# THE UNMET NEED IN CANCER GENETIC SERVICES: **CONDUCTING AN ENVIRONMENTAL SCAN OF THE CANCER GENETICS SERVICES IN AN IRISH CONTEXT UNDERPINNED BY A MIXED METHODS APPROACH**

## Report prepared for the Irish Cancer Society[1]

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UCC
University College Cork, Ireland
Coláiste na hOllscoile Corcaigh

Irish Cancer Society

| ABBREVIATIONS | |
|---|---|
| BMJ | British Medical Journal |
| BRCA | Breast Cancer Gene |
| CGS | Cancer Genetic Services |
| CGT | Cancer Genetic Testing |
| CGTC | Cancer Genetic Testing and Counselling |
| DCIS | Ductal Carcinoma in Situ |
| DOH | Department of Health |
| FHQs | Family History Questionnaires |
| GC | Genetic Counselling |
| GP | General Practitioner |
| GT | Genetic Testing |
| HBOC | Hereditary Breast and Ovarian Cancer |
| HCPs | Healthcare Professionals |
| HCRA | Hereditary Cancer Risk Assessment |
| HSE | Health Service Executive |
| JBI | Joanna Briggs Institute |
| MDTs | Multidisciplinary Teams |
| MMAT | Mixed-methods Appraisal Tool |
| NA | Not Applicable |
| NCCP | National Cancer Control Programme |
| NCHD | Non-Consultant Hospital Doctor |
| NHS | National Health Service |
| NICE | National Institute for Clinical Excellence |
| PRISMA | Preferred Reporting Items for Systematic Reviews and Meta-Analyses |
| RCPI | Royal College of Physicians of Ireland |
| SD | Standard Deviation |
| UCC | University College Cork |
| UK | United Kingdom |
| USA | United States of America |


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# Contents



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## DISCLAIMER

The views, information, or opinions presented within the research findings (Chapter 1) reflect the perspectives of the individuals who volunteered to be involved in the research; they do not necessarily reflect the perspectives of the Health Service Executive or other providers of cancer genetic services in an Irish context. The research findings reflect individuals' perspectives at the timeframe (2020) of data collection.

# FOREWORD

Inherited faulty genes play a major role in 5-10% of all cancers. For those affected, having early access to genetic testing can give them the knowledge they need to understand their risk. It can empower individuals at risk to take preventative steps to reduce their chances of getting cancer. It can also enable a patient's medical team to tailor their treatment to their particular genetic mutation, thereby increasing the chances of success.

Global advances in genetic testing, preventative treatment and personalised medicine thereby offer huge hope. Investment in, and equitable access to, these services has been proven to save lives and improve quality of life for people with a higher genetic risk of getting cancer.

Sadly, as is clear from this report, Ireland lags far behind other countries in this regard and the consequences of that for the individuals and families affected are devastating.

We thank our colleagues at University College Cork for their thorough work in considering and collating the perspectives and experiences of users of genetic services and the professionals who work in the field.

What is clear from this important piece of research is that services have been starved of investment and resources. Healthcare workers involved in cancer genetics and follow-on services are doing incredible work, but are completely overstretched. Some patients have had to wait for up to two years for testing and counselling. Patients found to be at a high risk have also reported that after they received their results, they had to wait two years for risk-reducing procedures.

In addition, fears of how their genetic information would be used after testing is not only causing great anxiety for those who are tested, it is also putting some people off getting tested at all, thereby depriving them of information that could be lifesaving. 154 people with experience or knowledge of the process were surveyed for this research. Over 4 in 10 of those had concerns over how their genetic information would be used after testing, including worries over whether it could be used against them by employers or insurers.

A separate survey of 52 healthcare professionals highlighted barriers for accessing the services with 6 in 10 saying they are under-resourced and 4 in 10 concerned about access to follow-up surgery for patients deemed to be at a high risk.

This report and its recommendations outline the step-change that is required to harness the potential cancer genetics brings, and what Ireland needs to do not just to keep up, but to catch up.

The Irish Cancer Society is committed to driving change in this area, in collaboration with individuals and groups affected and healthcare professionals.

To achieve meaningful change for families, Government needs to not only expand capacity for testing and counselling, but also ensure that the follow-on services that are needed by people diagnosed with a genetic risk of cancer are in place and can be accessed swiftly.

The Irish Cancer Society will seek a better-defined and better-resourced pathway for people who test positive with a faulty gene which increases their lifetime risk of developing cancer, so they can avail of the preventative options they need.

We will advocate for the appropriate screening, chemoprevention, risk reduction and psychological services and supports that are needed once people are discharged from the care of genetic services.

We cannot do this alone and we are thankful to all the people who shared their experience to give such a strong evidence-base for change in this area. Everyone involved in the planning and provision of genetic services can learn from its findings.

The National Cancer Strategy set out a vision for cancer genetics in Ireland to "become a leading example of how to incorporate genetics into healthcare". We will continue to push for change until the necessary resources are provided to achieve that ambition, delivering a brighter future for people with a suspected or diagnosed genetic risk of cancer.

**Averil Power**

April 2021

## REPORT OVERVIEW

# SUMMARY

The number of individuals diagnosed with cancer annually is on the rise. Inherited genetic mutations play a major role in about 5 to 10 percent of all cancers, though the contribution to individual cancers varies widely. A proportion of cancers are familial and involve mutations of multiple susceptibility genes that increase an individual's risk of cancer. Researchers have associated mutations in specific genes with more than 50 hereditary cancer syndromes.

The assessment of an individual's genetic profile plays a critical role across the continuum of cancer care from screening to the use of targeted therapies. A large proportion of the work of any cancer genetic service is the management of familial colorectal, breast and ovarian cancer, and these areas exemplify opportunities for increased access to gene testing and follow-up support in the first instance.

A reduction in the life-time burden caused by cancer can be achieved by implementing enhanced surveillance and timely evidence-based interventions. Even with improvements in the understanding of the role of genetic information in cancer care, health care providers globally face many challenges in providing uniform access to timely genetically guided health and oncology care. Progress towards more individualised and family-centered oncology care requires enhanced understanding of genetic and genomic information by patients, their health care providers and policy makers.

It is apparent from engaging directly with service users that waiting lists exist at every point on the pathway for people who need genetic services. For those who may have a genetic risk of cancer, the wait times for access to testing alone (before counselling treatment, prophylactic surgery etc.) can be up to 2 years. Barriers to accessing cancer genetic services include costs of tests, long processing time for referrals to tests, restrictive referral criteria, and difficulty in accessing information on cancer genetic services.

Additionally, there exists a lack of data on population statistics for mutations which can impact on the services' ability to plan and to scale. In order to move forward and ensure that people can get the best available treatment, we must also think about a systems level approach towards supporting decision-making, ensuring the right data and resources are in place, and crucially that not one single person is left behind.

Many recommendations were outlined by participants in this study. The recommendations reflect the general direction of the narrative outlined within the Irish National Cancer Strategy 2017-2026.

### Recommendations

1. Implement a hub and spoke model with genetics expertise within the dispersed oncology system. Genetics needs to be formally integrated into the cancer treatment pathway with uniform access to genetic testing, molecular tumour boards and access to genetics expertise and support at the point of care for both patients and their clinicians.
2. Build and further develop the genetics workforce and capability.
3. Increase cancer genetic diagnostics capability and expertise in Ireland.
4. Use a data management system that tracks referrals, appointments, and receipt of diagnosis with associated key performance indicators in terms of time to appointments, time to receipt of genetic test results and time to receipt of follow-up interventions (if required).
5. Streamline the genetics pathway to optimise online data collection and processing of data ensuring that follow-up counselling and health promoting interventions for individuals with positive mutations is optimised.
6. Increase knowledge and awareness of health care professionals, patients and the public of genetics and genetic services.
7. A dedicated pathway for individuals with specific syndromes or mutations with audited quality assured key performance indicators is required e.g. BRCA, Hereditary Breast and Ovarian Cancer Syndrome, Lynch Syndrome. Such pathways will ensure coordination of timely access to evidence-based surveillance, screening, surgery, and treatments as needed for individuals with specific mutations.
8. Test interventions that support the communication of information relating to genetic mutations with family members.
9. Explore and address the barriers to cascade testing of at-risk relatives.
10. Address concerns relating to the management of clinical samples and genetics data.

# EXECUTIVE SUMMARY

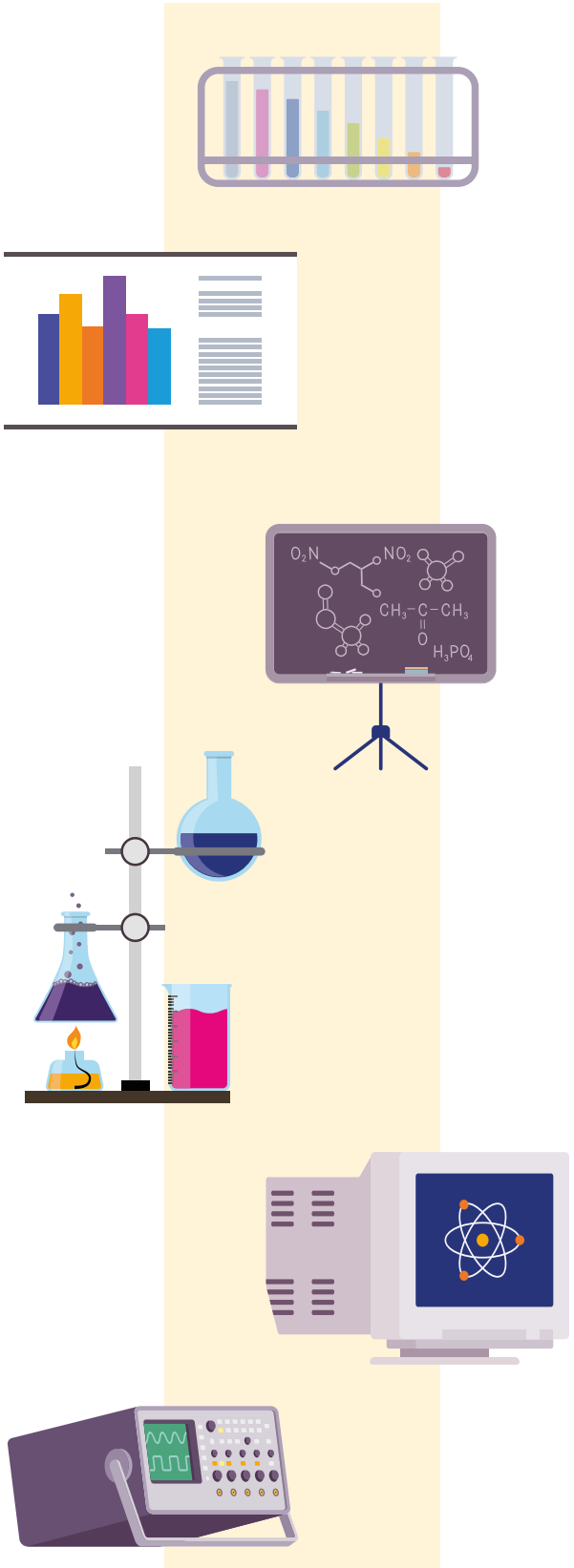
## Background

Cancer is caused by certain changes to genes (genetic mutations) that control the way our cells work, most especially how cells grow and divide. Some of these changes are inherited or familial genetic mutations, which are changes that are passed down through families from one generation to the next. These types of changes may increase a person's risk of developing cancer and are known to play a major role in about 5 to 10 % of all cancers. A patient with a known history of certain cancers or family members of individuals diagnosed with certain cancers can be referred to "Cancer Genetic Services" for the assessment of their cancer family history, and potential suitability for cancer genetic testing. Such genetic testing can: help an individual understand whether an inherited health condition may affect them, their children or other family members; show if the individual is at higher risk of getting certain health conditions, including some types of cancer, and guide doctors in deciding what treatments best suit the person diagnosed with certain types of cancer. Cancer Genetic Services can support families with a history of these types of inherited mutations by providing specialist advice. Thus, access to cancer genetic services is an important component of the health services for patients with cancer and their families.

## Method

This study sought to use a multifaceted environmental scanning approach which sought to hear about healthcare professionals, patients, family members and members of the public experiences of cancer genetic services in an Irish context. This was complemented by a systematic review of the international empirical literature.

A mixed methods approach was used with individual interviews (n=21 patients, 15 family members and 15 health care professionals) and online surveys with patients, families, and members of the public (n=154) and health care professionals (n=52).



# IRISH CONTEXT

## Survey data revealed

### Waiting times for genetic testing

One in seven respondents were waiting 13-24 months and one in 27 were waiting over 24 months for their counselling and testing appointments.

Many noted they changed from the public system to the private system to speed up access to genetic testing.

### The highest ranked barriers to accessing cancer genetics services:

Public: worried results could be used against them (over four in ten); referral took a long time to process (one in three); the tests were too costly (one in five); cost/lack of medical insurance cover (one in seven) and difficulty in getting information about cancer genetic services (one in seven).

Health care professionals: services under-resourced (nearly six in ten); lack of services to implement guidelines about follow-up prophylactic surgery for those diagnosed with elevated risks resulting from hereditary cancer mutations (more than four in ten); lack of services to implement guidelines for cancer genetic testing and counselling (nearly four in ten); lack of national guidelines about who should be referred (nearly four in ten ); referrals take a long time to process (more than one in three); referrals poorly coordinated (nearly one in three); lack of services to implement screening/ surveillance guidelines (more than one in four) .

### Waiting times for genetic testing

The cumulative waiting time from referral to counselling, testing, receipt of genetic test results and onwards to screening, surveillance or prophylactic treatments is lengthy (can be four years) which is seen as time lost in terms of cancer prevention and early intervention.

Family members start the process from the time of receipt of communication from the index person.

### The highest ranked facilitators to accessing cancer genetics services:

Public: perception of information benefiting their future (nearly three in four); wanting to know their future risk of cancer (two in three); the importance of being proactive (nearly two in three); information benefiting their family's future (six in ten); going seemed important (more than half); doctor's recommendation (half); having a family history of hereditary cancer (more than four in ten).

Health care professionals: having information resources for patients/family members (half); national guidelines regarding patients that require referral (half), medical insurance cover (nearly four in ten) and medical card cover (nearly one in three).

## Qualitative data revealed:

| | |
|---|---|
| Access to cancer genetic services is suboptimal | Advantages of earlier and uniform access to genetic counselling, testing and results include information can be used to inform surgery and treatment options, earlier cancer prevention conversations and access to surveillance and screening.<br><br>However, the process of accessing genetic testing varies. The waiting times for accessing genetic testing and results can be greater than two years for some. Many patients whilst waiting seemed to abandon the waiting and sought a private appointment if they had the financial resources or health insurance cover.<br><br>Criteria used for referral to genetic testing was viewed as being overly restrictive. |
| Experience of patients and family members | The model of family has changed and there was variation in experience of sharing information with family members.<br><br>Communication of positive genetic test results, a key initial step, presents its own challenges.<br><br>The time waiting for genetic test results was associated with increased anxiety and the period after being told about a particular mutation involved adjustment to having a genetic mutation and a new normal. |
| Access to follow-up support and care | Lack of uniform approach to the coordination of follow-up services and support was evident.<br><br>Current services are dependent on the individual with the mutation being proactive and seeking out services/follow-up as opposed to being offered automatically as part of a predefined pathway to all requiring it.<br><br>The need for dedicated syndrome or mutation specific pathway and clinics was articulated. |
| A preference for a hub and spoke model and an integrated genetics pathway | A preference for an integrated hub-and-spoke network design was articulated with genetics expertise available within the dispersed oncology system.<br><br>Mainstreaming as an approach was highlighted as a mechanism of ensuring that patients had uniform, criteria-based access to cancer genetics. |
| Barriers and facilitators to accessing cancer genetic services | Knowledge and awareness about cancer genetics and cancer genetic services needs to be enhanced across the oncology workforce and among GPs.<br><br>There needs to be development and expansion of the cancer genetics workforce.<br><br>Inequalities in access to cancer genetic services were evident. |
| Management of samples and data | Concerns re management of samples and genetic data emerged as a concern for some participants. Issues were raised with the use of multiple laboratories for genetic testing.<br><br>A vision for a state wide database that logs all of the individuals that come to see genetic services, linking up families to the pedigrees and integrated with the main hospital system to link clinical data was articulated by some. |

***One participant noted:***

***"-- [need] much better counselling and facilitating as to what this means for the wider family. Sending the person who has been tested and confirmed to have an inherited genetic mutation off with letters under the arm to hand out to other family members is not sufficient. It is like throwing a scud missile into the middle of the family and it can have all kind of consequences. This can be particularly difficult if the person tested and diagnosed is sick and going through treatment".***

# INTERNATIONAL CONTEXT

## Systematic review objectives

- To identify barriers and facilitators to accessing cancer genetic testing and counselling.
- To identify disparities among populations in terms of access to cancer genetic testing and counselling.

| Systematic review (n=122 included studies) revealed: | |
|---|---|
| Barriers to accessing cancer genetic counselling and testing (CGTC) | Individual-level barriers were most prevalent with the cost of genetic testing as the overarching barrier (n=35.9%). Additional important barriers included fear of positive results and their impact on insurability and employment (26.1%), insurance concerns (25%), and lack of knowledge and awareness regarding cancer genetics and services (21.7%).<br><br>Among the service-specific barriers, a lack of referral/ recommendation (10.9%) coupled with insufficient knowledge and awareness of CGTC and genetics in general among HCPs (9.8% and 7% respectively) were mostly reported.<br><br>The most commonly reported barrier at a national level was the geographical location of CGTC centres (15.2%), followed by a lack of genetic services and genetic workforce (5.4%), and difficulty navigating the healthcare system/systemic barriers (n=4.3%).<br><br>Ethnic disparities in access were frequently reported (n=21). |
| Facilitators to accessing cancer genetic counselling and testing | Individual-level facilitators were the most frequently reported. Knowledge of familial history (25.9%), proactive health attitudes and beliefs (n=20), and family obligation, responsibility, and support (24.7%) were the facilitators most often reported at an individual level.<br><br>In terms of service-specific facilitators, the use of alternative methods to deliver counselling, such as the web or telephone (7.4%), HCP access to training on CGTC (6.2%), access to professional/national guidelines on cancer genetic testing (4.9%), awareness of CGTC and benefits (4.9%), and strategies to facilitate access to and efficiency of appointments (4.9%) were identified as the most important facilitators.<br><br>The most common national level facilitators were positive health behaviours among socially influential individuals (6.2%), national awareness campaigns (4.9% and availability of information in different languages (3.7%). |

## Summary

The assessment of an individual's genetic profile plays a critical role across the continuum of cancer care from screening to the use of targeted therapies. A large proportion of the work of any cancer genetic service is the management of familial colorectal, breast and ovarian cancer, and these areas exemplify opportunities for increased access to gene testing and follow-up support in the first instance.

A reduction in the life-time burden caused by cancer can be achieved by implementing enhanced surveillance and timely evidence-based interventions. Even with improvements in the understanding of the role of genetic information in cancer care, health care providers globally face many challenges in providing uniform access to timely genetically guided health and oncology care. Progress towards more individualised and family-centred oncology care requires enhanced understanding of genetic and genomic information by patients, their health care providers and policy makers.

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There exists a lack of population level data for mutations which in turn impacts on the services' ability to plan and to scale up interventions. In order to move forward and ensure that people can get the best available treatment, we must also think about a systems level approach towards supporting optimum decision-making, ensuring the right data and resources are in place, and crucially that not one single person is left behind.

# CHAPTER 1: MIXED METHODS APPROACH USED TO OUTLINE THE CANCER GENETIC SERVICES AVAILABLE IN AN IRISH CONTEXT AND HEALTH CARE PROFESSIONALS', PATIENTS' AND FAMILY MEMBERS' EXPERIENCES OF ACCESSING AND USING SUCH SERVICES.

## BACKGROUND

Genetics and genomic medicine have helped revolutionise our understanding of cancer aetiology and offers new and exciting prospects in terms of personalised medicine. Worldwide health services are developing, sharing and integrating advances in genomic technologies and knowledge into routine clinical practice which ultimately benefits patient care. Through genetic counselling and testing health care teams seek to assist individuals with cancer and their families to make decisions facilitating cancer prevention in some cases, promoting earlier cancer detection and providing more targeted cancer treatments.

To ensure a systematic and uniform translation of cancer genetics into clinical practice it is important that we understand the experience of cancer genetics in an Irish context.

***Thus, this research seeks to outline the cancer genetic services available in an Irish context and provide detail on health care professionals', patients' and family members' experiences of accessing and using such services.***

## DESIGN

A mixed methods approach was used with individual interviews and online surveys being conducted concurrently, using a (non-probability) volunteer sampling strategy.

Environmental scanning refers to the collection and utilisation of information regarding events, relationships, and patterns and trends in an area, and the use of the acquired knowledge in shaping the future service delivery plans and objectives. It uses information to formulate a picture and an understanding of service delivery patterns and potential obstacles and facilitators.

### SAMPLE

The sample included:

- Multidisciplinary team members involved in the delivery of services to patients receiving treatment for cancer and individuals who have a coordination or management roles in relation to the organisation of cancer or cancer genetic services within the Irish context.
- Individuals who have/have had cancer who have sought access to cancer genetic services in an Irish context.
- Members of the public who have sought access to cancer genetic services in an Irish context based upon concerns regarding inherited or familial genetic mutations.

