample processing and analysis
- result transmission
- result discussion.

The pathway was developed by the Research team reviewing 55 clinical genetics records where potential clinical incidents were identified and avoided.

Clinical genetics teams in five other European centres were asked to test the pathway and suggest extra or missing steps.

The pathway provides a framework for all aspects of the project:

- Helping healthcare providers outside of genetics appreciate all the steps that are necessary for safe ordering of genetic testing.
- Allowing genetic teams to identify risk areas in their practice
- Helping identify possible system errors within units where intervention or improvements could prevent harm.
- Training tool for non-genetic staff about the steps needed for safe and ethical genetic testing as testing goes mainstream.

Patient Pathway of key elements in the Clinical Genetics patient journey

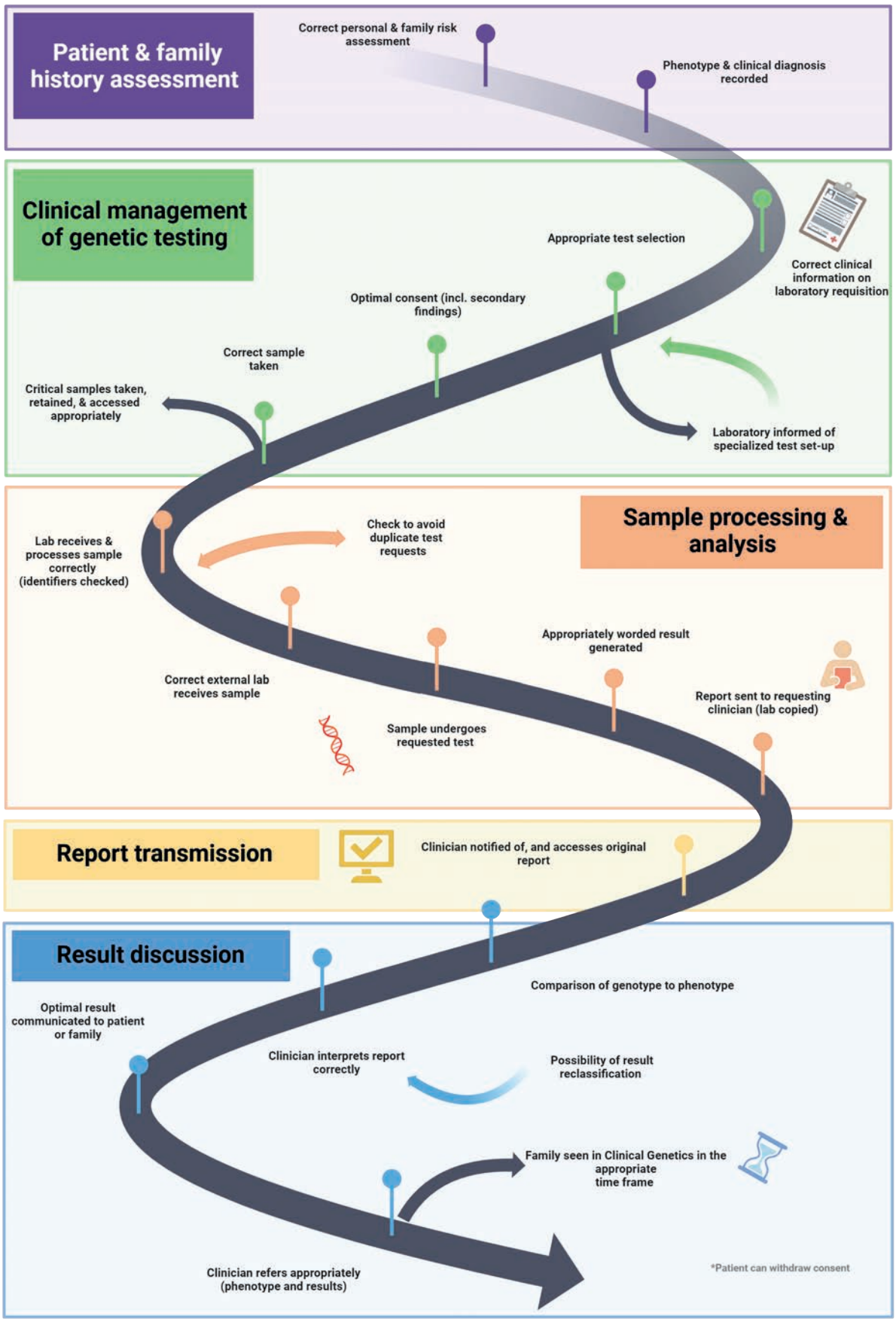


Image generated by Biorender.com

The Research team aimed to identify and measure risks seen in clinical genetics as a quality improvement tool.

Two groups of genetic charts (pregnant and non-pregnant) which had identified risk events were reviewed retrospectively. The patient pathway was used in conjunction with the HSE traffic-light risk matrix system HSE Risk Assessment Tool to rate risk severity.

The HSE risk assessment matrix was used as guidance within the exercise as it is a robust assessment tool with a proven history in Risk assessment within healthcare settings and was applicable to the range and disparity of scenarios observed by the team.

Risk Impact categories - how can risk harm?	
Harm to person	National Planning
Harm to Staff	Adverse publicity/reputation
Service user experience	Financial - hospital
Clinical service disruption	Financial - patient
Objectives/Compliance	Environmental

In addition, the CHI@ Crumlin Clinical Genetics team completed a prospective risk survey for all cases where risk occurred over a six-week period.

1 in 10 appointments had a risk event and nearly half of those had risk at more than 1 step in the pathway.

The majority of risk was attributable to:

- long waiting lists
- IT deficiencies
- insufficient clinical and support staff
- gaps in non-geneticists' genomic knowledge including consent and testing

The six-week risk survey was completed by six clinical genetics centres: - Dublin, Ireland; Belfast, Northern Ireland; Craiova, Romania; Oulu, Finland; Oxford and Newcastle, England. Risk events across the pathway varied between centres due to differing processes. Some centres suggested that they could be under-reporting risk events.
Reference Table 4: Clinical genetics centres risk survey findings.

	Dublin	Craiova	Belfast	Oulu	Oxford	Newcastle
Appointments with risk events	10.7%	20.3%	3.6%	0.8%	12.5%	1.3%
Risk cases with ≥2 pathway steps broken	42.0%	20.0%	52.4%	25.0%	34.8%	46.2%

Table 4: *Clinical genetics centres risk survey findings.*

The team noted risk reducing practices in other centres that could be implemented in Ireland:

- genomic resource associates to support early steps in the patient pathway
- robust IT infrastructure and connectivity
- genetic testing directory
- clinical and laboratory gatekeeping

All centres were asked to suggest improvements to the pathway, which they observed to be a beneficial and a good teaching tool. The Oxford centre have introduced use of the tool across the South-west of Britain network.

The team were grateful for the input from the State Claims Agency and HSE's Clinical Risk personnel to aid development of pathway into an assessment tool that can be used to:

- Monitor risk events specific to clinical genetics
- Investigate any future serious incidents to identify possible system errors that need addressing
- Inform compensation for State Claims Agency

The risk assessment tool was re-audited in both Dublin and Oxford in January/February of 2024. Findings from the re-audit show the value of using the tool and a reduction in the number of red categories.