### SURVEY

The patient/member of the public survey sought information about the person's age, gender, nationality, details of hereditary cancer syndrome/mutation, details of cancer genetic services accessed and factors that made it easy or harder to access such services, and satisfaction levels with genetic services. The health care professional survey sought information about the person's profession/speciality, gender, nationality, factors that made access easier, more difficult and comments regarding future developments in the service. The listing of barriers and facilitators in both surveys was informed by the review of literature and a pre-existing survey (Anderson et al., 2012).

Details of the online survey were accessed through email links or via the Irish Cancer Society website. Key professional contacts were asked to distribute an invitation email to participate via multiple channels e.g. through professional networks, Irish Cancer Society website and contacts, cancer support and advocacy groups, with individuals asked to arrange for their onward distribution and communication within their networks. If individuals were interested in partaking in the survey, they were advised to click on the survey link provided which took the individual to the consent form and online survey. Access to the survey was permitted once the person indicated their consent to participate in the online platform. Online survey logic allowed the participant to skip non-relevant questions as needed.

Quantitative data were analysed using SPSS and presented as percentages, means (SD), median (IQR) as appropriate.

## QUALITATIVE INTERVIEWS

Participants who indicated an interest in participating in an interview via their survey response or via an email to the research team or via mobile text were subsequently contacted.

A semi-structured interview schedule guided the interview process. Participants were asked to identify the main genetic services accessed and their experiences of accessing such services. Participants were also asked to outline their support, information, follow-up, and care needs. Barriers and facilitators to accessing such services were discussed. Subsequently, participants were asked to consider the future optimum genetic services and pathways to such services.

Each interview was audio-recorded. Notes were also kept by the researcher. Qualitative data analysis was iterative and began immediately after the first interview, such that, analysis of early interviews informed the content of future interviews.

Qualitative data analysis used latent content analysis, which refers to analysis of the underlying meaning of the text. Methodological trustworthiness was maintained by 1) audit trail 2) peer debriefing and 3) maintenance of a reflective diary by the researcher(s).

Potential barriers to the use of genetic services, were defined as anything that currently impedes patient access to genetic services or that which introduces variability into a process or system that would cause decreased accessibility of, efficiency of, timeliness of, or inequitable access to genetic services in an Irish context. Potential facilitators of the use of genetic services, were also identified.

## ETHICAL AND DATA MANAGEMENT CONSIDERATIONS

Ethical approval was sought and granted from the Clinical Research Ethics Committee Cork and other relevant stakeholders as deemed appropriate. All individuals were reassured that the information that they provided would remain confidential.

Full study information (information leaflets) was provided in all correspondence. Written consent was attained from all interview participants. Online consent using a tick box mechanism accompanied by a declarative statement was obtained for all survey participants. The team used the screening/survey platform functionality within online surveys to direct participants away from the survey if they indicated that they did not consent. The IP addresses were not collected by the survey tool. All responses (once the study is complete) will be deleted from the online survey platform. The resulting data file that is used for data analysis is free of any identifiers, including IP addresses or other electronic identifiers.

Data management is in line with published GDPR national guidance. All data are anonymised and stored in electronic format in UCC on a password protected device for ten years. All sociodemographic data was coded, and anonymity will remain in place for participants. All transcripts were anonymised.

***In summary, a mixed methods design was used to collect concurrent qualitative (interview data) and quantitative (survey data).***

# SURVEY RESPONSES (patients, family members, members of the public)

The survey was completed by 154 individuals representing "patients, families, and members of the public" and 52 health care professionals. The majority were female, of Irish nationality and located in Leinster (Table 1).

**Table 1 Sociodemographic characteristics of sample that completed the survey**

| Variable | Details | Patient, family, and public perspectives (n=154) % | Health care professionals (n=52) % |
|---|---|---|---|
| Gender | Male | 7.4 | 9.6 |
| | Female | 80.7 | 90.4 |
| | Other | 1.9 | |
| Ages (years) | 18-24 | 1.9 | NA |
| | 25-34 | 17 | |
| | 35-44 | 39.6 | |
| | 45-54 | 30.2 | |
| | 55-64 | 7.6 | |
| | 65-75 | 1.9 | |
| | 76-85 | 0 | |
| | Over 85 | 0 | |
| | Rather not say | 1.9 | |
| Nationality | Irish | 88.9 | 92.3 |
| | Other | 9.3 | 7.7 |
| | Rather not say | 1.9 | 0 |
| Public and private health insurance | Have a medical card | 28.3 | NA |
| | Have a GP visit card | 0 | |
| | Have private health insurance | 52.8 | |
| | No medical card/GP card/private health insurance | 20.7 | |
| | | 3.8 | |
| Region | Leinster | 54.8 | 61.5 |
| | Munster | 29.2 | 25 |
| | Connaught | 15.4 | 9.6 |
| | Other | 1 | 5.8 |

| Variable | Details | Patient, family, and public perspectives (n=154) % | Health care professionals (n=52) % |
|---|---|---|---|
| Health care professional-profession or specialty? | Advanced Nurse Practitioner | NA | 7.7 |
| | Clinical Nurse Specialist | | 23.1 |
| | Nurse | | 20.9 |
| | Oncologist | | 11.5 |
| | Surgeon | | 5.8 |
| | Genetics Counsellor | | 5.8 |
| | Consultant | | 15.4 |
| | Support worker | | 9.6 |
| Patients, families, members of the public | Diagnosed with cancer | 36.1 | NA |
| | Family member of an individual with hereditary cancer | 16.7 | |
| | Diagnosed with a hereditary risk of cancer | 20.8 | |
| | Member of the public undergoing or has undergone testing for hereditary cancer risk | 6.9 | |
| | Other (some of whom had sought access to genetic services and not been successful in their request (n=5)). | 19.2 | |

For the patients, family members, public group who highlighted the details of the hereditary cancer (n=98) details included: 41.9% ticked either hereditary breast cancer or hereditary ovarian cancer or ductal carcinoma in situ (DCIS) or triple negative breast cancer or hereditary breast and ovarian cancer (HBOC) syndrome; 20.5% ticked either hereditary colon cancer/ colorectal cancer or familial adenomatous polyposis or Lynch syndrome (hereditary non-polyposis colorectal cancer); 2.1% hereditary kidney cancer; 4.1% blood cancer; 2.1% appendix cancer, 3.1% hereditary prostate cancer; 13.3% known hereditary cancer susceptibility syndrome; 2.1% Muir Torres syndrome, mother of a child with alveolar rhabdomyosarcoma (1%), retinoblastoma (1%), not known yet (5.2%), panel test too limited, nothing found (1%), other (4.1%) (some respondents ticked more than one option).

## CANCER GENETIC SERVICES ACCESSED

When asked which cancer genetic services respondents (in the patients, family members and public category, (n=124)) had accessed, the majority ticked counselling (74.3%) and genetic testing (91.2%), whilst 60.5% noted they received a clear plan of action (e.g. follow-up screening requirements) and 66.2% had risk-reducing specific surveillance (e.g. screening) and 29.9% had risk-reducing prophylactic surgical interventions (Table 2, Figure 1).

Nobody ticked the options "I have cancer and I have had genetic testing of cancer (tumour) cells for possible genetic changes" or "I have had molecular and genetic profiling of tumour cells to guide specific targeted therapy" which may indicate that either these options were not clear to respondents or that these details are not routinely shared using the language used in the questions.

In terms of time from referral to having genetic testing: 33% and 42% of respondents respectively had their genetic counselling and testing appointments within six months (Table 2); 25.9% and 21.4% noted they got their counselling and testing appointments in 7-12 months. Some participants (7.4% and 18%) were waiting 13-24 months and more than 24 months respectively for their genetic testing appointments.

***Many noted they changed from the public system to the private system to speed up access to genetic testing.***

On a scale of 0 (extremely dissatisfied) to 100 (extremely satisfied), participants were asked how satisfied they were with their experience of accessing cancer genetic services in an Irish context? Satisfaction scores were mean±SD (63.9±21.5) and ranged from 25-100; 15% of responses were below 50%.

Respondents were asked when their most recent contact with the cancer genetic services was; 71.3% noted their most recent contact was within the last four years (2017-2020) (Figure 2).

| Service accessed/used | Yes Used service % | 0-6 mths % | 7-12 mths % | 13-24 mths % | >24 mths % | Missing time-frame data % | Not satisfied/ slightly satisfied combined % |
|---|---|---|---|---|---|---|---|
| Genetic Counselling | 73.4 | 33.0 | 25.9 | 3.7 | 3.7 | 33.7 | 10.0 |
| Genetic Testing | 91.2 | 42.0 | 21.4 | 14.3 | 3.7 | 18.6 | 12.0 |
| Received a clear plan of action | 60.5 | 36.0 | 10.5 | 5.3 | 0.0 | 48.2 | 30.0 |
| Risk-reducing specific surveillance (e.g. screening) | 66.2 | 17.7 | 23.5 | 0.0 | 0.0 | 58.8 | 6.1 |
| Risk reducing chemoprevention | 9.7 | 0.0 | 0.0 | 11.1 | 0.0 | 88.9 | 0.0 |
| Risk-reducing prophylactic surgical interventions | 29.9 | 12.5 | 18.8 | 12.5 | 6.3 | 50.0 | 15.0 |
| Surgical breast reconstruction | 24.2 | 15.0 | 0.0 | 15.4 | 7.7 | 61.9 | 25.0 |
| Had a breast implant | 12.1 | 15.0 | 8.3 | 8.3 | 8.3 | 60.0 | 18.2 |

**Table 2 Services accessed, time waiting for service, level of satisfaction as reported by patients, family members and members of the public (n=124)**

Note: Not all participants completed the timeframe and satisfaction questions. % timeline and satisfaction data presented as a percentage of the individuals that used that service. Mths=months.

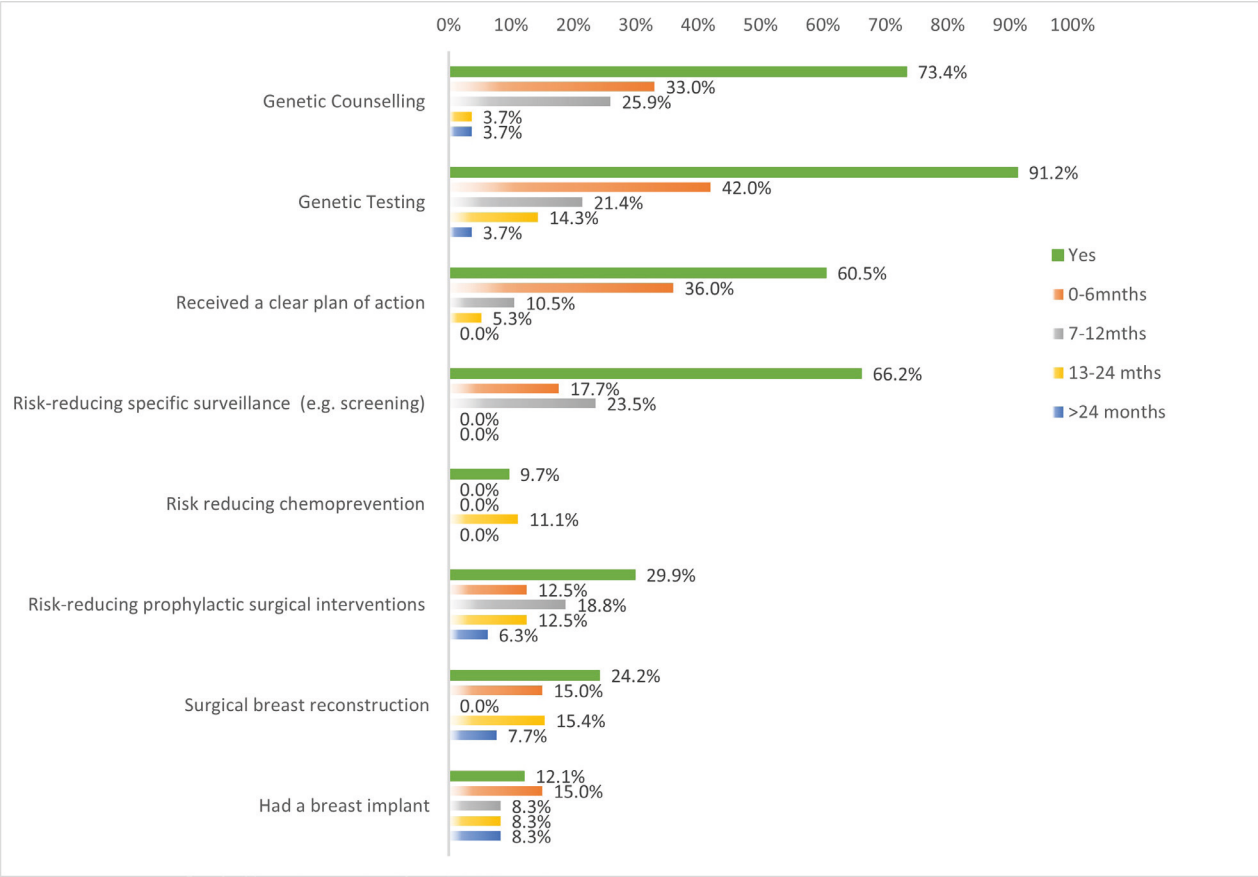



**Figure 1 Services accessed, time waiting for service, level of satisfaction as reported by patients, family members and members**

Participants (in the patients, family members and members of the public category, n=124) ticked "yes" to services received (green) and indicated the timeframe (months) of receipt of such services (orange 0-6 months; grey 7-12 months, yellow 13-24 months, blue > 24 months). Not all participants who ticked yes, completed the follow-on time and satisfaction questions.

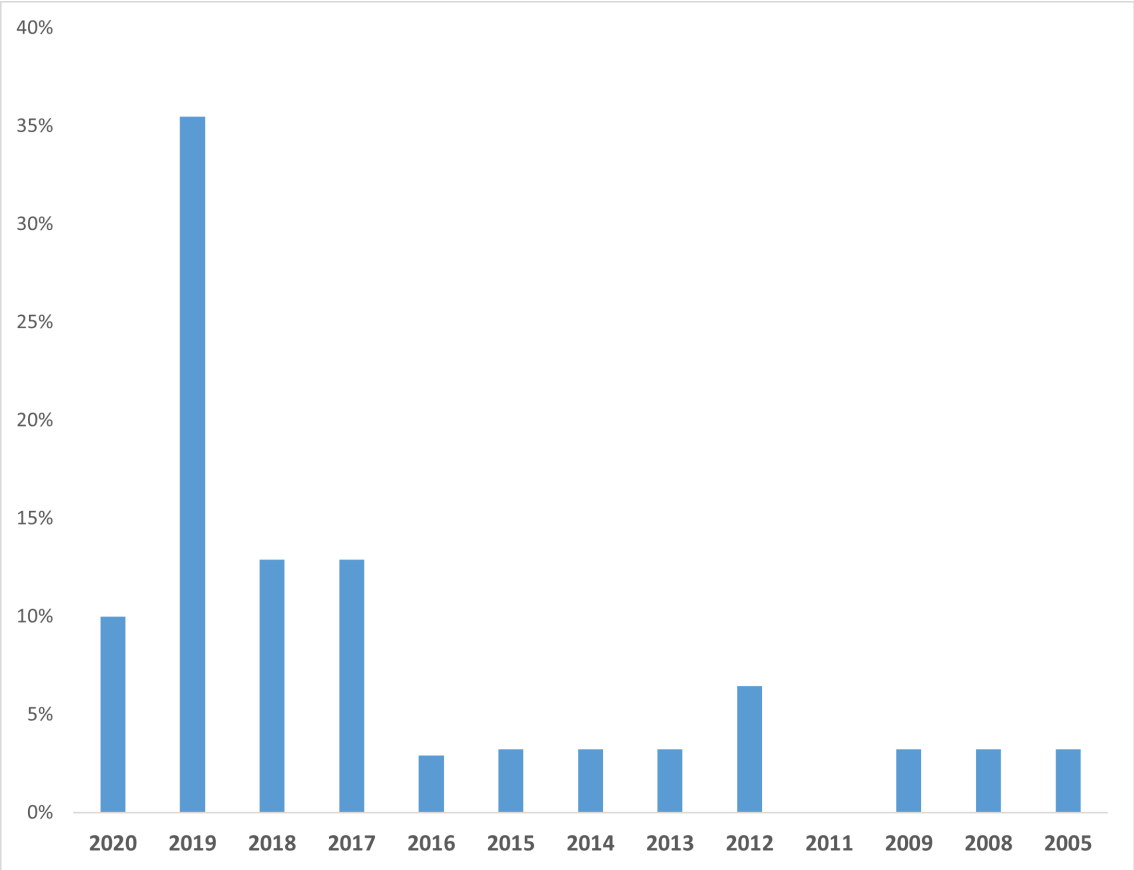


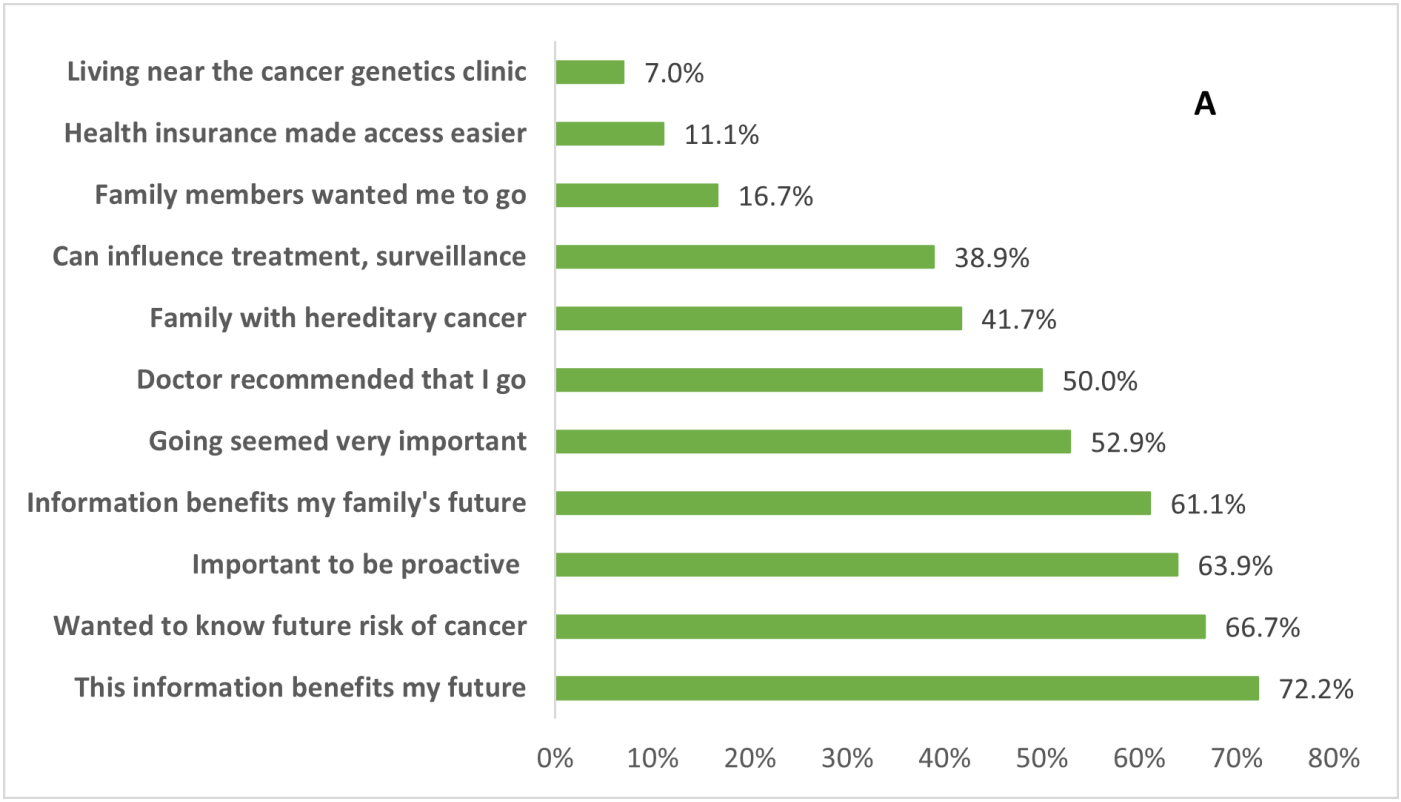

**Figure 2 Year of last or most recent contact- participants (in the patients, family members and members of the public category) reported on their most recent contact with the genetic services (n=122 completed this question)**

## FACILITATORS TO ACCESSING CANCER GENETICS SERVICES

Participants were asked what helped to make their access to cancer genetics services easier.

Participants (in the patients, family members and members of the public category) highest rated facilitators were perception of information benefiting their future (72.2%); wanting to know their future risk of cancer (66.7%); the importance of being proactive (63.9%); information benefiting their family's future (61.1%); going seemed important (52.9%); doctor's recommendation (50%); having a family history of hereditary cancer (41.7%) (Figure 3A).

Health care professionals noted that having information resources for patients/family members (48.1%); national guidelines regarding patients that require referral (48.1%) and medical insurance cover (38.8%), and medical card cover (31.5%) were facilitators (Figure 3B).



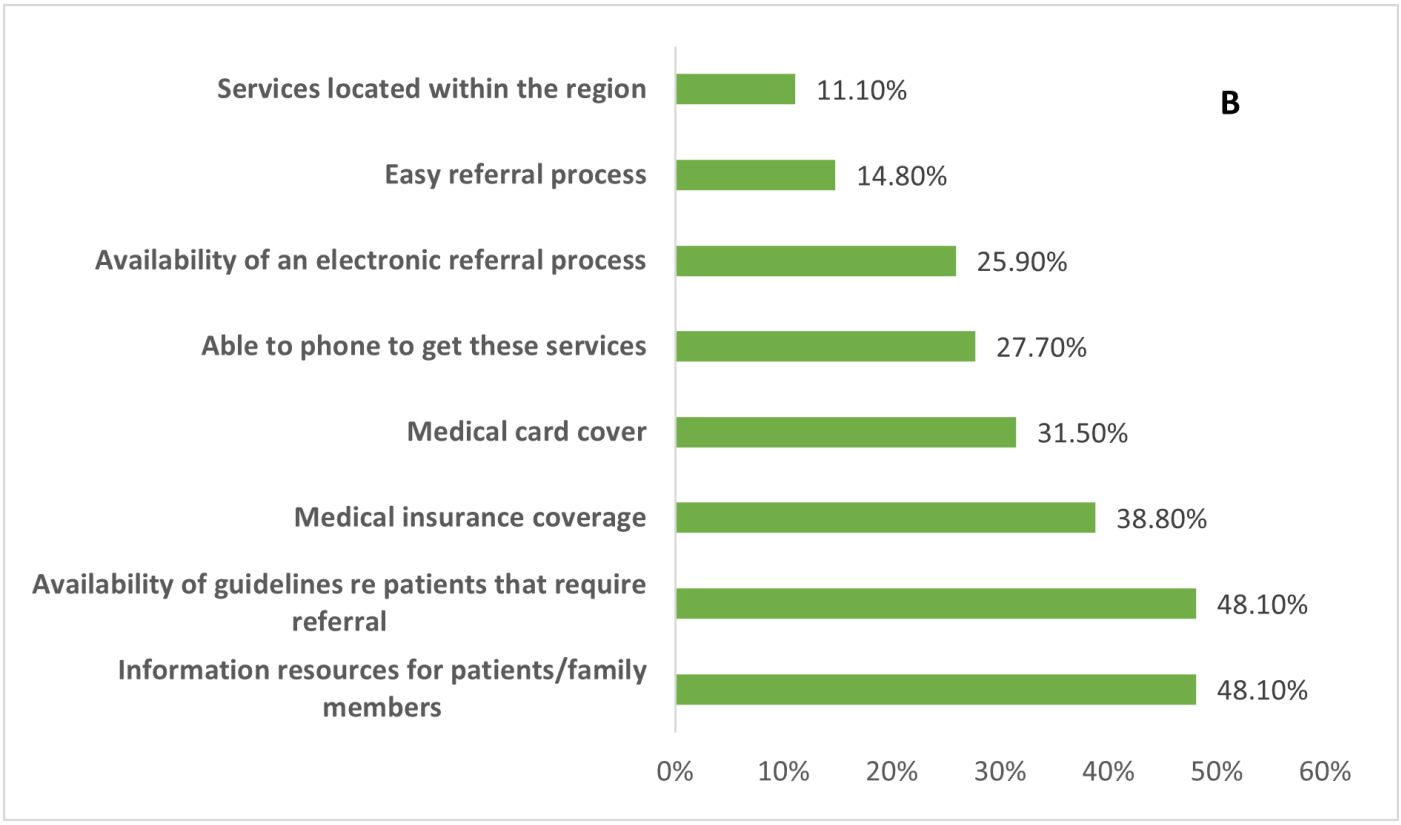




**Figure 3 Facilitators to accessing cancer genetics services, perspectives of A. Patients, family members and the public (n=124) and B. Health care professionals (n=54)**

## BARRIERS TO ACCESSING CANCER GENETIC SERVICES

Various factors were highlighted as potential barriers by respondents. The highest ranked barriers were: worried results could be used against me-by employer, insurance (42.8%); referral took a long time to process (33.1%); the tests were too costly (19.4%); cost/medical insurance cover (14.5%); and difficulty in getting information about cancer genetic services (14.53%) (Figure 4A).

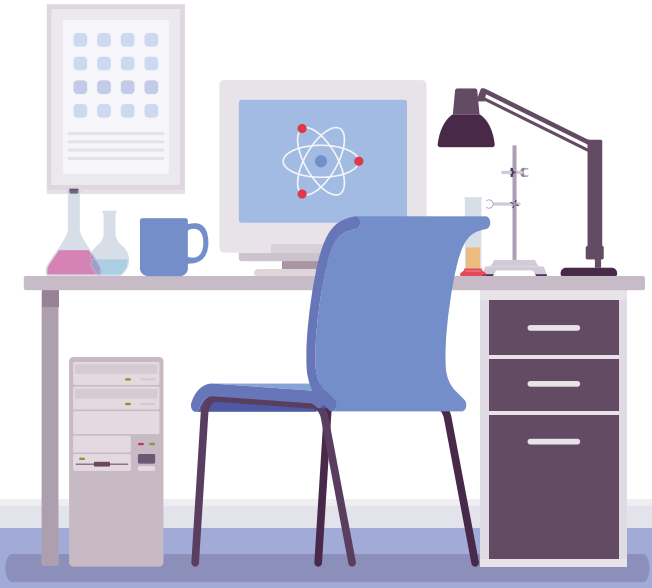
Younger age was highlighted as a barrier in two participants' comments: "I was discouraged multiple times by various people saying I was too young to find out, including a GP. I really don't feel this is true and I have been proactively living a better lifestyle since I found out [about positive genetic test result]".

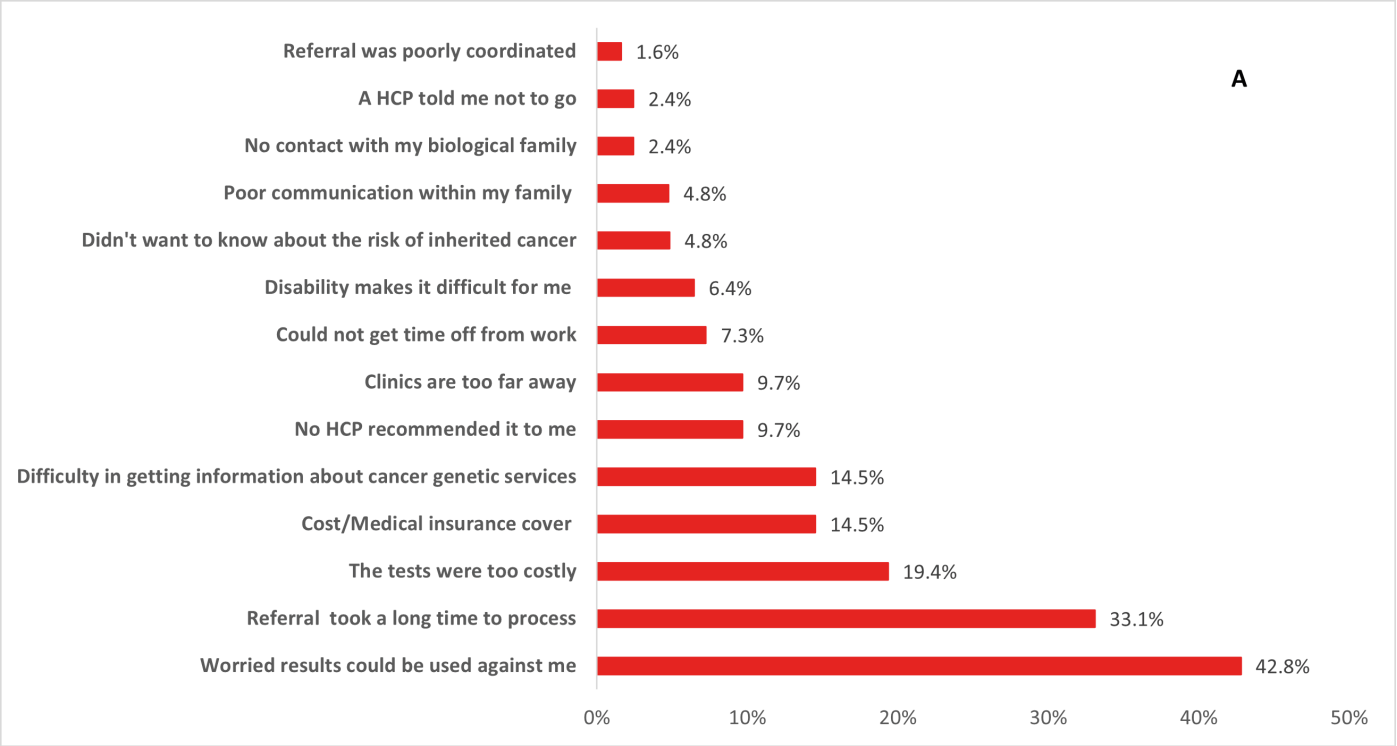
For those who completed the survey but did not use the genetic services (n=11) these were the top ranked reasons cited for not using the service: no one ever recommended it (30.4%); didn't know enough about genetic testing or services (12%); and suggested but didn't go (10%).

Health care professionals cited barriers to accessing cancer genetic counselling and testing as: services under-resourced (57.1%); lack of services to implement guidelines about follow-up prophylactic surgery for those diagnosed with elevated risks resulting from hereditary cancer mutations (42.9%); lack of services to implement guidelines for cancer genetic testing and counselling (38.8%); lack of national guidelines about who should be referred (38.8%); referrals take a long time to process (34.7%); referrals poorly coordinated (30.1%) (Figure 4B).

***In the qualitative commentary, within the survey, one participant noted "the wait times for public testing were too long and my medical insurance did not cover the test. I paid to have the test results sent to Germany so I could make a quick decision regarding surgical options (unilateral or bilateral mastectomy?)"***

***Another participant noted "what will happen in the future re insurance and career if the data gets into the wrong hands and there is a risk that this has already happened".***






Many options had no response (not perceived as barriers) including being too busy; lack of transportation; childcare; other life issues; cultural or religious beliefs; lack of awareness; feeling access would be futile; lack of trust in the health system and Covid-19. HCP= health care professional.

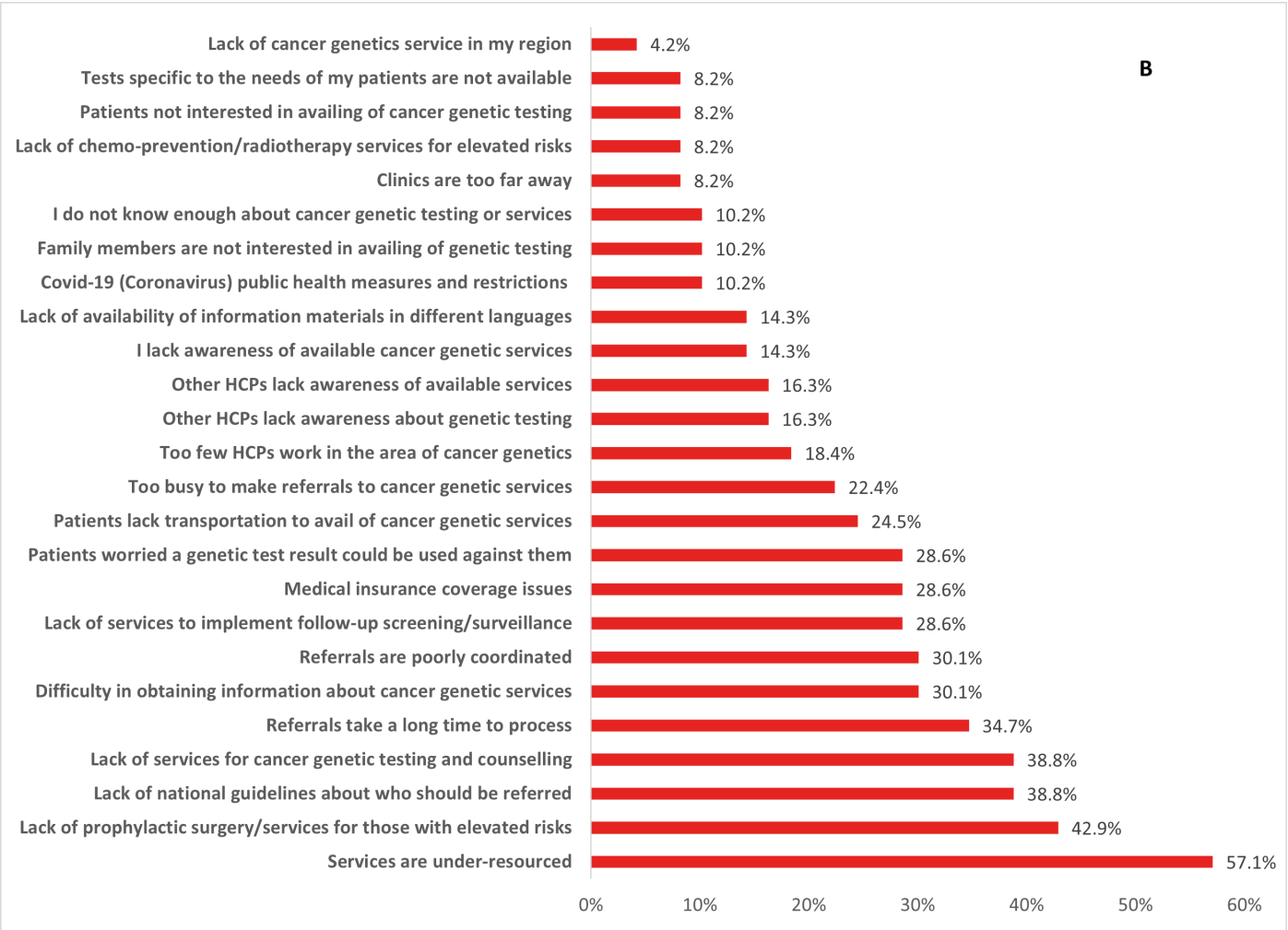



**Figure 4 Barriers to accessing cancer genetics services, perspectives of A. Patients, family members and the public (n=124) and B. Health care professionals (n=49)**

## OPEN ENDED SURVEY QUESTIONS

Open ended questions were asked where respondents could make additional comments, some of these comments have been integrated into the themes in the qualitative write up. One question related to "suggestions regarding potential changes that could be made to referral and access pathways to cancer genetic services in an Irish context". Most of the comments related to:

1. Reducing the waiting times with specific reference to the time it takes to get the results after the initial appointment by increasing the fiscal and human resourcing of the genetics service.
2. Recommended someone accompany the individual to get their results.
3. More follow up for patients to check in on how they are absorbing the information.
4. Support with communicating with the family.
5. Increased support and updated information provided to GPs (patients first point of contact) to aid identification of those who might benefit from referral to a cancer genetics clinic.
6. Overly restrictive criteria for access to cancer genetic testing.
7. Need to increase awareness of health care professionals of cancer genetics and cancer genetic services.

One respondent noted: "The communication of my test results was a frustrating aspect of my experience (both the tumour and blood results). It required a level of proactivity on my part that was very challenging at the time as I was recovering from surgery and undergoing chemotherapy".


***One particularly powerful comment read as: [need] much better counselling and facilitating as to what this means for the wider family. Sending the person who has been tested and confirmed to have an inherited genetic mutation off with letters under the arm to hand out to other family members is not sufficient. It's like throwing a scud missile into the middle of the family and it can have all kind of consequences. This can be particularly difficult if the person tested and diagnosed is sick and going through treatment".***

***The waiting list for family members is perceived to be long: "waiting lists are quite long, my family are waiting over 18 months for their appointment".***

***Some participants felt they were not able to access testing as there family was small- difficult to show a positive family history: "I was penalised by the system because my father is an only child and my mother only had one brother. I could not ever qualify for public testing".***

## QUALITATIVE SAMPLE CHARACTERISTICS

Qualitative individual interviews were conducted with 21 patients, 15 family members and 15 health care professionals. Most participants were female (patients n=16; family members n=12; healthcare professionals n=13).

Patients included individuals with cancer who also: had BRCA mutations (n=9), Lynch syndrome (n=5), lung cancer mutation (n=1), not offered genetic testing (n=3), and some were waiting results of testing (n=3).

Family members included: those with BRCA mutations (n=4), Hereditary Breast and Ovarian Cancer (HBOC) syndrome (n=2), Lynch syndrome (n=3), those testing negative (n=2), have uncertain mutation (n=1), and some were waiting genetic counselling appointments (n=2) and results of testing (n=1).

Health care professionals included: oncologists (n=4); consultant with special expertise in genetics (n=1), genetic counsellors (n=2), surgeon (n=1), nurses (n=6), support worker (n=1).

Data were organised around several themes reflecting the focus of the research aim (Table 3). For each theme, quotes are provided that exemplify how the participants framed the discussion about these themes.

| Themes | Subthemes |
|---|---|
| Access to cancer genetic services | Process of accessing genetic testing<br>Waiting times for accessing genetic testing and results<br>Perspectives on access to counselling and testing |
| Experience of patients and family members | Model of family has changed and variation in experience of sharing information<br>The anxiety associated with being tested and the period of adjustment to having a genetic mutation |
| Access to follow-up support and care | Lack of uniform approach to the coordination of follow-up services and support<br>The need for dedicated syndrome or mutation specific pathway and clinics |
| A preference for a hub and spoke model and an integrated genetics pathway | A centralised diagnostic service versus an integrated hub-and-spoke network design<br>Mainstreaming as an approach |
| Barriers and facilitators to accessing cancer genetic services | Knowledge and awareness about cancer genetics and cancer genetic services<br>Funding and workforce<br>Inequalities in access to cancer genetic services |
| Management of samples and data | Concerns re management of data and samples<br>A vision for a state-wide database |

# ACCESS TO CANCER GENETIC SERVICES

## PROCESS OF ACCESSING GENETIC TESTING

Access to cancer genetic services and investigations for hereditary cancer syndromes in an Irish context is predominantly through clinical genetic services at two hospitals St James Hospital, and the CHI Crumlin Hospital. Genetic counselling and testing can also be accessed through a number of private clinics. Direct-to-consumer DNA tests are becoming increasingly available.

In addition, some physicians access genetic testing for their patients as part of a clinical trial and some genetic tests may be supported by pharmaceutical companies as part of the access protocol to a particular medication. Having information regarding genetic mutations helps plan the treatment, surveillance, and prevention strategies for many hereditary cancers. For example, germline and tissue BRCA testing is needed prior to prescribing PARP inhibitor olaparib[2] which can be used in the treatment of platinum-sensitive relapsed BRCA mutated (germline and/or somatic) high grade serous epithelial ovarian cancer. The NCCP have a procedure for the ordering of inherited (germline) and tumour (somatic) BRCA gene mutation testing by medical oncologists to inform the decision to use the PARP inhibitor olaparib in selected patients.

The NCCP National Executive Management Team provides leadership and governance for the National Cancer Control Programme (NCCP); the NCCP Executive Management team includes the National Clinical Programme Lead in Cancer Genetics.

## WAITING TIMES FOR ACCESSING GENETIC TESTING AND RESULTS

Earlier and uniform access to genetic counselling, testing and results was seen as a positive by all participants. Advantages were seen as: information can be used to inform surgery and treatment options; earlier cancer prevention conversations and access to surveillance and screening.

In the Irish context most participants articulated the fact that they had a conversation with their GP or consultant and having a referral sent to the genetic services. Wait time for the initial genetic services consultation varied substantially from one month to over two years.

Reasons for moving from the public system to the private system were cited as: "I just wanted to get it done and have that box ticked"; "just prefer to know"; "anxious about it"; "gives people reassurance whether there is some other target for treatment or not". In addition, some participants noted that "Covid19 is causing such a back log in the public system".

Most individuals interviewed seemed to have the consultation and bloods completed during the one visit. The wait time between the initial consultation and the final genetic test results in the public system were lengthy in some circumstances with wait times of between two months and over one year articulated.

The follow-up test results were mostly communicated over the phone with a follow-up letter sent to the individual and their nominated physician, however participants also noted they had the option of attending in person if they desired. The level of satisfaction with the actual service delivered and the persons delivering the service in the Irish context was high. One survey participant noted: "I received excellent genetic counselling in the public and private systems [named services]. The staff were very knowledgeable and thorough. I was informed of the demand for the service but was seen in a relatively timely manner. That required follow up on my oncology nurse's part in order to facilitate an appointment".

***"Yeah so I was diagnosed with breast cancer in [month] 2019 and I was referred on to the cancer genetics department in [Dublin, public] Hospital because my Oncologist felt because of my age I was thirty seven they would test me for the BRCA gene -- so I was only waiting a month for my appointment--- I was only waiting over two months for the results but I asked for the results to come back as quickly as possible because -- it would have determined what kind of surgery I needed-- I had a BRCA2 mutation then it was recommended that I have a double mastectomy-- so I had quite a positive experience with them [genetic services] because I would have heard people are waiting months for these appointments. My brother and dad are waiting over a year for their appointments as well because they [brother and dad] both don't have health insurance so they are waiting on that public appointment so they wouldn't have quite a positive experience in that line." [female with breast cancer].***

***"I was referred -- around the end of April [2020] -- through the public system but I didn't get any [appointment], I was on the waiting list and didn't hear anymore though in August I asked the team to refer me to see [name of consultant] privately instead, so I got that referral and I saw him in the [name] clinic in September so I had met the specialist nurse, done the family tree about cancers and I have had the initial consultation on the blood test so I am waiting to get the results, takes about three months" [female with breast cancer, family history of breast cancer, 38 years]***

***"The list is not a true reflection of a list because people come off that list because they go privately because of the length of time they are waiting, they just can't bear the wait and they have an ability to organise it privately" [male with Lynch Syndrome]***

[2] *NCCP Chemotherapy Regimen Olaparib Monotherapy available at: https://www.hse.ie/eng/services/list/5/cancer/profinfo/chemoprotocols/gynaecology/olaparib-monotherapy.pdf and at https://www.hse.ie/eng/services/list/5/cancer/profinfo/medonc/sactguidance/brca%20testing%20for%20olaparib.html*

Another survey participant noted: "I was very impressed with the clear information and advice provided by my genetic counsellor prior to undergoing genetic testing. The information received helped me to make an informed decision. Follow up to explain my results & offer relevant support was excellent. However, the very long waiting periods for initial appointment and for the results of my genetic testing caused anxiety & stress. The lengthy wait also affected my identical twins' treatment for an unrelated medical condition as her biologic was stopped until our cancer risks were better understood". A survey participant noted: "I was waiting too long to get results back; so, missed a dose of extra chemotherapy drugs".