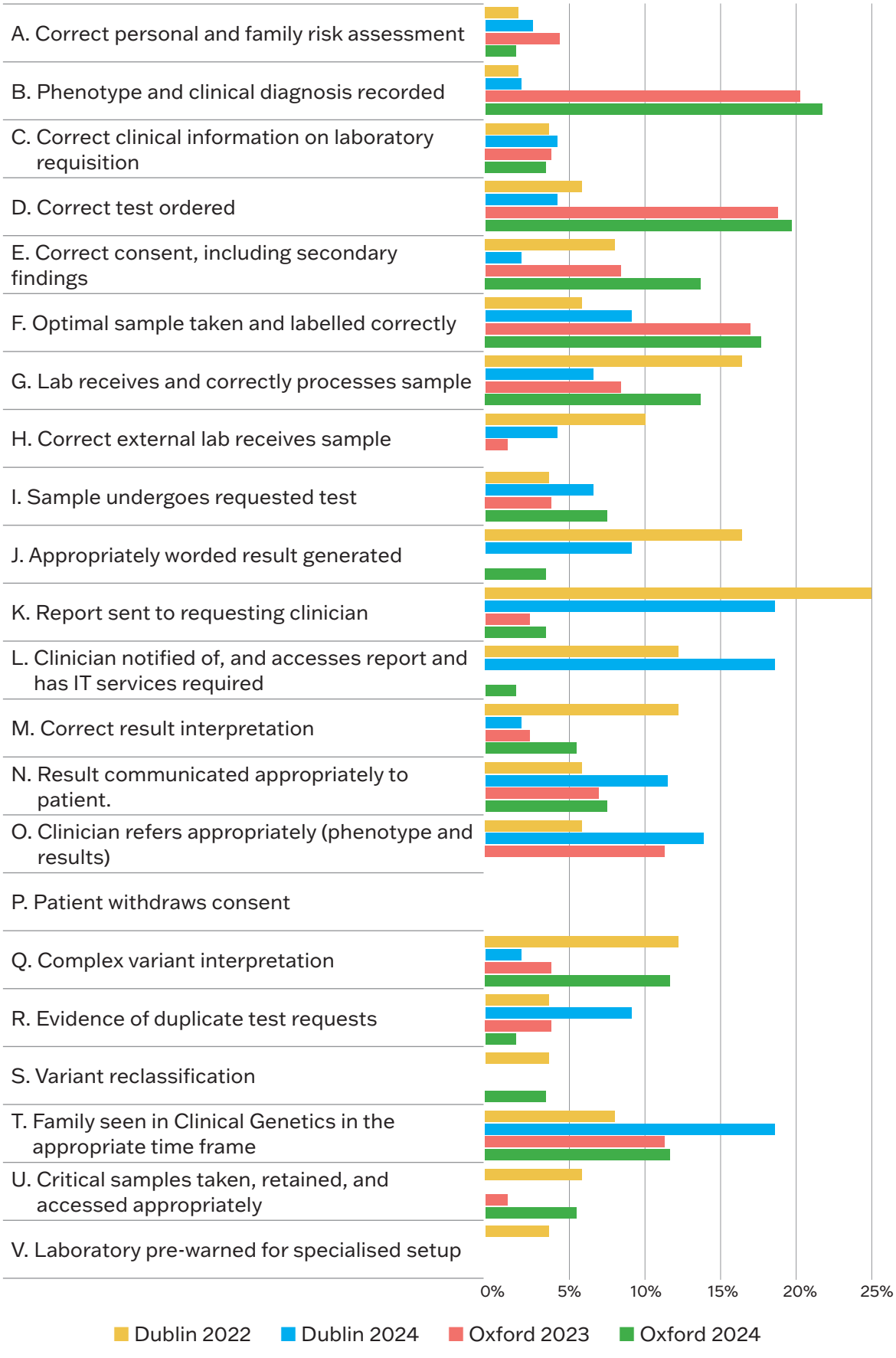
Through the repeat audit, the Oxford team noted a reduction in events in 2024. They feel that proactive training and education of mainstream clinicians, encouraging them to complete a form introduced to record informed consent along with a record of the discussion with the patient has resulted in improvements in the issues they were noting particularly at receipt of sample.

The repeat audit conducted by the Irish team reported the same number of events, although a reduction in serious events such as loss of critical samples and referrals for secondary findings was noted. **Reference Table 5:** Results of Risk Assessment re-audit.

Dates Inclusive	Dublin 2022	Dublin 2024	Oxford 2023	Oxford 2024
	09/2022-10/2022	15/01/2024-23/02/2024	19/03/2023-30/04/2023	29/02/2020-11/04/2024
Number of Clinical Genetic appts	591	671	1240	1458
Number of Events	55	57	80	42

Table 5: *Results of Risk Assessment re-audit*

The following chart illustrates the number of categories assessed and compares findings identified by the Risk Tool in 2022 and 2024.



4 Project Findings & Recommendations

Summary Findings

Patient/Family experience

Families requiring access to genetic testing and clinical genetic expertise are facing significant challenges in their personal lives due to the lack of access to correct expertise and ongoing extended waiting times.

The researchers recommend investment across the field of clinical genetics to ensure the delivery of safe and effective services by:

- increasing staffing levels
- investing in new roles such as genomic resource associates
- Promoting the importance of Clinical scientists who can advise on consent, optimal and most appropriate testing, and gatekeeping

Investment in these core areas will drive an improved service and experience for families.

Genetic Counsellors

Maintaining registration through genetic counselling supervision and CPD requires employer support, and this needs priority attention. Considering the increasing number of pre-registration genetic counsellors, support for professional mentoring is a requirement.

- Formal recognition of the profession is necessary to ensure Genetic Counsellor is a protected title, thereby safeguarding patients and Genetic Counsellors.
- Genetic counsellors (and indeed Genetic Clinical Scientists) are not included in the 26 professions represented by the National HSE Health and Social Care Professions (HSCP) Office.
- The HSCP office provides strategic leadership and support to maximise HSCP potential and achieve the greatest impact for the delivery of people centred, integrated care and develop National policy. As a result, Genetic Counsellors and Clinical Scientists cannot avail of opportunities to promote and develop their specialty and denies access to funded courses/training ad¬ministered by the HSCP office. These professions need support to access continuous professional development to maintain standards. The team would strongly advocate for this issue to be resolved.
- A national network of genetic counsellors may help to promote cohesiveness of the workforce, especially for those based in mainstream roles.
- Due to the misconception around the role of Genetic Counsellors, a public education program is essential.
- A workforce plan is needed to stabilise and future-proof the profession so that recruitment deficits are addressed and managed on an on-going basis.

Laboratory / Gatekeeping

Laboratory genetic testing & gatekeeping practices require investment in areas such as:

- Development of an interconnected IT infrastructure
- National Testing Directory
- Inclusion of Genetics laboratory staff within HSE HSCP registered specialities
- Training laboratory staff in gatekeeping practices
- Consideration towards a more centralised service

Education

An education program is required to address both public and professional sectors in:

- Improving Genetic and genomic literacy and competency to support the growth in testing in mainstream healthcare settings
- Consent
- Understanding the implications of secondary and add-on testing, with a particular focus on optimal and ethical genetic testing in minors.

Risk Tool

The deployment of the patient pathway as a research and audit tool will support quality improvement and overall patient safety.

Use of this tool during the audits revealed its flexibility; It can be used to analyse individual risk events, be used as a continual risk assessment monitoring tool or used whilst doing a retrospective audit of a department to identify possible high-risk areas.

- Use of the pathway may address the high level of red and amber risk ratings on clinical genetic patient charts by highlighting stages in the process which need attention, including monitoring the effectiveness of any interventions.

Recommendations

The researchers made 8 overall recommendations for both Policy and Practice based on the integrated findings from all aspects of the project.

Table 6: *Recommendations for policy and practice*

1	There should be timely access to clinical genetics expertise to ensure families have an understanding of the implications of a genetic diagnosis and have access to care pathways.
2	Ongoing HSE support and investment for clinical genetics teams is required: to ensure the delivery of a safe and effective service, to increase current staffing levels and invest in new roles such as genomic resource associates and laboratory gatekeeping staff.
3	HSE and employer provision of protected time and funding for genetic counsellors is needed to access continuing professional development, and counselling supervision to maintain competency and registration
4	There should be formal HSE recognition of the profession to ensure Genetic Counsellor is a protected title to safeguard patients and practitioners.
5	A national genetic testing directory is required to support clinicians and laboratories in optimal and ethical testing and implementation of gatekeeping
6	A national educational framework should be developed to support the growth of mainstream genetic and genomic testing; to develop health care professional competency in genomic testing and consenting procedures.
7	Centralised, digitally accessible HSE laboratory infrastructure to permit genetic testing visibility and interoperability across laboratories and clinicians is urged.
8	A Clinical Genetics risk assessment tool by teams engaged in genetic and genomic testing should be used to allow accurate risk assessment and support quality improvement and patient safety

5 Conclusion

Conclusion

Genomic technologies have become a significant part of mainstream medicine.