Two participants referenced wait time limits in other jurisdictions that there were service specific KPIs for example the UK National Health Service (NHS) wide policy is that an individual should be offered an appointment within 18 weeks of the referral being received; with some specific services having a target of offering an appointment within six weeks and 97% of referrals hitting that target.

There was a perception that the criteria applied for deciding who gets access to genetic testing being too narrow. One female participant noted: "I was refused the first time as they said there wasn't enough family history. Luckily, my oncologist pushed again [wrote a second referral request] and I got accepted. It turned out I do have BRCA2 and have had preventative surgeries as a result. If I had not have pushed it might be a different story—they [genetic service] need to trust the oncologists as well". Overall, it took over two years from first request to having the result, in the interim she had a lumpectomy, chemotherapy and radiation. Once she received her BRCA2 diagnosis she opted for a double mastectomy with breast implant and a bilateral salpingo-oophorectomy. This lady also noted that some individuals would have accepted the first refusal, she highlighted that she had four daughters and having this information is vital for their future wellbeing.

*"I am seeing delays of between four and five months to get a patient seen. They are trying to streamline it which I think is amazing so they are trying to get the blood sent off beforehand especially if you have a really convincing history-- So they are flexible and they do understand the need to try and move things along but they are just inundated with requests." [medical oncologist].*

*"I found it to be a much longer process than I imagined it would be. The long waiting periods for assessment, counselling/testing and then the results left a lot of time for worry and anxiety to grow" [female with breast cancer].*

## PERSPECTIVES ON ACCESS TO COUNSELLING, TESTING AND SHARING RESULTS

### PRE-TEST COUNSELLING SESSION

There was a perception among a number of participants that the pre-test counselling session could be shortened or adapted to: incorporate use of virtual technology; access to online pre-counselling educational packages or apps or websites; provision of pre-test education reading; streamlined online data collection, use of an online platform to collect, collate and review data regarding family history in an integrated way. Extra personnel such as intake assistants or a family history report agent can speed up the process of completing a family history questionnaire, chasing pathology/histology test results, and thus freeing up the time of genetic counsellors.

The profile of individuals accessing cancer genetic services is also changing thus some participants felt that the collection of a detailed family history could be postponed to after having the genetic test results. One female participant who was BRCA2 positive noted: "it involves digging around to get the history trying to find cancer, trying to justify the need to be tested." In addition, the family structure has altered with much smaller family units making family history of cancer less observable. Whilst others noted difficulties in sourcing this information as the cause of death may not have been discussed openly within a family e.g. breast cancer among males.

### POST-TEST SHARING RESULTS

The post-test conversation could be enhanced by having access to genetic test results, family history, and pathology details. In some international services the genetic counsellor has the conversation (in person or virtually) with the individual whilst they are receiving treatment e.g. "embedded chemotherapy chair time".

For those who test negative, many participants felt that the post test results could be shared by letter (with screening recommendations[3]) ± a telephone call, unless there was some other reason to bring them back. But for those who test positive an in-person appointment is really important, as this is the opportunity to discuss the genetic test results and their implications with the offer of a follow-up contact and an appointment/check-in at one year. One consultant with expertise in cancer genetics said, "that post-test conversation is where you need to hold out the family tree and say your brother is as much at risk as your sister". The importance of offering an open door to the genetics services was mentioned; by keeping the contact details within a registry, this enables one to "blast out emails with updated guidelines easily" which keeps patients and family members updated.

*"--testing now it is more driven by the pathology type of the cancer and [the service] usually see about fifty per cent not having a family history never mind having a [history of a] mutation so it's still important to collect that family history information however you could potentially do that post-test" [female with recent experience as a health care professional within genetic services in Ireland and another country]*

*"So immediate at source testing I don't believe that there is need for a genetics referral unless the BRCA mutation is positive" [medical oncologist].*

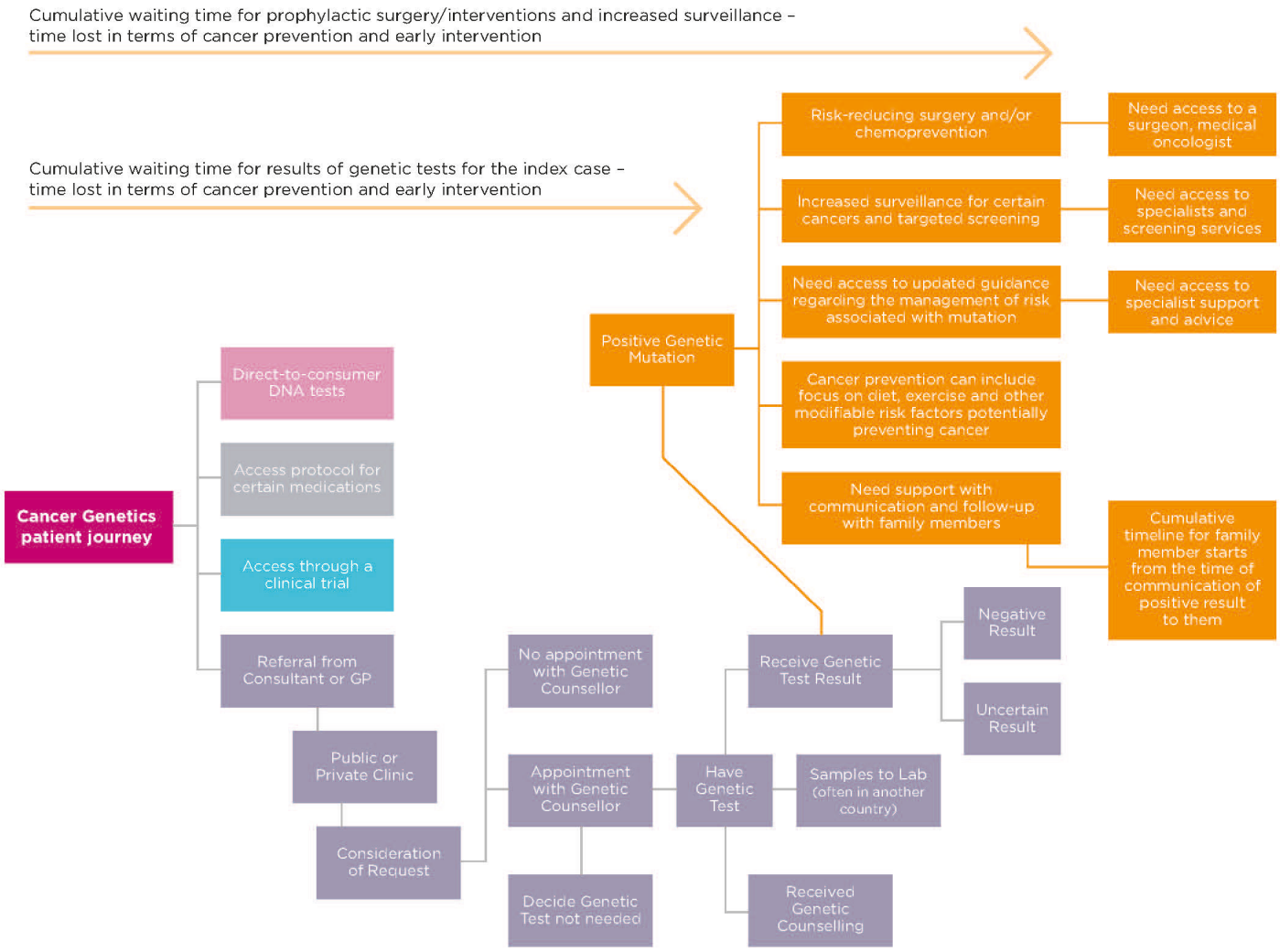



**Figure 5 The individuals' cancer genetics journey**

When accessing genetic tests through a clinical trial where participation in a trial is contingent on having a certain genetic mutation, individuals are tested for a specific mutation and this, whilst serving the purpose of the trial, leaves participants with unanswered questions in terms of "what if wider panel testing was done would it have given a better picture?" This leaves the individual wondering and, for "peace of mind", access to a broader standard panel should be discussed with the person and offered, if relevant [male with lung cancer].

Participants also noted that when having a discussion with their oncologist or surgeon post biopsy or tumour removal that in "addition to the information about the surgery, lymph node status, they would like to hear that there were tumour markers X and Y, genetic mutation Z and we do genetic testing to look for these and in your case this is what we found out for you" [female, 27 years with a cancer of the appendix who sought genetic testing but did not receive it].

Genetic counsellors highlight that most genetic conditions are multifactorial, whereby manifestation of the condition is a result of a complex interaction between genes and the environment. However, the health promotion, disease prevention piece did not really come through within the conversations despite probing.

*The post-test conversation with the index case in the Irish context was mostly over the phone. A woman with breast cancer BRCA2 positive said:*

*"I was happy enough to have the result on the phone. So yeah then I had a couple of queries, sorry she [genetic counsellor] asked me then did I want to go back in after she gave me the result if I had any more questions and I had a couple of things but I used to email her and she used to come back to me and she rang me a few times -----the letter went to my Oncologist and to my GP and a copy came to me and the letter was three pages long and the description of conversations that we had, then the recommendation from the Consultant---- so I knew the steps that I needed to take and you know follow ups ---- got the names for the consultants pancreatic, gynaecological, dermatologist -- but I got the appointments very quick which I was very surprised about to be honest-- So yeah and then the Gynae end of it then I had my ovaries removed in July because of the ovarian cancer risk --- my GP has it [letter] and has probably not read the full ends and outs of it so I have that in the back of my head to follow up on that [dermatologist] myself. So, there is a bit of self-management in there as well that I have to take charge of that".*

[3] *Noting that a negative genetic test result is reassuring, but does not completely exclude a hereditary cancer risk. There is still a need to explain the results, and discuss prevention and screening options if necessary.*

## MODEL OF FAMILY HAS CHANGED AND VARIATION IN EXPERIENCE OF SHARING INFORMATION

Some individuals found having the responsibility to share the details of the genetic test results difficult whilst some families were very open, others had particular family dynamics that made sharing the information more challenging. Family cooperation is often necessary to collect family medical histories, some may have difficulties with sharing this and don't really want to engage in this type of a conversation, as genetic information can affect an entire family rather than only one individual, and can potentially affect the choices of future generations making this a complex area.

Most services globally use a shared decision making model with family members being encouraged to share the information using standard 'open relative letters' with a family reference number/ details of the mutation, very broad health related information and contact numbers being provided. Yet for individuals this creates some difficulties: "who is responsible, is it the individual themselves or should the physicians be involved?" Some individuals feel able to have the conversation "You know when I discovered I had it, I didn't need anybody to tell me that I need to talk to my children--- I mean I have three adult children, two of them have been tested, one is positive and one is negative and the third one in her forties doesn't want to the tested. So, you know it brings up different issues. The lady that is positive has three children she had finished having her children, so she is considering a Hysterectomy" [male with Lynch syndrome].

One lady with BRCA2 mutation noted she had told her wider family "which was a horrible experience - - - some have got tested, others didn't--turns out I am the only one with the mutation". She warned her wider family that this was "not for public news" as she had not told her daughters yet; she highlighted that she would really like to have help with that conversation so she could tell her daughters "this is the plan" and it will be really good to have "someone watching over them should they test positive". She had sought advice and was told that it was best to tell her daughters when they were in their twenties.

As mentioned previously most services globally use a shared decision-making model with family members being encouraged to share the information. However if a person refused to share the information, two scenarios were presented by respondents with experience in multiple jurisdictions: respecting the autonomy of the person whilst highlighting the consequences for that family and/or taking a more active role in contacting the family. Such cases may be discussed at an "MDT meeting" to discuss the options and plan the best way forward depending on the risk and the circumstances of the individual. In some other countries, if it is challenging for the individual to share the information with family members, the genetic service actively collects the contact names and telephone numbers/ email addresses of family members. The genetic service (genetic counsellors) then takes on the responsibility of sharing the information with family. If there is a high index of worry the service may contact the GP. This is a very complex legal and ethical field with multifaceted and possibly conflicting responsibilities which balance the premise that 'genetic information belongs to the index person' versus the 'family members right to have access to health promoting possibly lifesaving information'. A recent legal case[4] also places a duty on the health service to be more proactive in contacting family members and to actively consider whether to disclose confidential information to at-risk individuals when patients refuse consent. In parts of the UK, a carrier register is maintained with details of the cancer predisposition variant and details of persons tested; a follow-up conversation is held with the index case at one year to check if all family members have been contacted and if they have been for testing. A carrier register has many advantages including access to individuals to provide informational updates; it can also be used for research, service planning and workforce planning.

The family member ordinarily receives the letter/information with contact details for follow-up. If the family member chooses to make contact with the genetic services either as a self-referral or via their physician the counsellor then discusses: the process of testing; possible outcomes of the testing and the repercussions for their cancer risk and their health care going forward; potential life insurance implications; considerations if having children (depending on age). Having a family member (e.g. parent) accompany younger adults to the appointment was seen by some as a facilitator and others felt it helped in picking up on the large volume of information transmitted.

The genetic counsellor responding to the family member can be challenged as there is no database that allows for easy tracking of the index person with the mutation in the family nationally and then linking that information up with a sister or brother or relative. Much of that work was done manually via a paper system and limited online processing and sharing of genetic and clinical information. Counsellors can have difficulty in accessing pathology information to confirm the cancer type when they are contacted by family members.

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[4] *Thornton, J. (2020). Judgment in ABC case rules on confidentiality. Lancet (London, England), 395(10226), 771-772.*

*The ruling by Justice Yip in the case brought by the relative of a man suffering from Huntington's disease https://www.theguardian.com/society/2020/mar/01/huntington-disease-ruling-doctors-duty-to-tell-patient-family`*

## THE ANXIETY ASSOCIATED WITH BEING TESTED AND THE PERIOD OF ADJUSTMENT TO HAVING A GENETIC MUTATION SHARING INFORMATION

Having to go for genetic testing was associated with increased anxiety, such anxiety was perpetuated by having to wait longer for the initial counselling session, genetic testing, and the long period of time between testing and receiving the results. A medical oncologist noted "It just hangs like a shadow in the room you know, did you get back my BRCA test, did you not take that five months ago, yeah you know it's still not back". The results were often seen as a relief in terms of "now knowing" but results can also worsen anxiety levels and there is a need for adjustment to this new situation.

Some participants with cancer hereditary genetic mutations felt guilt that they had passed this on to their children. One woman with breast cancer noted that "He [dad] was more upset that he had given me the gene [BRCA2], he felt guilty". Some participants felt there was a stigma associated with a positive genetic result.

The optimum time to communicate with children was a concern articulated by many, one survey participant noted: "I do think it might be a good idea to have a follow up with the genetic counsellor in a year's time, best time to tell your kids, managing guilt etc."

Once referrals are made the counsellor role with that individual generally ends with the door remaining open if needed. However, a continual familial relationship potentially remains with other members in the family.

The need for more support has been articulated to deal with the familial context with some participants mentioning the benefits of the family therapy system in the predictive context. Family system therapy is where you have different families that come in for group genetic counselling, where it's not just the person who has been found to have cancer is seen, thus the counsellor is having a more of a holistic conversation with the family. Such systems could be co-designed with individuals with cancer and their families.

***"Even though I have a background in genetics, I was not prepared for the emotional aspect of this genetic test result. I had convinced myself I would be fine; I knew about genetics and this was good for me. However, it has brought with it a level of anxiety and uncertainty that I have had to have counselling for. I think this is in part because I have not seen a genetic counsellor to talk this through with. The nurses I met and team I met were lovely, but the support systems need to be there for the long run. Furthermore, the mammogram machine in [Dublin hospital name] has not been working for some time now (or so we were told) so my mother has not been screened in over a year and is high risk. We think it is unfair that we were encouraged to have this testing without the proper supports and facilities in place on a constant basis" [female with BRCA mutation].***

## ACCESS TO FOLLOW-UP SUPPORT AND CARE

### LACK OF UNIFORM APPROACH TO THE COORDINATION OF FOLLOW-UP SERVICES AND SUPPORT

Knowledge and understanding regarding one's predisposition to germline cancer enables the adoption of appropriate prevention measures. Cancer prevention can include focus on diet, exercise, and other modifiable cancer prevention behaviours; increased screening; risk-reducing surgery and chemoprevention.

When questioning about the follow-on pathway (i.e. access to screening, prophylactic treatments/surgery, follow-up monitoring a varied picture emerged. In the Irish system, one health care professional noted "there is a big gap after we [genetic services] see them, where are all those supports and all that information that they can access, to then help them make the best decisions for them as a person and their kids." In the Irish system the involvement of a counsellor seemed to end with imparting the genetic test results and with the letter which signposted the individual and family member (with the mutation) and need for follow-up which could be organised though the referring physician or GP. Often the follow-up involves multiple consultants and the difficulty of sharing information across hospitals was highlighted: "--accessing information between different hospitals is extremely difficult. You know it is as if you are dealing with separate entities" [male with Lynch syndrome].

Younger adults were particularly challenged in accessing services as they fell outside of the screening recommendations in some instances and they perceived they were left without a clear pathway and plan for surveillance and follow-up. Two participants managed to secure access to ongoing screening in other European countries through family connections which gave them "peace of mind". In addition some participants from their interaction with healthcare professionals felt they were burdening an overstretched service which was designed for patients with cancer, with one individual with a BRCA mutation being dismissed by a physician "why are you here, I'm seeing cancer patients".

In some jurisdictions the counsellor would coordinate the referrals to the surgeon, oncologist, specialist, screening services, support services that the person needs. For the rare cancer types the counsellor would have to find a specialist who knew about the particular syndrome for example and if the identified specialist didn't know about it, the counsellor would send on information about the condition and its' management. For complex cases the cancer genetics service may continue to maintain a coordination and checklist approach checking the person had received all the required follow-up, the example of PTEN Hamartoma Tumour Syndrome was given as an example highlighting the multiplicity of follow-up requirements associated with the increased risk of breast, thyroid, renal cell, endometrial, colon and melanoma cancer.

Some participants had great support from their physician in organising the follow-up requirements whilst other participants had the opposite experience. One participant noted"--my doctor wasn't interested even though he was a young doctor, he said sure you know more than me, my oncologist wasn't making any discussions about it and I didn't get any indication as to how I would monitor myself going forward". Another noted he found out what the newest approach to managing Lynch syndrome was and then he requested that, rather than the physician informing him: "I had no physician holding my hand" [male with Lynch syndrome]. Individuals referenced the fact that they were reading the newest guidelines and requesting screening, treatments based upon the most up to date evidence: "a few years back, I added a baby Aspirin[5]. So, it involved me asking my doctor to prescribe it for me" [male with Lynch Syndrome]. In many instances there appeared to be no coordinating physician looking at the overall picture.

### THE NEED FOR DEDICATED SYNDROME OR MUTATION SPECIFIC PATHWAY AND CLINICS

The lack of ongoing support was highlighted both in terms of the newest evidence-based approaches to managing individuals with hereditary cancers and the lack of support with the ongoing challenge of communicating details with family members and the need to sometimes support family members to make the decision regarding genetic testing. "I am relying a lot on actually the Marie Keating Foundation they have a great support for BRCA and a lot of information there [website, webinar, monthly coffee morning] and that's where I get all my information about it, you are relying on a charity rather than a hospital for support-- there is no like central resource for BRCA" [female with breast cancer, BRCA2 mutation].

A survey participant noted: "underwent screening for a BRCA2 variant in my family when I was around 21 years old. I did not receive any genetic counselling. I simply received the result, was asked if I had any questions and was told I would need to wait until I was 25 to start any screening. I am 25 now and hoping to begin screening soon. I had questions about hormonal contraception that were not really answered, but I think this is because there is not much research on this topic and it's interaction with HBOC".

Another survey participant who had sisters in other jurisdictions noted: "Following my results of being BRCA2 positive, my family then had the option to be tested. I feel

*[5] NICE committee agreed that aspirin use for at least 2 years should be considered in people with Lynch syndrome. Cited within: National Institute for Health and Care Excellence. (2020) Colorectal cancer (update): Effectiveness of aspirin in the prevention of colorectal cancer in people with Lynch syndrome. NICE guideline NG151 FINAL (January 2020).*

there is very little if any support following testing in Ireland". Another lady noted: "The genetic counselling and services in themselves were excellent. However, once I had my BRCA status, I had to work very hard to get the surveillance I needed. Even though, the services were superb. I had to put in many hours to find my breast and plastic surgeons, longer to get decent advice on HRT - I had to go to London for that. People who are just into the system have very little chance of a good outcome surgery-wise. The lack of support afterwards - -there should be centralised services for people in this situation".

One participant who completed the survey noted: "I accessed tests privately and there was a minimal wait however, on receiving a positive result there was a gap in where to access the next step or a cohesive plan / model of care. I had to do the research and seek help myself with limited resources or where to go for information. I had BRCA without a cancer diagnosis, I fell through the cracks and was almost a fraud taking a place among women with a cancer diagnosis".

Another survey participant noted: "Was only tested for BRCA1&2. Came back negative but no other option for further genetic testing was offered or suggested. High hx [history] of various cancers in my family so would like to be able to join the dots and see what's going on." Another participant noted: "I've BRCA 2, I'm given a mammogram & breast MRI yearly since the age of 39 and I'm now 42 and on a waiting list for preventive surgery which I could be waiting years for". The perception that the panel test being too limited was articulated by both patients, the public and some health care professionals. The importance of looking at a broader panel which incorporates both high and moderate risk mutations was articulated. To support an earlier diagnosis, promotion of health and prevention of illness, some participants felt that expanded panels could be used and that the potential of a polygenic risk score which combines the effect of many risk variants needs to be exploited. Higher levels of cancer risk stratification for the general population could be achieved by incorporating polygenic risk score, mammographic density results, and other factors into risk models. The long-term investment in the cancer genetic services should be based upon maximising the early identification of all individuals at moderate and high risk of cancer.

Women with hereditary breast cancer, BRCA genes can consider the option of prophylactic surgery with women in this study citing the psychological benefits of reducing their risk profile. Others acknowledged the particular psychological support that women need if considering risk reducing surgeries and the benefits of having access to clinical physiological support. Group sessions were seen as a particularly scalable option in this context. One participant noted "once I heard I had BRCA2 the surgery couldn't happen quick enough—delighted with the surgery—heard later there is an increased risk of cancer associated with [name of] implant, oncologist and surgeon have reassured me not to worry--- also have a crinkle or dimpling of the implant- more of aesthetic problem than anything else". The need for rapid access to risk reducing surgery was highlighted by several patients, their families and nurse specialists. "Once the individual knows they have BRCA and if surgery is seen as the most evidence-based approach, then they just want it done ASAP, its' is a huge worry for them whilst they wait" [female nurse specialist].

One participant noted that such a clinic would help as then "you don't have to be the middle- man between lots of consultants who may be contradicting each other with their advice" and it would lead to a more holistic evidence-based approach.

***The need for dedicated syndrome or mutation specific pathway and clinics was highlighted. For example, for patients with BRCA positive mutation the patient could get to see the breast team, gynaecologist or urologist and other relevant physicians and offer the potential for a psychological support referral all in one visit.***

## A PREFERENCE FOR A HUB AND SPOKE MODEL AND AN INTEGRATED GENETICS PATHWAY

### A CENTRALISED DIAGNOSTIC SERVICE VERSUS AN INTEGRATED HUB-AND-SPOKE NETWORK DESIGN

The Irish cancer genetics services were described as being a centralised diagnostic service concentrated within a particular geographical region rather than being part of an integrated care pathway. "Disjointed" and "fragmented" were two words which were consistently used to describe genetic services. Disadvantages included: longer waiting times for initial appointments and for test results; transportation difficulties; and lack of follow-up. This can hamper efforts to deliver the best care possible to patients. One genetic counsellor who had exposure to the US, UK and Irish genetic services noted that the "Irish genetic services are two decades behind the UK".

A preference for a hub and spoke model and an integrated genetics pathway were articulated. The genetics programme needs to be planned based on a long-term vision and requires investment.

*"---- embedding genetics within the multi-disciplinary team and the care and the pathway, it shows the patient it [genetics] is integrated more within the patients routine care, it's the best model I've seen" [female with recent experience as a health care professional within genetic services in Ireland and another country]*

***The hub-and-spoke network design is a service-model consisting of an anchor facility (hub) which offers a full array of services, complemented by secondary establishments (spokes) which offer more limited services, and referring individuals needing more intensive services to the hub as needed. Such an organisational model would embed genetics and genetic counsellors within the dispersed oncology system with expertise being available to the local MDTs within the centres of excellence; genetics could be more integrated into routine care facilitating streamlined, automated and direct access to genetic services; facilitating consistency across services and allowing for more structured follow-up.***

## MAINSTREAMING AS AN APPROACH

"Mainstreaming" was mentioned as a solution used in other jurisdictions, particularly for breast and ovarian cancers, to speed up the access to cancer genetic testing. In the UK NHS mainstreaming means that any clinician can order the genetic testing for cancers in their own specialist areas. The aim of mainstreaming was to make cancer gene testing part of routine cancer patient care, by integrating testing into the cancer patient pathway; oncologists or surgeons can order cancer genetic testing directly at the point of diagnosis for certain cancer types for patients who fulfil certain criteria. A pilot had been run through the Royal Marsden for Irish patients (n=100) to access such direct testing in the past; the pilot at the time was taken up with enormous enthusiasm, a report of same is with the NCCP. A similar process was also offered through a clinical trial in the past. Outside of that the individuals interviewed were not aware of mainstreaming being more widely adopted in the Irish context.

Concerns about mainstreaming relate predominately to the lack of specialist genetics training for oncologists and physicians and the lack of genetics expertise within the dispersed oncology system. Thus, there is a need to upskill clinicians and to train more specialist genetic counsellors to facilitate this model. Reference was made to specialist postgraduate online training for physicians particularly specialist registrars in medical oncology being made available through The Royal College of Physicians of Ireland (RCPI)[6] and availability of online training and a Scientific Training Programme[7] to prepare for "mainstreaming" in the UK. Support is required for mainstream clinicians throughout interpretation, reporting and follow-on processes. When questioned about the cost of such a model, participants mentioned the cost neutral/potential for cost savings elements as using an existing appointment; means that the specialist genetic services are freed up to support those with a positive result and it can result in potential savings in terms of quality adjusted life years and savings in terms of treatment costs.

At the moment whilst mainstreaming is not formally available within the Irish services a "fragmented and disjointed version of mainstreaming has evolved [as a workaround], primarily out of frustration with lack of access, where physicians and surgeons refer patients to go privately or to self-fund for specific tests. This is problematic as firstly the test results are not supported by a clinical genetic counselling and testing service with associated family follow-up and cascade testing as needed; and secondly if testing is negative and there is a strong family history an individual may still need to come and talk to a counsellor to discuss their family history; thirdly an uncertain result- needs to be discussed regarding next steps--- don't have that backup if working outside of a mainstream clinical genetics service and fourthly may miss something labelled as 'not genetic' or worse a variant may be misattributed as being pathogenic and managed inappropriately" which can lead to litigation down the line [a consultant with expertise in cancer genetics].

*"Genetics services should be seen more as a therapeutic process rather than just a diagnostic or screening process and incorporated into the treatment pathway" [female with breast cancer]*

*"I'd like a one stop shop--you go in and you get everything done, take your females, even have your colonoscopy, you can have your testosterone screen, you have, you know, attend your gynaecologist have whatever tests you need there, dermatologist is another one, I forgot about that- maybe even some psychological help as well, they are all under the one umbrella. So you are not going here and there and yonder and I would assume that you have a coordinator for all of that--- it's not the individual themselves with the diagnosis seeking services, the services are made available to the individual-- for the people that are not proactive--- with a bit of luck they will be okay, but it's luck they'll need" [male with Lynch syndrome].*

***At the moment whilst mainstreaming is not formally available within the Irish services a "fragmented and disjointed version of mainstreaming has evolved [as a workaround], primarily out of frustration with lack of access.***

***One consultant noted that whilst there is "a lot of negatively around the cancer genetic services, the clinicians working in the service are fantastic with a world class service, lots of positive things coming in terms of increased genetics workforce, education and implementation of the cancer strategy."***

[6] *Details available at: https://courses.rcpi.ie/product?catalog=Certificate-in-Cancer-Genetics*

[7] *Details available at: https://nshcs.hee.nhs.uk/programmes/stp/*

## KNOWLEDGE AND AWARENESS ABOUT CANCER GENETICS AND CANCER GENETIC SERVICES

Whilst genetic specialists and counsellors have familiarly with the guidelines for cancer genetic testing and the profiles of individuals that need testing, such information needs to be more broadly communicated and discussed within the different cancer centres and within GP practices. Oncology as a specialist discipline requires healthcare providers to be proficient in a myriad of areas, including genetics which can be challenging given the rate of evolution of knowledge in this area.

A key obstacle to the increasing use of genetic testing in healthcare was considered to be a lack of information and sufficient competencies among healthcare professionals. The need for the genetics expertise to be integrated at the point of oncology clinical care was noted. Genetic counsellors or highly skilled specialist nurses dispersed within the oncology network may offer part of the solution.

The idea that individuals will be aware of their genetic profile in terms of health risks through commercial means was also mooted. Thus, many participants noted the need to raise awareness and knowledge, amongst GPs, oncologists, surgeons, nurses particularly nurse specialists, about genetics and genetic testing and the need for a broader conversation from a patient and public perspective. Whilst there was an acknowledgment that basic genetic science and its application in the health care context is included in the curricula of undergraduate health care programmes, there was a perception that the genetics content of undergraduate curricula could be enhanced. Most of the healthcare professionals interviewed particularly oncologist and nurse specialists noted that their cancer genetics specialist knowledge was gained by "osmosis" and their search for information given their interest in the field rather than by a particular strategic drive to increase their knowledge and awareness. Some of the medical oncologists particularly referenced their experience in other jurisdictions and colleagues with specialist knowledge in the area as being particularly helpful. One medical oncologist noted that "My colleagues would not feel confident in educating patients about a BRCA mutation and what that might mean for them and their families". Individuals with cancer generally commended the knowledge of their oncologists and clinical nurse specialists and highlighted their role in supporting their access to follow-up treatments and surveillance.

A genetics counsellor noted: "requires the clinician's knowledge and experience with rare cancer types and sometimes they don't get that education necessarily and you know if they have a special interest and they will talk about them [genetics] well and good but broadly they often don't know what to look out for and so that's a kind of a barrier for patients to then access genetics because they may not be getting that or having that conversation with their doctor". There was a perception among many participants and some participants gave concrete examples of where they were refused access to genetic testing with some of these testing positive later.

A female with breast cancer and BRCA2 mutation noted that "There is a discrepancy in knowledge [of genetics] between different members of the team". Whilst a male with Lynch syndrome highlighted that there is a "lack of awareness of people, of doctors, of the syndrome, Lynch Syndrome and the follow up requirements associated with it --it is a barrier". One participant noted that a GP said that because the "BRCA mutation was on the father's side of her family, that she was not to worry" there was no need for her to get tested. However a woman who has a strong family history of breast or ovarian cancer on her father's side (e.g. her dad's mother or sisters) has the equivalent risk of having an abnormal breast cancer gene as a woman with a strong family history on her mother's side.

Some participants felt they should have had access to cancer genetic services or at the minimum better understand why they were not offered such services. Some suggested there was a need for a "National information campaign about what genetic information is and what is genetic testing what happens to that information why it is important for you to get that information and that becomes a thing just like a normal conversation". One participant noted "a good GP makes all the difference", noting that whilst her GP initially did not have the knowledge, he sought it out.

One participant with BRCA2 cited the power of having celebrates highlight particular mutations and the risks associated with them. She said: "Angelina Jolie yeah she brought it [BRCA] into the common language in a way that this was something that we should be looking out for".

Mechanisms of increasing awareness and knowledge of genetics were suggested as: educational updates and webinars for health care professionals and the public; having a repository of up to date evidence-based information for both the public and healthcare professionals that is easily accessible. Many of these guidelines are updated very regularly, thus such a repository would help healthcare professionals to access the most up to date information. A lack of follow-up information either online or written was highlighted. Such information would help individuals as they adjust to their diagnosis and their new normal. The need for more dedicated private spaces for the nurse specialists, oncologists, or genetic specialists to have discussions with patients was also declared.

*"My mum's brother had died of bowel cancer-- and her sisters are in America and funny they had said that the Consultants in America said that if there is anyone that had bowel cancer in your family you should really go and push scopes-- My mother had stomach cancer-- my dad then last year was diagnosed with colorectal cancer-- I've been going to a Doctor, I started getting heart burn-- the Consultant recommended because of my family history that I go for scopes every two years just to keep an eye-- no-one looked at this objectively in terms of my family history and see -- do I meet the criteria or not, they [doctors] didn't kind of do that kind of an assessment--- So I did actually ask [for genetic testing] and they said that well you know if you do your scopes and that you know you are more preventative--- they may have been treating me more from an emotional worry point of view --it was more the emotional support in relation to you know am I worrying unnecessarily-- I would like to see that there is a genetic test for two reasons one for myself as a person but also then it gives you a kick in the ass in a way in relation to okay maybe that it not genetic but maybe your lifestyle is not right and there is my family to think of" [female member of the public].*

*"I am 27 and he [ doctor] said it to me because of my age and the rarity of this cancer as well, it was all very unusual and he asked if I had interest in genetic testing and had my Oncologist mentioned it— [when I suggested it to] my Oncologist more or less dismissed it--- I still would have liked to have information about genetic services and what it means in relation to my particular cancer, my level of risk and my family members, I think I went off, I Googled that and then you don't know if you are at risk or if it is reliable information" [female in her twenties with appendix cancer].*

***"GP's wouldn't necessarily have the understanding that would be required, it would be more the oncologist. My oncologist and gynaecologist they were brilliant and knew all about the risks. I went to see the doctor, the professor to talk about [my risk of] pancreatic cancer and he sat there on his computer Googling about pancreatic cancer he looked up some I don't know medical journal and he was reading out from it and I was going Jesus and he didn't even know much about it, do you know the way. My radiation oncologist, I had to sit there and tell him all about it but he was genuinely interested because you know they don't seem to be very informed about it as the risks, he didn't know anything about the elevated risks to melanoma or pancreatic cancer, he was writing it all down-- I have definitely met a few that haven't a clue" [female with breast cancer, BRCA2 mutation].***

## FUNDING AND WORKFORCE

As our knowledge of genetic mutations and the genetic basis of certain cancers increases so too does the demand for the services. The need for more people working within the genetic services was highlighted as was the need for increased funding. The need for a targeted strategy to develop the expertise in the area of cancer genetics was noted and the need to invest in the training of genetic counsellors was highlighted. "We have a public wait list for family members of up on two years; specialist genetic staffing should be doubled; based upon European recommendations we should have 30-40 genetic counsellors across the country- we have 15; we need more administrative support staff to do the family histories and admin work, and we need many more genetic consultants and we need training programmes, an MSc for genetic counsellors" [genetic counsellor]. Investment in the provision of genetics education to masters' level would ensure a pipeline for the genetics workforce.

Disparities in access to genetic services, and consequent reconstructive prophylactic surgeries were highlighted. Access varied according to geographic location, tumour type or diagnosis and whether a person has access to health insurance. Waiting times for counselling and receipt of results were long particularly for family members. There is an over reliance on the knowledge and understanding of individual consultants and physicians, which isn't always equal across the country and across tumour types, thus there is a need for more automated processes e.g. a particular pathological and molecular diagnosis leads to a particular genetic test, with particular treatment pathway(s) clearly defined.

In addition, there is a need for enhanced access to cancer drugs and treatments nationally. One oncologist noted "from the Lynch syndrome point of view, the endometrial cancer with Lynch syndrome should get access to immunotherapy but it's actually not available in Ireland so even though it's the appropriate treatment to give them I cannot access it. So there are compassionate access programs whereby I will ask [name] to have an immunotherapy agent I will say 'I have an endometrial Lynch syndrome I would like access to an immunotherapy' and sometimes you are lucky sometimes you are not".

Internationally cancer genetic services facilitate access to tumour-based sequencing, germline sequencing and whole genome sequencing on a diagnostic basis for specific cancers. The Irish system was seen as being quite similar to the UK-NHS in terms of germline sequencing and follows much of the NHS directory. Deficits in access to onsite or national tumour-based sequencing which inform the need for targeted therapies and personalised cancer care was seen as an area that needed to be developed. The importance of The Genomics Tumour Advisory Boards was reiterated by medical oncologists. One consultant with specialist expertise in cancer genetics noted due to rapid technical advances, reduced sequencing costs, and growing number of targeted therapies, it is anticipated that the use of extensive tumour sequencing is becoming the standard of care. This means that clinicians will be challenged with increasingly complex genetic information and multiple test-platforms to choose from. Therefore weekly "structured MDTs- Molecular Tumour Boards" and "global Molecular Tumour Boards" to deal with complex cases have been suggested as solutions. Molecular Tumour Boards bring together the requisite skills to interpret and translate the genetic findings into the clinical context and optimum treatment protocols. This would involve making the expertise of genetic counsellors available to MDTs within the centres of excellence either with counsellors being located centrally (in the 'Hub') and attending targeted clinics and engaging virtually to the 'spoke' centres of excellence or having the genetic counsellors located in the nine spokes and linking in with the hub. Many clinicians had a preference for the later. One consultant noted in the Irish context there is a "heavy leaning on pharma input" in the extensive tumour sequencing space, where it might be better to have this funded consistently and nationally through the NCCP.

*"No good waiting for people to get sick and then they [health services] throw money at it, wait for somebody to get cancer and to give us chemo -- why not discover the potential that cancer is going to happen before it actually happens—polyps, they can tackle that with a colonoscopy before it becomes a problem or they can diagnose people at an earlier stage -- You have to invest to actually yield the benefit from a prevention and a targeted treatment perspective ---- funding is the big one you know, more geneticists and counsellors -I'd like to see you know a lot more education maybe in the GP sphere, more research, a genetics register so that there is a central point where people can access information ---- At the moment it's fragmented, at the moment nobody can tell you how many people in Ireland have been identified with Lynch syndrome" [male with Lynch syndrome].*

***They test all the [women] irrespective of age or family history, every endometrial cancer gets tested [for mismatch repair deficits in this hospital] and this is not happening in every hospital --- not uncommon it's about twenty, twenty five per cent of endometrial cancers, a proportion of them maybe about five to ten per cent may have a syndrome called lynch syndrome--- so once they have a mismatch repair defect I will send a referral to Dublin-- there is a big difference with access to pathological tests across the country-- equally there are other tests that are done in other places that we mightn't do here -- so they [consultants in other hospitals] don't know that these patients may have a lynch syndrome and I think that has colossal implications because if you are not picking up lynch endometrial; you are exposing a whole family to potential cancer risk in the future" [consultant oncologist].***

## ACCESS AND INEQUALITIES IN ACCESS TO CANCER GENETIC SERVICES

Participants articulated the need for more widespread access to routine genetic testing on the premise that having such information helps with cancer prevention and early detection thus saving money. Examples cited included that all ovarian cancer and breast cancer patients will get BRCA testing and all colorectal cancer patients will all get access to dual panel tests at some point in the future that is genetic testing is part of their routine care rather than being an exceptional requirement.

The current genetics system is designed based upon the articulate, educated person usually women who can proactively manage their own health, for individuals who were not of that mind-set or who lack such skills, there was a common understanding articulated that such individuals "would fall through the cracks alright". Individuals with private health insurance were able to bypass the public system but some perceived that they had no way to access the services needed after their genetic results were received.

One health care professional noted: "The Irish travelling community- needs face to face, building a relationship--tailor information in a way that suits their culture, their communication their way of living --- It's a different way of communicating".

It was noted that the length of time since the diagnosis of the index case was a factor; that is the longer the timeframe the less likely family members will come forward for testing.

Financial barriers were highlighted as travel costs and costs of consultations and tests if going privately. One participant said, "So somebody on the dole [social welfare payment] or whatever is going to have an issue going private so they are totally dependent on the public system you know, which is slow". Another participant noted: "I'm glad I got the testing done but having to go private cost a lot of money".

***Particular groups who may be challenged in accessing cancer genetics services were highlighted as: men, individuals with rarer cancer susceptibility mutations or syndromes; those from minority ethnic or cultural groups; those whose first language was not english; those with low health literacy and individuals with little familiarity with the irish health system and how it works.***


## MANAGEMENT OF SAMPLES AND DATA

Concerns re management of samples and data emerged as a concern for some participants. A small number mentioned that they feared that results could be used against them in terms of perceived health threats when seeking employment or insurance cover. Participants did not reference legislation covering this[8]. In the context of health insurance, concern re misuse of genetic data was cited where an individual could potentially be pushed out of insurance pools as a result of their declarations within the pre-quote screening surveys either as a result of being charged inordinately high premiums or being denied insurance cover.

## CONCERNS RE MANAGEMENT OF DATA AND SAMPLES

One medical oncologist cautioned around GDPR and that "data needed to be protected as genetic information is exceptionally vulnerable". One survey participant who used the public services in 2013 asked about her data: "The real problem now is with my data. Where is it?" she queried how was her data protected and how could she access it. Another male with lung cancer noted "all patients with a similar diagnosis should have the same testing with the maximum number of relevant genes tested and a standard of feedback structure so that the person knows what was tested for and what was found --- it may not have significance at this time but this may have significance for the individual or their family in the future". This is important for example in the case of "uncertain test results". The participant also noted "there is a need for rules and regulations and governance regarding what is done with genetic samples in both public and private settings and the co-existing clinical and genetic data--- there is a need for broader discussion involving the Department of Health, health care professionals, patients, data managers and key stakeholders to look at how the data can be safely managed and used to support patient care".

One genetic counsellor highlighted the importance of having a "fully functioning laboratory co-located with a genetics hub". Issues were raised with the use of multiple laboratories

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[8] *The Irish Disability Act 2005 (Section 42.2) prevents the use of genetic testing results for employment purposes, (health) insurance, life assurance, and pension or mortgage applications.*

*Insurance companies have a voluntary code of practice in this regard as well.*

for genetic testing. The disadvantages of using multiple laboratories were cited as: more costly when outsourced; difficult to access the control sample and associated details when sample are in multiple locations (e.g. US, UK, Germany), increased turnaround time. It is better if the laboratory can have direct access to the clinician and it is better to have samples from Irish patients located in Ireland. The need to centralise the data in one repository was highlighted as this allows one to track and connect variants and offers the potential to link to other data sets (e.g. National Cancer Registry)

Two participants highlighted the questionable value of direct- to consumer genetic testing highlighting that their use needs to be carefully monitored.