As each human being carries millions of DNA variations it is important that staff are trained in interpretation of the data and that informed consent is taken.

Investment is required for both clinical genetic staff (medical and genetic counsellors) and trained laboratory staff, to ensure Ireland has a safe system to optimise care of Irish patients going forward.

This study highlights the imperfect system staff are currently working in.

It identifies key areas where investment would introduce a safer system for Irish patients.

It identifies areas of waste which if addressed could allow re-direction of money to areas where investment has been deficient.

The key areas that require fixing include regulation of Genetic Counsellor as a profession, improved interoperability of IT system and education of mainstream staff in taking optimal consent.

The development and implementation of the risk assessment tool should allow Clinical genetic units to monitor high risk areas that emerge and put controls in place to minimise patient harm.

6 Scientific Papers

Genetic Services survey - Experience of people with rare diseases and their families accessing Genetic Services in the Republic of Ireland.

A Ward, D Butterly, D Lambert, University College Dublin School of Medicine, V McGrath, Rare Disease Ireland, J. O'Byrne, Mater Misericordia University Hospital, Sally Ann Lynch, University College Dublin School of Medicine, and Children's Health Ireland, Crumlin, Dublin.

Waste not, want not; measuring waste and potential clinical risk from limited gatekeeping of Rare Disease genetic testing in the Republic of Ireland

Sally Ann Lynch, Dearbhla Butterly, Deborah M. Lambert, Catherine Clabby, Bronagh O'hIci, Louise Johnston, Stephanie Kelly, Denise McDonald, Jennifer McDaid, Alana Ward

(Submitted March 2024 Irish Medical Journal-Paper in Press)

Metabolic paper Experience of people with Inherited Metabolic Disease and their families accessing Genetic counselling and Genetic testing in the Irish Republic

Arnott C, Ward AJ, Lambert DM, Butterly D, McGrath V, Lynch SA, O'Byrne J

(Journal of Community Genetics)

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Research team

- Professor Sally Ann Lynch, Principal Investigator
- Dr. Alana Ward, Project Researcher
- Debby Lambert, Project Researcher
- Dearbhla Butterly, Project Researcher
- Vicky McGrath, Rare Disease Ireland, Co-Investigator
- James O'Byrne, NCIMD Mater Misericordiae Hospital Co-Investigator
- Hosting Institution: University College Dublin

Project Collaborators

- Dr. Karen Flood, Rotunda Hospital

International Service Comparisons

- Alistair Hodgett, Polwarth Strategy

Laboratory Review:

Deep Dive CHI@Crumlin Genetic lab

- Jennifer McDaid Head of Genetics Laboratory CHI@Crumlin
- Catherine Clabby Molecular Clinical Scientist CHI@Crumlin
- Bronagh Ó'Hicí Molecular Clinical Scientist CHI@Crumlin

Identifying possible duplication of chromosome array testing:

- Louise Johnston Senior Scientist Laboratory Medicine. Tallaght University Hospital
- Ms Stephanie Kelly CHI@Tallaght
- Dr Denise McDonald CHI@Tallaght

Development of Risk Tool:

Risk Tool Collaborators

- Ioana Streata Craoiva Romania
- Outi Kuismin Oulu Finland
- Helen Stewart Oxford
- Mari Bandiola Oxford
- Gillian Rea Belfast
- Marta Bertoli Newcastle

Risk Tool Re-audit 2024

- Sofiia Kravets UCD Medical student
- Anna Griffin University of Glasgow Medical student

Children's Health Ireland

- Deborah Behan, Risk & Clinical Compliance Lead

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- Elaine Kilroe, Enterprise Risk Management, HSE

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- Sheila King, Clinical Risk Advisor
- Natasha Coen, Senior Clinical Risk Manager

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THE ADELAIDE HEALTH FOUNDATION