## A VISION FOR A STATEWIDE DATABASE

A vision for a state wide database that logs all of the individuals that come to see genetic services, linking up families to the pedigrees and integrated with the main hospital system to link clinical data was articulated by some. Such a database could combine high-quality information technology with genome and clinical data for both clinical and research utility giving a population perspective to help with planning and design of services.

Current processes for linking families and pedigree information (e.g. between private and public genetic services) are labour intensive and involve requests for information via confidential emails requesting such information and the retrieval of the information from a hybrid paper/online system. A consent to share such information with family members would have been attained as part of the original genetic counselling processes with the index case. Whilst staff are very helpful, email requests are difficult to track, take time to process and requests can be misplaced.

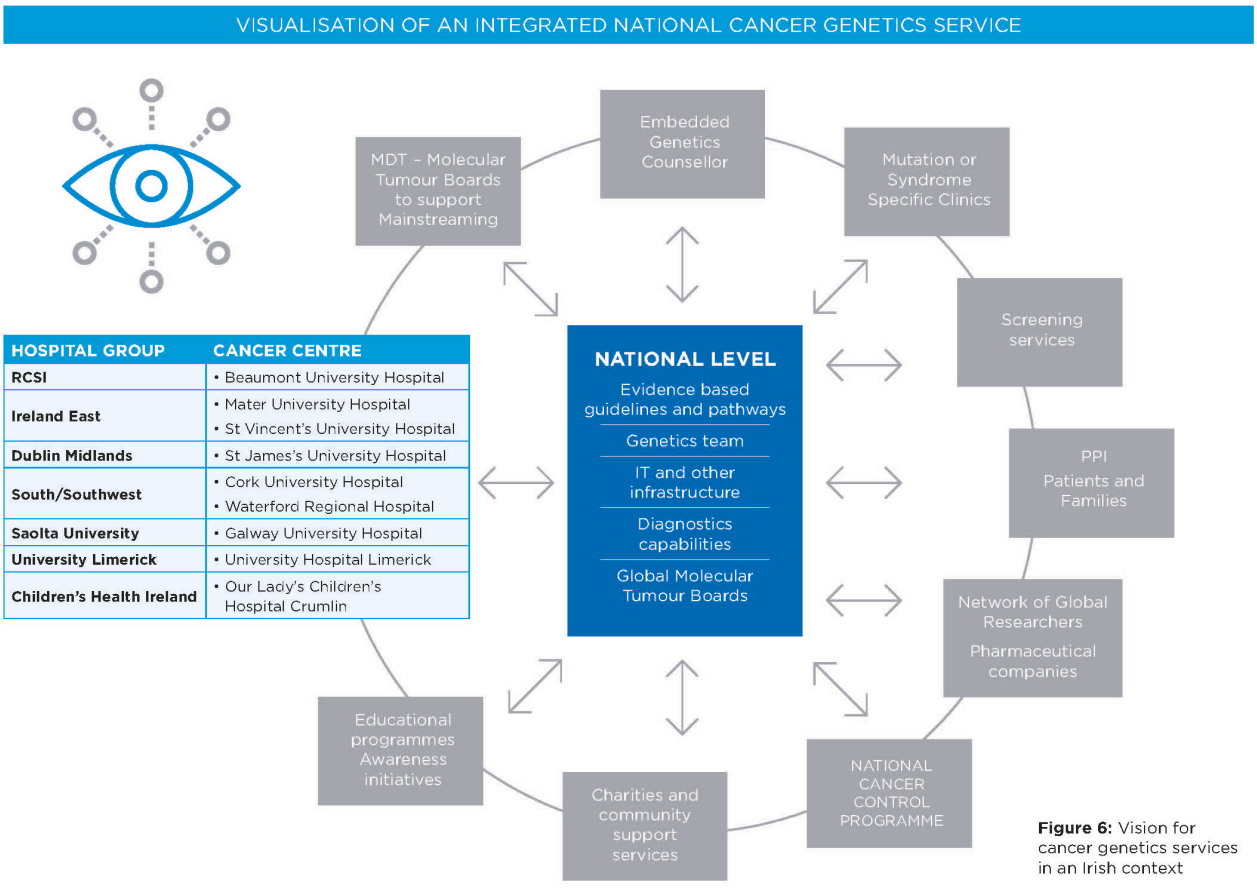


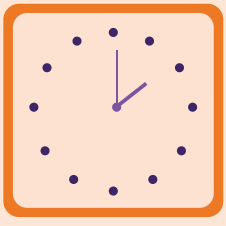

**Figure 6:** Vision for cancer genetics services in an Irish context

**Figure 6: Vision for cancer genetics services in an Irish context**

# DISCUSSION

The National Cancer Strategy 2017-2026 highlighted the growing role of cancer genetics in cancer prevention and cancer care. The HSE's National Cancer Control Programme is working closely with clinical genetics and cancer genetics services in Our Lady's Children's Hospital, Crumlin, and St James's Hospital to develop the services to meet the growing need. Under the strategy, investments are being made in additional staff and to enhance testing capability. The National Cancer Strategy (pg. 75) notes that "At present cancer genetics services in Ireland are underdeveloped and underfunded---- Approximately 1,800 new patients avail of the cancer genetics service each year". The Strategy goes on to note that "results with therapeutic relevance for patients undergoing treatment are delayed and healthy individuals are not being informed of their inherited cancer risk in a timely way" ..... "It is likely that many new patients diagnosed with breast, colorectal, ovarian and endometrial cancers will soon benefit from genetic testing. Other patients, such as those with prostate, thyroid, gastrointestinal stromal tumours and phaeochromocytomas will also require testing in the future. Clinical cancer genetics in Ireland requires a strategic approach that will include increased infrastructural and financial support." The Strategy makes reference to National Cancer Genetics Service and an integrated cancer genetics service, additional specialist genetics workforce, national management protocols, nurse/counsellor led clinics, a coordinated national recording of genetic test results, telemedicine, and biobanking. Much is happening with moving this agenda along, however this study highlights that much remains to be done.

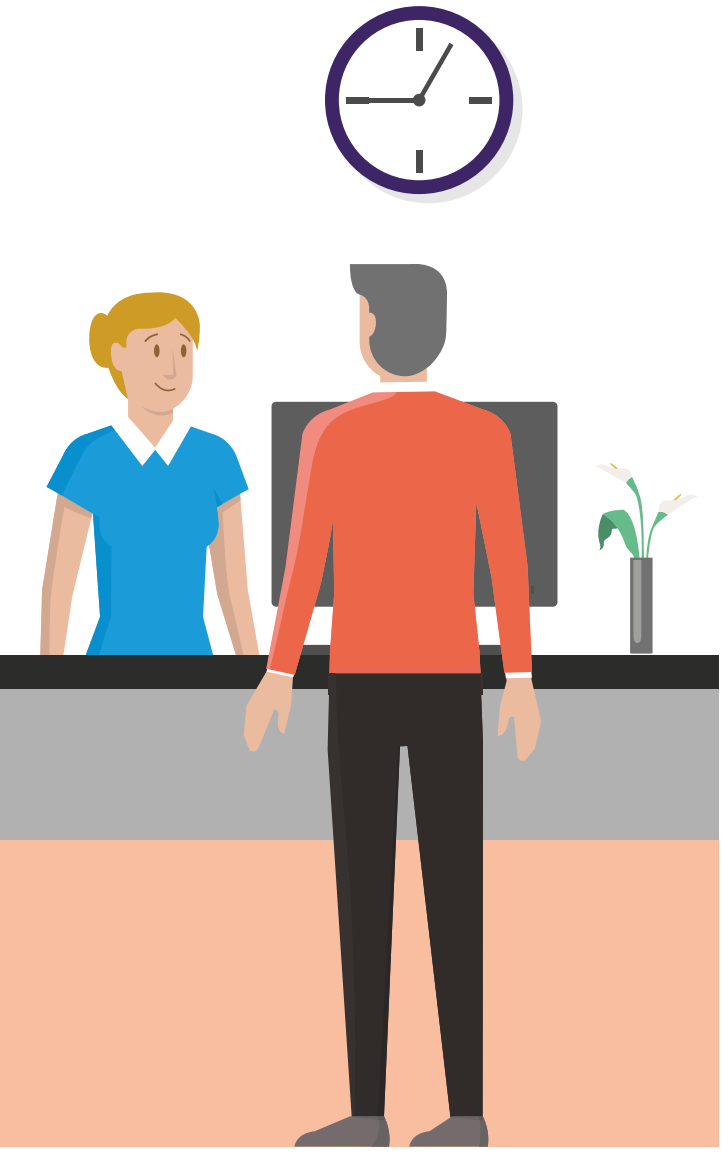
Genetic testing can have an impact on the psychosocial well-being of the patient and their family; participants in this study cited the increased anxiety associated with waiting for appointments and test results. Given the limitations on genetics workforce, infrastructure and resources, the genetic testing services are organised so that they can review patients most at risk of having a hereditary predisposition to cancer, which is mostly based upon a strong family history (McVeigh et al., 2014). A family history questionnaire must be completed by each patient referred for testing to allow for the selection of individuals for genetic testing. Unaffected first-degree family members of an individual with proven mutations may then be offered pre-symptomatic predictive testing. In the Irish context UK guidelines such as NICE are regularly referenced. However, controversy exists over the criteria used for referral for genetic testing. It is perceived that the eligibility criteria used to decide who should qualify for genetic testing in the public system are overly restrictive, relative to other European countries. In contrast with many other European countries, Ireland does not make genetic testing available to all asymptomatic women at high risk of BRCA related breast cancer (The Health Policy Partnership, 2019). McVeigh et al., (2020) note that "in light of the limited resources available in the public health system, it may be most beneficial to the greatest number of people to loosen criteria for testing of the highest risk actionable genetic variants that will modify treatment and/or risk management,



***According to the Clinical Genetics Medical Workforce in Ireland (2019) report "The national genetics service is staffed by consultants, trainee NCHDs, non-trainee NCHDs and Genetic Counsellors. …Inadequate staffing levels and long waiting times are perceived as precluding referrals to the service… Currently the priority waiting list is between 15-18months and routine referrals wait > 2 years to be seen… A Clinical risk assessment associated with the waiting list is currently being performed and a number of serious adverse outcomes have been noted … A fully functioning national service cannot be accommodated. This has led to poor practice in terms of testing requests and also poor-quality foreign laboratories handling Irish samples".***

rather than offering larger panel testing to restricted number group of patients" (pg. 860). Rosenthal et al., (2017) noted that expanding genetic testing beyond BRCA1/2 significantly increases the number of women who are candidates for enhanced screening and other risk reduction measures, most of whom would not have been identified through family history assessment.

Preferences for an integrated hub-and-spoke network design which embeds genetics and genetic counsellors within the dispersed oncology system and linked to the national screening services (such as BreastCheck, CervicalCheck and BowelScreen) was expressed. Such an approach would extend the expertise and resources of National Centre for Genetics to the wider oncology community. Cancer genetics comprises approximately 30% of the public health genetics current workload. The main genetics specific database, known as iGene is based at CHI at Crumlin. Ireland's actual ratio of consultant Clinical Geneticists per 100,000 has been calculated as a consultant headcount of 4 which is less than the 15 recommended by the HSE in a Review of the Clinical Genetics Medical Workforce in Ireland (2019); notably this report deals with genetics more broadly.



Many participants bemoaned the lack of national data on the numbers of individuals with specific mutations in an Irish context, this hampers the ability of the services to visualise the scale of the issue and to plan and deliver an effective service.

Patients whilst they seemed to be familiar with details of the mutation that they have tested positive for, only two patients mentioned results of the tumour based sequencing with one highlighting that they would have liked to hear more about this with a written record of what tests were performed and what was found. Mullally et al., (2020) in a survey of 84 patients in an Irish hospital identified that 42% of respondents were familiar with genetics and 90% stated they would pursue cancer genetic testing, if available.

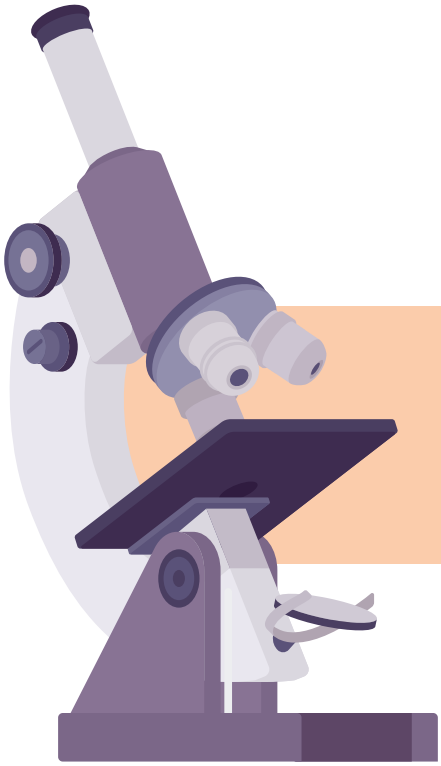
Within our study we found an acknowledgement and enthusiasm for the potential of genetics/genomics to improve health through the prevention and treatment of cancer and the potential to assist family members through lifestyle changes and additional surveillance. Genetic information is complicated, and the growing availability of increasingly complex testing options challenges clinicians' ability to communicate findings to patient and families and to translate the genomic findings into routine clinical practice. A positive result has repercussions in terms of further management, in some instances involving the consideration of prophylactic surgery, and screening for other cancers, as well as need for predictive testing of pre-symptomatic first-degree relatives (cascade testing). Long-term follow-up, support and coordination is essential for those with positive test results because of the need for information on risk-reducing strategies, surveillance, and the need to translate new and emerging approaches into the routine care and follow-up of these individuals. This is particularly cited in the literature pertaining to BRCA mutations and Lynch Syndrome (Hunter et al., 2017; Peterson et al., 2018; Seven et al., 2020). The public health benefits of cancer genetics have yet to be fully realised as we continue to have inequitable access to genetic testing and associated personalised medicine and surveillance (Allen et al., 2020).The benefits of risk management multidisciplinary clinics designed to quantify cancer risk, offer advice on preventative strategies and coordinate access to surveillance and targeted intervention strategies were highlighted. Risk management of BRCA mutation carriers for example include access to risk-reducing surgery (risk-reducing mastectomy and risk-reducing salpingo-oophorectomy), chemoprevention with hormonal therapy, and surveillance with mammogram and breast MRI. Guan et al., (2021) in a systematic review regarding initiatives to scale up and expand the reach of cancer genomic services outside of specialty clinical settings noted that such efforts are limited outside of traditional oncology and genetic clinics. These authors noted this is a missed public health preventative opportunity because evidence thus far suggests that these efforts can be successful in expanding the reach of genetic services with the potential to reduce health inequities in access.

Despite routine recommendation to individuals undergoing genetic testing about communicating risk to family members, it is estimated that between 20–40% of at-risk family members remain unaware of relevant genetic information or chose not to engage in genetic testing (Hodgson et al., 2014). While family communication between first-degree relatives happens frequently, communication with second and third-degree relatives is becoming increasingly challenging as

family structures alter and families become more dispersed (Petersen et al., 2018). One glaring gap in the evidence is the perspective of family members who do not go for testing and who chose not to engage with their genetic history; understanding their perspectives would be important as we move forward with developing educational materials for the general public.

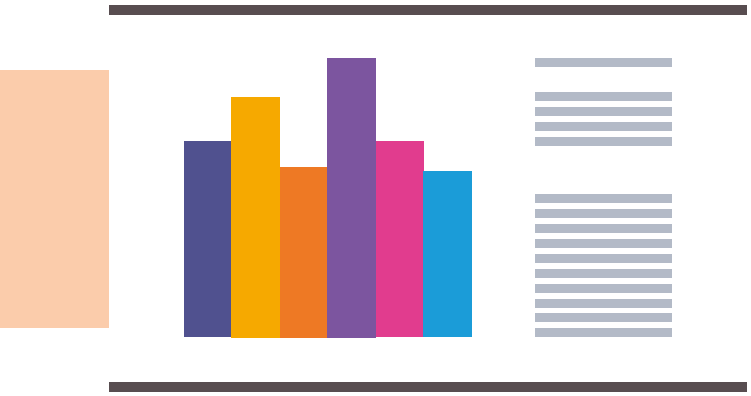
Index patients (with a known cancer susceptibility mutation) play an essential role in the communication of information regarding a mutation and the importance of the associated preventive measures. They generally feel a moral and ethical obligation to communicate the information and they generally seek to encourage relatives to get tested (Seven et al., 2020). Similar to other research in this area (Aktan et al., 2011; Petersen et al., 2018), participants in this study cited that this is a challenging process for which they feel ill prepared. Findings in US studies, suggests that closeness among family members, concern for family and future generations, and awareness of cancer risk act as facilitators for information sharing and encouragement for genetic testing whereas cancer risk as a challenging topic to convey and disrupted family structures and family relationships can act as barriers (Chopra and Kelly, 2017; Petersen et al., 2018). Dattilo et al., (2021) in a systematic review noted that parental disclosure of test results may be challenging due to concerns regarding age of the child/young adult, developmental appropriateness of the disclosure, and the potential for emotional burden. Participants in this study articulated feelings of guilt for passing on the mutation to offspring, similarly Frost et al., (2019) in a mixed methods international study interviewed 32 women diagnosed with breast cancer who also identified feelings of guilt on passing on a mutation to a child.

Hereditary breast and ovarian cancer (HBOC) syndrome (BRCA1 and BRCA2 genes) and Lynch syndrome (MLH1, MSH2, MSH6, and PMS2 genes) are recognized as having cumulative evidence supporting the benefits of early interventions where genetic profile and family history models support such interventions (Hunter et al., 2017; Buchanan et al., 2020). Alternative strategies for more rapid and increased access to cancer genetic testing were articulated in this study. These can vary from population level screening to mainstreaming. Genomic screening of the adult population can identify previously unrecognized individuals at increased risk of cancer and other diseases and facilitate risk management and early cancer detection. Zhang et al., (2019) evaluated the impact of offering preventive population genomic screening to adults aged 18–25 years in Australia; the researchers found that population screening would reduce variant-attributable cancers by 28.8% and cancer deaths by 31.2%. Population level screening whist viewed as a health prevention intervention can be associated with concerns regarding the feasibility of and ethics of population genomic screening and the potential for genetic discrimination, marginalization, or stigmatization. In the UK, the Mainstreaming Cancer Genetics Programme has been developing simplified eligibility criteria and testing access processes to improve access to BRCA testing. NICE recommends offering testing for Lynch syndrome to people who are diagnosed with endometrial cancer and recommends testing all patients with colorectal cancer, when first diagnosed, using immunohistochemistry for mismatch repair proteins or microsatellite instability testing to identify tumours with deficient DNA mismatch repair, and to guide further



sequential testing for Lynch syndrome (NICE Guidelines, 2017 and 2020). Kemp et al., (2019) evaluated mainstream genetic testing within the NHS, using simple cancer-based criteria in patients with cancer (n=1184) and found that mainstream testing offers promise in terms of efficiently delivering consistent, cost-effective and patient-centred BRCA testing. Concerns regarding mainstreaming include: fears regarding a lack of consistency in services and patient management including the interpretation of genetic variants, the educational requirements of non-genetic specialists who may be required to offer testing, a lack of resources within clinical genetics to support mainstream services, lack of pre-existing mainstreaming guidelines or protocols, concerns regarding workload and the perceived relevance of genetic testing for patient care in the short-term (pg. 294). The proliferation of genetic variants might cause clinicians and others to misread their clinical relevance, potentially leading to overestimation of risk, overdiagnosis, and overtreatment (Scott et al., 2020). Scott and colleagues cautioned that "moving from genetic risk profiling of rare monogenic disorders within families to wider polygenic profiling for more common diseases in large asymptomatic populations carries considerable potential for harm and waste. When choosing predictive genetic tests, clinicians and consumers must avoid commercial hype, ask relevant questions, and advocate for rigorous evaluation". The authors highlighted that testing for BRCA1 and BRCA2 mutations was an example of a near ideal test given the potential for better outcomes.

Follow-up of index patients (with cancer and a mutation) is usually coordinated through the treating oncologist, many participants cited the benefits of having specific pathway that allowed patients and family members with specific mutations to access referrals, screening, surveillance and access to evidence-based treatments. Suggestions included the central hub genetics centre taking a coordination role and having mutation or syndrome specific regional clinics. Asymptomatic BRCA mutation carriers face challenging decisions regarding cancer prevention, screening and early detection, risk-reduction surgical and pharmacological



options, and menopausal hormonal management which require a multidisciplinary and individualized approach (Stan et al., 2013). It is estimated that up to 20% of ovarian cancers have an inherited genetic basis with the most common being BRCA1/2 mutations; risk-reducing bilateral salpingo-oophorectomy is often performed. Hickey et al (2021) in a systematic review of the psychosexual effects of risk-reducing bilateral salpingo-oophorectomy in female BRCA1/2 mutation carriers noted that women do not feel adequately prepared for the psychological and sexual side effects for instance removal of the ovaries results in surgical menopause with immediate effects. Thus, healthy individuals harbouring a BRCA mutation constitute a population with unique unmet needs, often overlooked by health services. Yershalmi et al., (2016) describe the potential benefits of a dedicated follow-up clinic for BRCA mutation carriers as impacting health, quality of life and survival of BRCA carriers. An open letter published in the BMJ highlighted that urgent improvements were needed to diagnose and manage Lynch syndrome within the UK NHS, thus highlighting the potential benefits of syndrome specific pathways and clinics (Monahan et al., 2017).

Mutations in breast cancer susceptibility genes (BRCA1 and BRCA2) put women at a higher risk of developing breast and/or ovarian cancer. For individuals with BRCA1, this equates to an estimated probability of developing breast cancer over a lifetime of 57–65% and that of ovarian cancer 39–40%; for individuals with BRCA2 mutations, the probabilities are at 45–49% for breast cancer and 11–18% for ovarian cancer (Antaniou 2003; Chen et al., 2006; Mylavarapu et al., 2018). Women with these germline mutations are more prone to develop these cancers at a younger age with more aggressive disease and poorer prognosis as compared to those with somatic mutations. It is estimated that approximately 20% to 30% of patients with cancer at high risk for BRCA1/2 mutations undergo genetic testing; these figures represent lost opportunities in terms of cancer prevention and early diagnosis. Genetic testing varies across ethnic groups, socioeconomic classes, and geographic regions with consequent varying access to health services. Forbes et al., (2020) in a systematic review, highlighted that regional and organizational guidelines differ for genetic screening, counselling, and treatment of patients with BRCA-mutated BC; authors noted that guideline harmonization would optimize identification and management of these patients. Samimi et al., (2017) highlighted that a "traceback approach" of retrospective identification of mutation carriers potentially provides an opportunity to offer informative genetic counselling, testing, and cancer risk assessments to probands and their family members who would previously have missed the offer of such a service.

A review of inherited cancer susceptibility syndromes highlighted the importance of clinicians being aware of the more common cancer syndromes, including hereditary breast and ovarian cancer, Li-Fraumeni, Lynch, familial adenomatous polyposis, retinoblastoma, multiple endocrine neoplasia, and von Hippel-Lindau (Brown et al, 2020). Review authors note that physicians need to be proactive in linking specific signs or inheritance patterns to a potential inherited cancer susceptibility syndrome and referring patients to an appropriate specialist, and initiating preventive care; this can reduce morbidity and mortality for both patients and their extended families.

The traditional model of cancer genetics has relied on individuals with a strong family history of cancer being referred to Clinical Genetics Departments, where family history information is assessed and confirmed with resources have been targeted at those with the greatest chance of having an inherited cause for cancer. Small family sizes, lack of contact with relatives and reduced penetrance mean that family histories cannot always reliably identify those at high risk. Practices in cancer genetics are evolving to address these challenges. Reduced costs of genetic testing, advances in tumour tissue analysis, next-generation sequencing technologies, simultaneous analysis of multiple genes, use of liquid biopsy (blood sample) and mainstreaming of cancer genetics mean that cancer genetics is becoming an essential component of routine clinical care in many cancer types (Harrison, 2019; Kentwell et al., 2017). Having genetics expertise available as close as possible to the patient on the cancer treatment pathway is important in order to prevent the wrong genetic test being ordered, genetic test results being misinterpreted, and inadequate genetic counselling (Brierley et al., 2010).

In this study, concerns emerged around loss of privacy and confidentiality, the use of multiple laboratories outside of the Irish jurisdiction and the longer-term management of genetic data and samples. The potential for conflicting variant interpretations between clinical laboratories exists associated with varying use of and interpretation of guidelines (Balmaña et al., 2016). Winkler & Knoppers (2020) discuss the ethical and legal aspects of precision cancer medicine including: the return of incidental findings and sequencing raw data to patients, the communication of genetic results to patients' relatives, privacy and communication risks with concomitant oversight strategies, patient participation and consent models. The authors note that many of these concerns can be addressed through addressing the "genomic literacy" of healthcare professionals working in the oncology field so that the full benefits of precision medicine can be realised.

System-level approaches to enhance family communication and provide support (e.g. specialty clinics, family counselling, support organizations, speciality websites and print materials) are needed. Cost-effectiveness data that examine the impact of such approaches on family communication and cascade testing may provide support for such approaches.

# STRENGTHS AND LIMITATIONS

This study was conducted over the period of the Covid-19 pandemic which meant all data collection processes were conducted remotely. The sample was a volunteer, motivated sample and this may introduce bias.

A mixed methods approach was used supported by a review of literature and a search for relevant Irish context specific literature which helped to triangulate, and sense check the findings.

# CONCLUSION

Mutations of genes are known to cause an increased risk of cancer, and these underlie approximately 3-10% of cancer cases overall. There is strong evidence that identification of cancer predisposition gene mutations has an impact on diagnosis and management of cancer patients and on prevention and early diagnosis of cases in their families. Emerging use of genetic testing to guide cancer therapies, combined with greater public and health care provider awareness, in the era of personalised medicine, has led to rising demand for publicly funded cancer genetic services. Genetic test results guide surgical decisions and direct choice of medication (pharmacogenetics) i.e. precision medicine.

Yet evidence of effectiveness and cost-effectiveness of genetic service models for the successful translation of genetic knowledge and targeted interventions leading to improvements for patient benefit are sparse. It is likely that the integration of cancer genetic testing into routine patient pathways, within a hub and spoke model, will prove to be the optimal pathway for most cancer patients. However, cost-effectiveness analysis is required to better understand the benefits that would be gained both from implementing new sequencing technologies and from broadening of testing access (Slade et al., 2016). In addition, targeted clinics and clinical pathways for particular mutations and syndromes would assist individuals in accessing targeted support and access to evidence-based treatment and support.

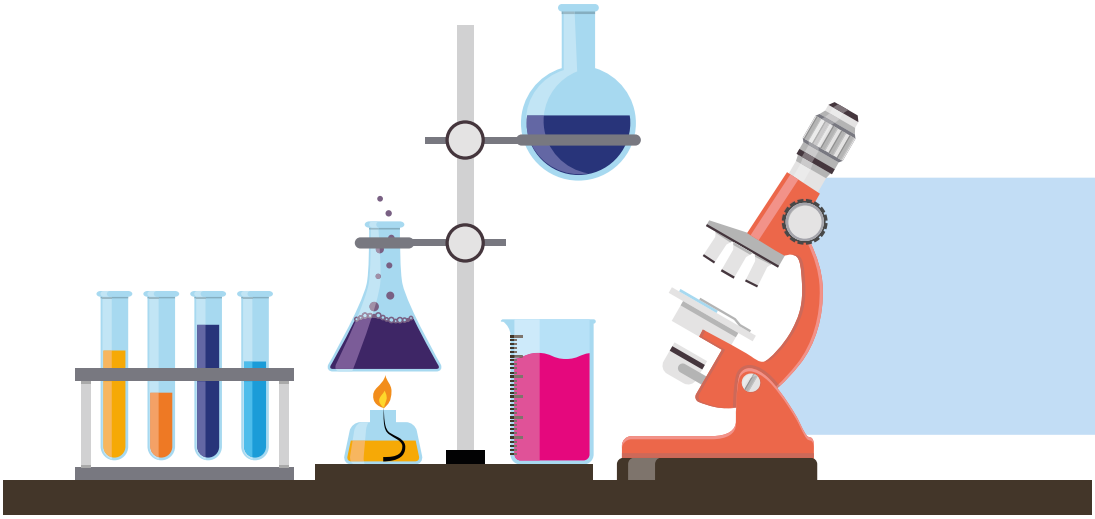
# INTRODUCTION

Hereditary cancers account for approximately 5 to 10% of cases in the general population (Foulkes, 2008; Garber & Offit, 2005). Within ethnicities, however, this proportion can vary widely. Women in developing countries have higher rates of deleterious mutations linked to early-onset cancers and, on average, are diagnosed with such cancers 10 years younger than their counterparts in developed nations (Daly & Olopade, 2015; Villarreal-Garza et al., 2013). Cancer genetic counselling and testing services have well documented benefits for at-risk individuals, including improved health behaviours and informed surgical decision-making (Schwartz et al., 2004; Watson et al., 2005). In a cascading effect, these benefits can be passed on by patients to their families leading to widespread improvements in hereditary cancer risk management (George, Kovak, & Cox, 2015; Mendes, Paneque, Sousa, Clarke, & Sequeiros, 2016). Despite the documented evidence surrounding the benefits of cancer genetic testing and counselling (CGTC), uptake has remained below expected levels, especially among at-risk ethnic groups (Willis et al., 2017).

Central to CGTC is the effective communication of cancer genetic information by clinicians to at-risk individuals and their families who can then make informed decisions to proactively seek genetic counselling and testing (Chopra & Kelly, 2017). Lower uptake rates in at-risk populations raise concerns over the awareness of, and accessibility to, these services while the adequacy of healthcare services to meet the demands of genetic consultations, especially in the increasingly more diverse populations in Western nations, has been called into question (Godard et al., 2003; Mikat-Stevens, Larson, & Tarini, 2015). Godard et al. (2003) examined European-wide social, ethical, and legal impacts on the provision of CGTC and highlighted equality of access as a key concern among professional experts in the field, thus echoing this concern.

Successful interventions have been implemented across various populations to improve the availability of cancer genetic counselling and testing. Examples include communication technologies (Buchanan et al., 2015), educational programmes for physicians (Carroll et al., 2011), targeted videos (Hurtado-de-Mendoza et al., 2019), and pre-appointment family history questionnaires (FHQs) (Kessels et al., 2017). These interventions, although effective, address only specific components of CGTC accessibility. The exact aetiology for the underrepresentation of ethnic groups is not fully understood but is most likely multi-faceted with a potential combination of individual, service-level, and national-level factors. These efforts to improve genetic testing and counselling uptake will be made most fruitful when synergistically combined within a specifically designed model of access that is informed by current evidence. Therefore, this systematic review aims to examine the international literature on patients, families, and clinicians' perspectives and experiences in accessing and providing cancer genetic testing and counselling. It specifically aims to address the following objectives:

1. Identify barriers and facilitators to accessing cancer genetic testing and counselling at individual, service, and national levels.
2. Identify disparities among populations in terms of access to CGTC.



# METHODS

Guidance from the Cochrane Library was used to conduct this review (Higgins et al., 2019), which is reported according to the Preferred Reporting Items for Systematic Reviews and Meta-Analyses checklist (Moher, Liberati, Tetzlaff, & Altman, 2009).

## ELIGIBILITY CRITERIA

The review inclusion and exclusion criteria were formulated beforehand. Empirical studies were included if they incorporated the following characteristics: (i) sample population: healthcare professionals (HCP), patients, or family members; (ii) area of interest: perspectives, opinions, and data relating to access to or experience of accessing CGTC (genetic testing or counselling), accessing assessments for hereditary cancer assessments and counselling, seeking referral to CGTC, barriers or facilitators to accessing CGTC, and/or disparities in accessing CGTC. Studies were excluded from the review if they (i) assessed the frequency of mutations types and/or influencing factors; (ii) assessed interest, attitudes or knowledge of cancer screening and results of surveys about interest in cancer screening in general populations; and (iii) tested targeted interventions to increase access to CGTC. Relevant data from experimental studies not directly related to the intervention were included for analysis. Conference abstracts, theses, dissertations, opinion pieces, and case studies were excluded.

## SEARCH STRATEGY AND STUDY SELECTION

A systematic search of CINAHL and MEDLINE was conducted on March 10, 2020 for eligible studies published between January 2010 and March 2020 in English. The following keywords were searched on title or abstract and combined using Boolean operators "OR" and "AND": Cancer AND (test OR tests OR testing OR test* OR service OR services OR service* OR counselling OR counseling) AND (genetic OR genetics OR genetic*). All records identified were exported to Covidence online software package for screening (Veritas Health Innovation, Melbourne, Australia). Records were screened by title, abstract and full text. Screenings were conducted independently by four reviewers. Each study was screened twice for a screening decision to be made. Screening conflicts were resolved by a third independent reviewer.

## QUALITY APPRAISAL

The tools used to appraise methodological quality were selected based on study design. Quantitative descriptive studies, qualitative studies, non-randomised studies, randomised-controlled trials, and mixed-methods studies were appraised critically using the Mixed-Method Appraisal Tool (MMAT) (Hong et al., 2018). Each item was judged on a "Yes," "No," or "Can't tell" basis. The Joanna Briggs Institute's critical appraisal tool (Joanna Briggs Institute, 2017) was used to assess the methodological quality of the included systematic reviews and each item was rated as "Yes," "No," "Unclear," or "Not applicable."

## DATA EXTRACTION AND SYNTHESIS

The following were extracted from each study using a standardised data extraction form (i) study characteristics (authors name, country, year of study, study design); (ii) sample characteristics (sample size, cancer types/test types, area of focus); and (iii) findings (barriers, facilitators, and disparities). Findings were analysed and synthesised narratively based on the aim and objectives.

# RESULTS

## STUDY SELECTION

A total of 2,453 recorded were identified from the search. Following deletion of duplicates, 2,053 records were screened on title and abstract and 1,763 irrelevant records were excluded. Full texts of 290 papers were then screened. Of those, 122 studies were included in this review (Figure 7).

## STUDY CHARACTERISTICS

Most studies were conducted in the USA (n=79) and were quantitative descriptive (n=56), qualitative (n=31), or non-randomised studies (n=21) (Table 4). Sample size ranged between 12 (Spencer, Rodgers, & Coffey, 2019) and 49,721 (Han & Jemal, 2017) participants. Approximately, half of the studies were conducted during genetic counselling (n=34), in cancer genetic services (n=28), or during genetic testing (n=26), using surveys (n=25), telephone interviews (n=19), or questionnaires (n=15). Individual-level barriers (n=234) and facilitators (n=156) to cancer genetic testing were the most reported outcomes in the included studies.

## QUALITY APPRAISAL

More than half of the qualitative studies met all MMAT criteria. Two qualitative studies did not have clear research questions (Duquette, Lewis, McLosky, & Bach, 2012; Halverson, Wessinger, Clayton, & Wiesner, 2020). Both randomised controlled trials met all MMAT criteria except for blinding the outcome assessor (Kinney et al., 2014; Kinney et al., 2016). Less than half of the included mixed-methods studies met the majority of MMAT quality criteria. Threats to methodological quality in the included systematic reviews related to the likelihood of publication bias and the absence of two or more independent reviewers. The majority of non-randomised studies had a representative sample and clear research questions, data collection processes, and outcome measures. Almost all quantitative descriptive studies demonstrated appropriate statistical analysis. However, the risk of nonresponse bias and of sample unrepresentativeness was not clear in almost half of these studies.

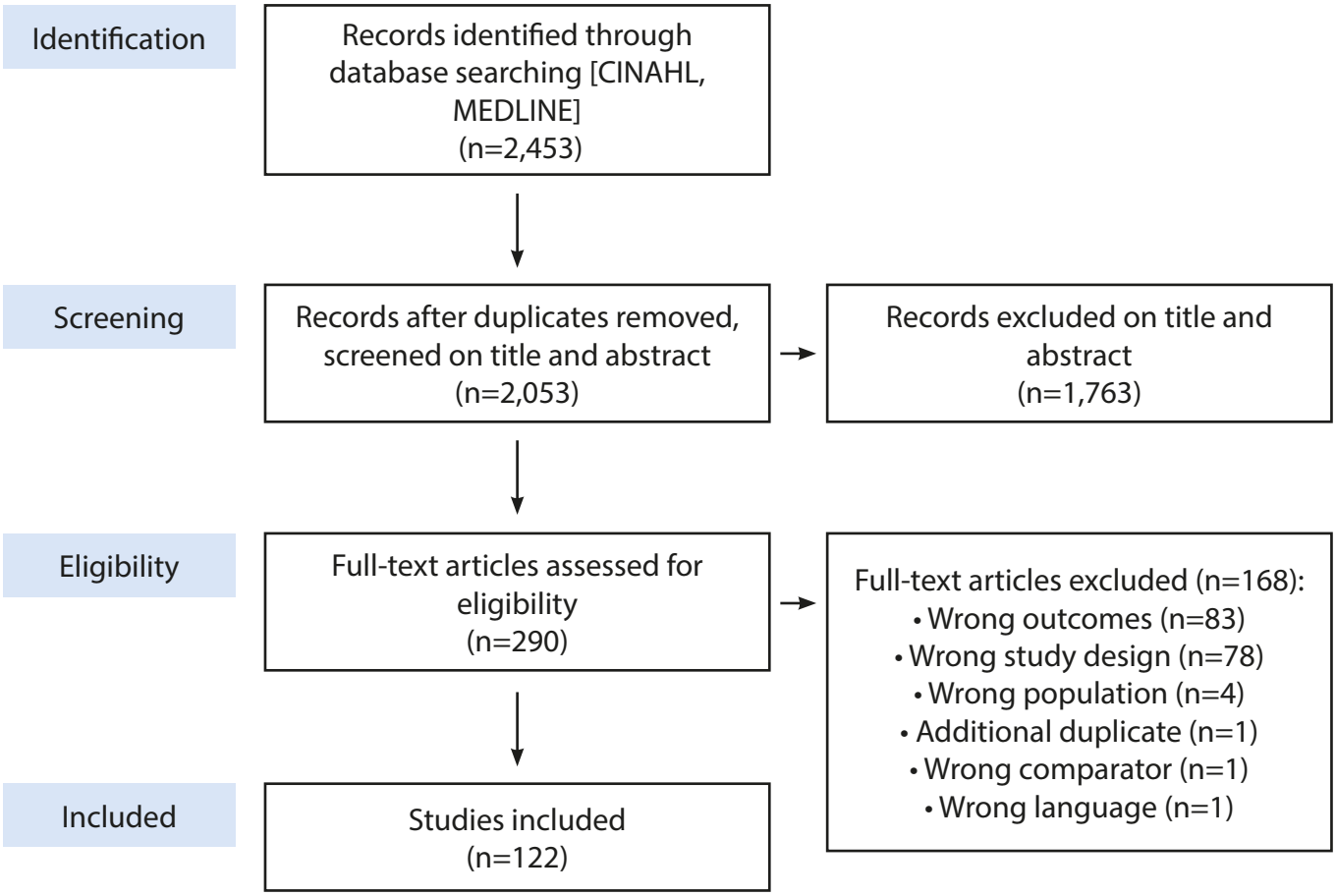



**Figure 7. Study identification, screening, and selection process.**

**Moher David, Liberati Alessandro, Tetzlaff Jennifer, Altman Douglas G. Preferred reporting items for systematic reviews and meta-analyses: the PRISMA statement BMJ 2009; 339:b2535**

## Table 4. Characteristics of the included studies (n=122).

| | |
|---|---|
| Country | USA (n=79)<br>Australia (n=8)<br>Canada (n=8)<br>UK (n=8)<br>Netherlands (n=6)<br>Singapore (n=4)<br>Germany (n=2)<br>South Korea (n=2)<br>France (n=1)<br>Japan (n=1)<br>Kenya (n=1)<br>Malaysia (n=1)<br>Not available (n=1)<br>Sweden (n=1)<br>Switzerland (n=1) |
| Research design | Quantitative descriptive studies (n=56)<br>Qualitative studies (n=31)<br>Non-randomised studies (n=21)<br>Qualitative and quantitative descriptive studies (n=6)<br>Systematic reviews (n=5)<br>Randomised controlled trials (n=2)<br>Qualitative with non-randomised study (n=1) |
| Sample size (min-max) | 12–49,721 |
| Settings | Genetic counselling (n=34)<br>Cancer genetic services (n=28)<br>Genetic testing (n=26)<br>Hereditary cancer risk assessment (n=11)<br>Others (n=23) |
| Data collection | Surveys (n=25)<br>Telephone interviews (n=19)<br>Questionnaires (n=15)<br>Retrospective data analysis (n=14)<br>Records (n=12)<br>Focus groups (n=5)<br>Others (n=32) |
| Outcomes** | Individual-level barriers to cancer genetic testing (n=234)<br>Service-level barriers to cancer genetic testing (n=36)<br>National-level barriers to cancer genetic testing (n=29)<br>Individual-level facilitators to cancer genetic testing (n=156)<br>Service-level facilitators to cancer genetic testing (n=39)<br>National-level facilitators to cancer genetic testing (n=19) |

** Some studies reported on more than one country.*

***Studies often reported on more than one outcome. n corresponds to the number of times an outcome was reported.*

## BARRIERS TO CANCER GENETIC TESTING

An overview of the frequency of barriers is presented in Figure 8. Of the 122 studies reviewed, 92 reported different barriers to CGTC and associated disparities. Individual-level barriers were most prevalent with the cost of genetic testing as the overarching barrier (n=33). Additional important barriers include fear of positive results and their impact on insurability and employment (n=24), insurance concerns (n=23), and lack of knowledge and awareness regarding cancer genetics and services (n=20).

Among the service-specific barriers, a lack of referral/recommendation (n=10) coupled with insufficient knowledge and awareness of CGTC and genetics in general among HCPs (n=9 and n=7 respectively) were mostly reported. The most commonly reported barrier at a national level was the geographical location of CGTC centres (n=14), followed by a lack of genetic services and genetic workforce (n=5), and difficulty navigating the healthcare system/systemic barriers (n=4).

Disparities in access across populations were evident. Ethnic disparities were most frequently reported (n=21). The majority of studies highlighting ethnic disparities were based in the USA (n=15) with individuals from African-American or Latino ethnic groups identified as those most marginalised in terms of accessing CGTC. Differences in access were also evident across age groups (n=13) with younger people typically more likely to access testing services compared to older people. Health insurance coverage (n=9), educational level (n=6), differences in socioeconomic status (n=4), and gender (n=2) were also reported to affect equitable use of CGTC, albeit to a lesser extent.

## FACILITATORS TO CANCER GENETIC TESTING

An overview of the frequency of facilitators is presented in Figure 9. Of the 122 studies reviewed, 81 reported different facilitators to CGTC. Individual-level facilitators were the most frequently reported. Knowledge of familial history (n=21), proactive health attitudes and beliefs (n=20), and family obligation, responsibility, and support (n=20) were the facilitators most often reported at an individual level.

In terms of service-specific facilitators, the use of alternative methods to deliver counselling, such as the web or telephone (n=6), was identified as the most important facilitator. This was followed by HCP access to training on CGTC (n=5), access to professional/national guidelines on cancer genetic testing (n=4), awareness of CGTC and benefits (n=4), and strategies to facilitate access to and efficiency of appointments (n=4). The most common national level facilitator was found to be positive health behaviours among socially influential individuals (n=5), which largely related to celebrities such as Angelina Jolie and her diagnosis with the BRCA gene. National awareness campaigns (n=4) and availability of information in different languages (n=3) were also important.

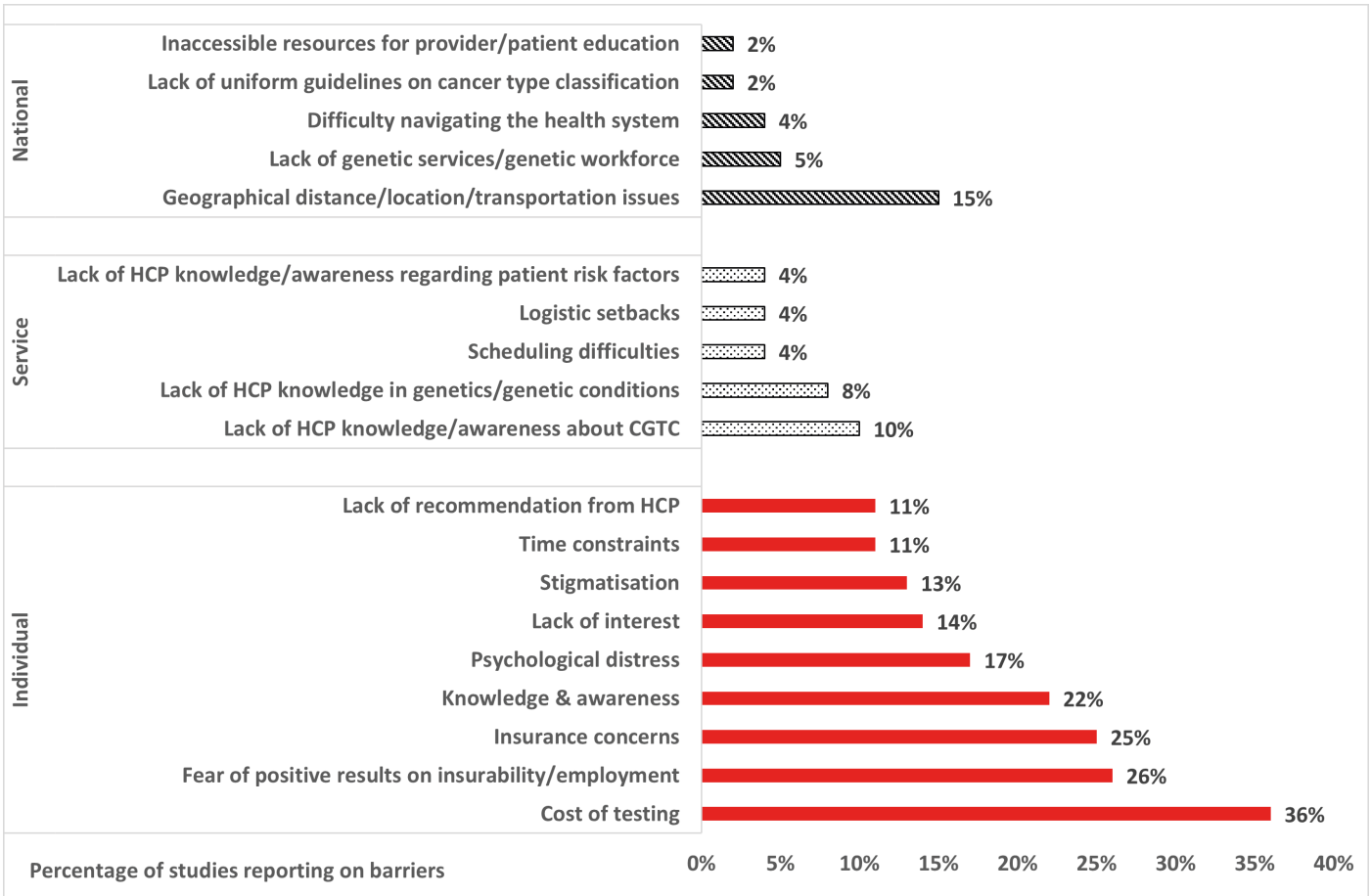



**Figure 8. The frequency of barriers within studies reporting on barriers (n=92)**

**Abbreviations: CGTC, cancer genetic testing and counselling; HCP, healthcare professional.**

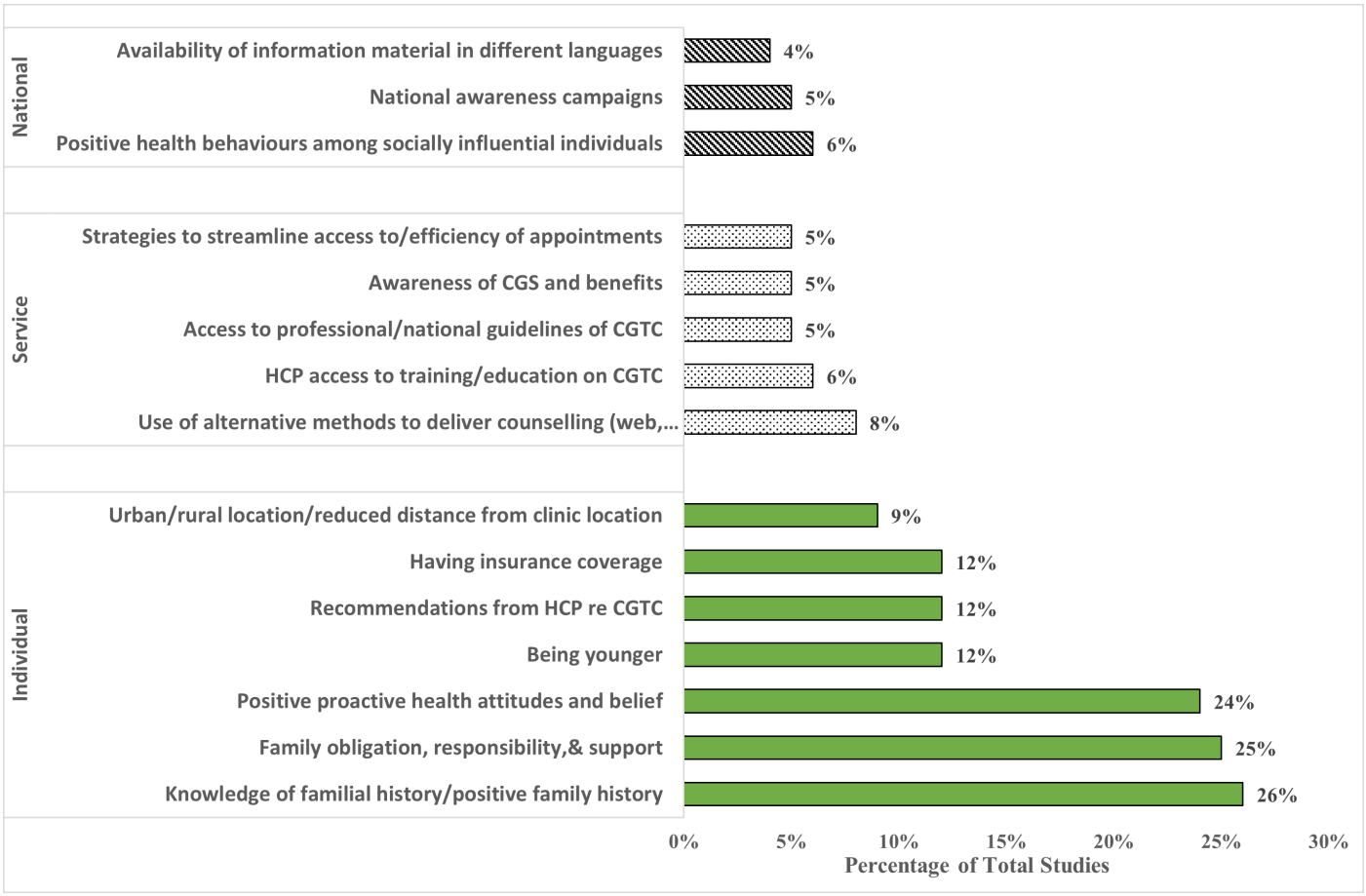



**Figure 9. The frequency of facilitators within studies reporting on facilitators (n=81)**

**Abbreviations: CGTC, cancer genetic testing and counselling; HCP, healthcare professional.**

# DISCUSSION

## BARRIERS TO CANCER GENETIC TESTING

Drawing on the findings of this review, the cost of genetic testing and insurance concerns are the predominant barriers to accessing CGTC. The majority of the studies highlighting such concerns focused on the USA and are likely related to their highly expensive and unequal healthcare system (Dickman, Himmelstein, & Woolhandler, 2017). This results in high out-of-pocket cost for basic healthcare with a rising percentage of individuals uninsured for healthcare (Berchick, Barnett, & Upton, 2019). Insufficient knowledge of cancer genetics and related services among HCPs were commonly highlighted as service-specific barriers. Scheduling difficulties, logistics, and technological setbacks were also identified and should be addressed together to provide effective access for all (Hurtado-de-Mendoza et al., 2019; Jones et al., 2016; Kentwell et al., 2017). At a national level, the geographical location of the centre was a major barrier to access (Crook et al., 2015; Pokharel, Hacker, & Andrews, 2017; Tutty et al., 2019). Patients and their families in rural areas may incur significant additional travel costs and time compared to their urban counterparts. This may add to the perceived excessive cost of accessing CGTC and potentially result in the marginalisation of those living in rural areas.

Advances in genetic and medical technology can reduce as well as exacerbate disparities in healthcare (Godard et al., 2003; Susswein et al., 2008). There appears to be specific underutilisation of CGTC by certain ethnic sub-populations, even when cost and insurance are covered (Baars, van Dulmen, Velthuizen, van Riel, & Ausems, 2017; Cheng et al., 2018; Shaw et al., 2018; Susswein et al., 2008). Latinos, Asian Americans, and African Americans in the USA plus Asian Australians were the ethnic groups most marginalised in the reviewed literature (Hurtado-de-Mendoza et al., 2019; Jones et al., 2016; Manrriquez, Chapman, Mak, Blanco, & Chen, 2018; Shaw et al., 2018; Spencer et al., 2019). In a European context, migrants were less likely to be referred for genetic counselling (Allford, Qureshi, Barwell, Lewis, & Kai, 2014; Nilsson, Nilsson, Silfverberg, Borg, & Loman, 2019; van der Giessen, van Riel, Velthuizen, van Dulmen, & Ausems, 2017; van Riel, van Dulmen, & Ausems, 2012). Despite a willingness to access CGTC services (Cheng et al., 2018; Cragun, Weidner, Kechik, & Pal, 2019), these minority groups are impeded by lower rates of referral for testing, less familiarity with the healthcare system, and a lack of culturally appropriate information (Allford et al., 2014; Augusto, Kasting, Couch, Lindor, & Vadaparampil, 2019; Gammon et al., 2011; Hurtado-de-Mendoza et al., 2019). Age disparities were also prevalent. Younger women (aged ≤ 50 years) were more likely to use and access hereditary breast and ovarian cancer risk assessment and testing services compared to older women (Chew et al., 2017; Demsky et al., 2013; Gammon et al., 2011; Hull, Haas, & Simon, 2018; Hurtado-de-Mendoza et al., 2019; Jones et al., 2016; Jones et al., 2019; Katz et al., 2018; Nikolaidis et al., 2019; Nilsson et al., 2019). However, two studies in the UK observed that younger people were less likely to participate in genetic cancer risk assessment through FHQs (Dancyger, Smith, Jacobs, Wallace, & Michie, 2010; Hanning et al., 2015).

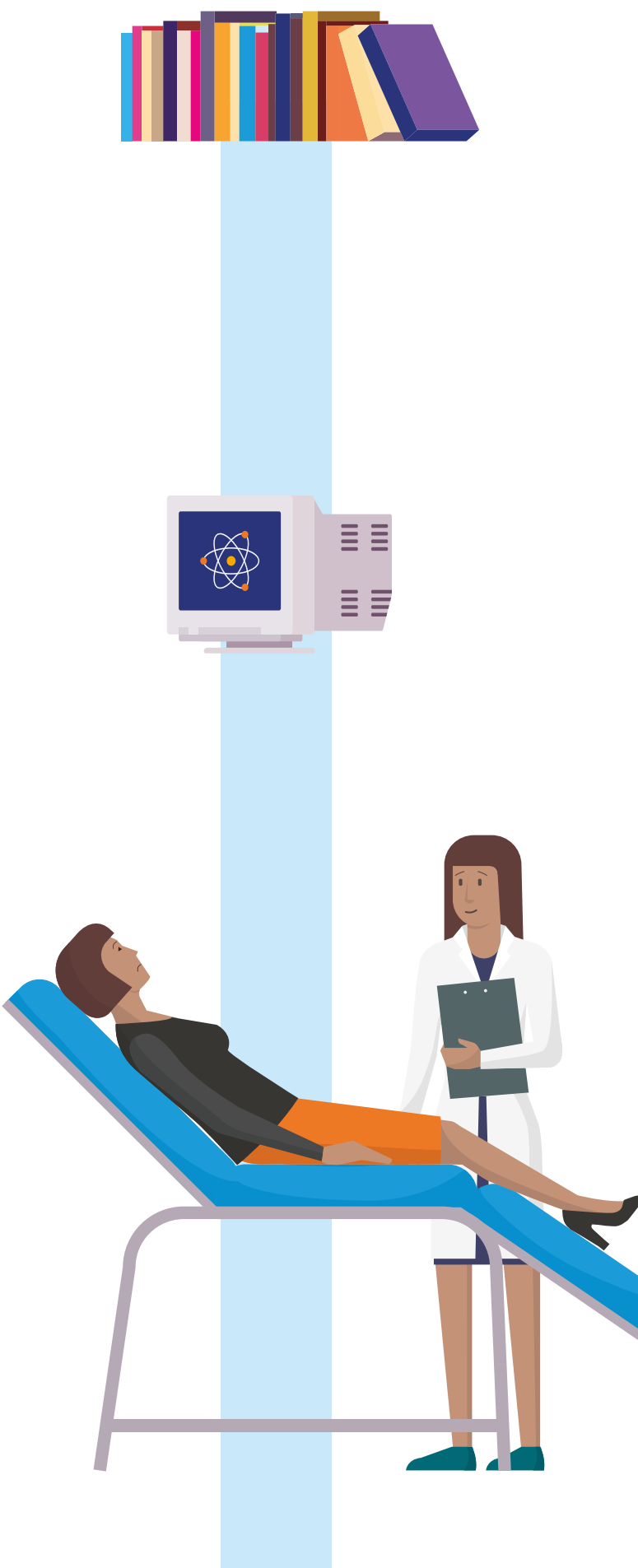
## FACILITATORS TO CANCER GENETIC TESTING

Drawing on the available literature, several factors, at differing levels, can support access to CGTC. An overwhelming majority of studies focused on individual level facilitators demonstrating a clear drive to empower individuals to be proactive in their cancer risk management. Knowledge of family history or a positive family history was the most mentioned facilitator (Allford et al., 2014; Hull et al., 2018; Jones et al., 2019; Nikolaidis et al., 2019; Nilsson et al., 2019). Family obligation, responsibility and support were familial dynamics that may also serve as facilitative factors (Dancyger et al., 2010; Dekker et al., 2013; Sussner, Jandorf, Thompson, & Valdimarsdottir, 2010; Vogel et al., 2018). An individual's family unit were identified as an important resource in improving access (Hurtado-de-Mendoza et al., 2019). Their importance has been discussed in other studies where family are vital in providing information, support, and social influence around cancer and testing, and are seen to play an important role in decision making (Ashida et al., 2011; Hobbs et al., 2015). Furthermore, shared decision-making can improve health outcomes with family being part of this process (Boland et al., 2019; Grad et al., 2017; Peek et al., 2010). It is vital that HCPs are aware of the key role of the family and their potential influence on continued cancer risk management by the patient.

The use of alternative methods to deliver cancer genetic counselling, such as the web or telephone (Buchanan et al., 2015), was the most common facilitator identified at the service-level. This is encouraging as it suggests that individuals are more willing to engage when measures are put in place that provide service information and ease access to CGTC. At a national level, positive health behaviours among socially influential individuals was the biggest driver of engagement with CGTC. For example, Angelina Jolie's public announcement of her use of BRCA gene testing services was cited as a basis for increases in overall BRCA testing rates, in what became known as the "Angelina Jolie effect" (Lee et al., 2017). Health behaviours among celebrities is often seen to influence individuals' health behaviours, both positively and negatively, and has been identified as an important public health issue (Brown & Basil, 1995; S. J. Hoffman et al., 2017; Steven J Hoffman & Tan, 2013; S. J. Hoffman & Tan, 2015). It has also been suggested that the influence of celebrities and the media on health behaviours, when regulated and monitored, can be harnessed for positive use, and that public health authorities can work in partnership with celebrities to endorse positive health behaviours (Steven J Hoffman & Tan, 2013). Noticeably few other national-level facilitators were identified indicating a potential deficit in the available literature.

# STRENGTHS AND LIMITATIONS

This review provides up-to-date peer-reviewed evidence regarding barriers and facilitators to cancer genetic testing and counselling in different contexts. Rigour was sought in the conduct and reporting of this review. Record screening was conducted by independent reviewers and data extraction and quality appraisal were cross-checked to minimise reporting errors. However, the search was limited to studies published in English between January 2010 and March 2020; therefore, older studies and studies published in languages other than English were not included. Many studies were carried out in the USA which may limit the generalisability of findings beyond the unique socio-cultural and economic context of the USA. Another limitation in terms of generalisability is a clear lack of research into male-specific cancers and cancer risk management.



# CONCLUSION

The present review highlights common barriers and facilitators to accessing CGTC. Individual-level barriers were predominantly linked to cost of testing and lack of insurance. Lack of referral coupled with insufficient knowledge and awareness of CGTC were identified as service-level barriers. Nationally, the geographical location of CGTC was a key barrier. These barriers were noticeably more pronounced among ethnic groups. In relation to facilitators, awareness of family history and positive health attitudes served as individual-level enablers to accessing CGTC. At service-level, the use of technology was identified as a key facilitator. Influential figures, particularly celebrities, were identified as national-level facilitators to accessing CGTC.

The identified barriers and facilitators should be considered in future research and health policy and education to develop approaches that increase access and uptake of genetic counselling and testing. A focus on implementing practices that address these factors in a multi-level framework, i.e. at individual, service, and national levels, is recommended. Special attention should be paid to the needs of ethnic and minority groups given the clear disparities in place. While several barriers have been identified as contributing to underutilisation of CGTC, further research is necessary to ascertain underlying factors to these barriers and determine how they can be effectively tackled. Developing strategies that seek to improve CGTC access through educating HCPs while simultaneously creating awareness in communities about CGTC and its benefits offer a potential means of further exploration. Continuous evaluations of such strategies can be undertaken in parallel to examine community participation and the impact of creating awareness on referral and/or mortality rates. Such insights can build on the existing evidence base to support increased uptake of CGTC to achieve the associated benefits for at-risk patients and their families.

# RECOMMENDATIONS

The number of individuals diagnosed with cancer annually is on the rise. Inherited genetic mutations play a major role in about 5 to 10 percent of all cancers, though the contribution to individual cancers varies widely. A proportion of cancers are familial and involve mutations of multiple susceptibility genes that increase an individual's risk of cancer. Researchers have associated mutations in specific genes with more than 50 hereditary cancer syndromes.

The assessment of an individual's genetic profile plays a critical role across the continuum of cancer care from screening to the use of targeted therapies. A large proportion of the work of any cancer genetic service is the management of familial colorectal, breast and ovarian cancer, and these areas exemplify opportunities for increased access to gene testing and follow-up support in the first instance.

A reduction in the life burden caused by cancer can be achieved by implementing enhanced surveillance and timely evidence-based interventions. Even with improvements in the understanding of the role of genetic information in cancer care, health care providers globally face many challenges in providing uniform access to timely genetically guided health and oncology care. Progress towards more individualised and family-centered oncology care requires enhanced understanding of genetic and genomic information by patients, their health care providers and policy makers.

Within chapters one and two the barriers and facilitators to accessing cancer genetic counselling and testing are outlined from both an Irish and International context, respectively. In the last two decades there have been rapid and revolutionary development of genomic technologies and extensive advances in knowledge of the impact of genomic variation on human disease. Cancer genetics is becoming a routine component of mainstream oncology clinical practice. Early and uniform access to cancer genetic testing based upon predefined criteria can lead to a reduction in morbidity and mortality.

However relatively low rates of referral for genetic counselling and testing are compounded by lack of patient and health care professional awareness of genetics services and barriers to their utilization. Both the qualitative and quantitative data reaffirmed that the health care professionals within the genetic services are doing a good job. However, the public cancer genetic services are overstretched. Thus, it is a challenge for many to receive timely access to genetic testing and the associated results.

Several recommendations emanate from this study. But many of these recommendations are not new as they have already been cited in prior strategy documents. Financial resourcing and full implementation of the National Cancer Strategy 2017–2026 recommendations around genetic services is key to reducing genetic testing wait times and to optimise the potential health benefits of advancements in our undertaking of the genetic basis of many cancers. The challenge remains to ensure timely and appropriate implementation of all the recommendations outlined within the National Cancer Strategy.

| Recommendations from this study | Quotes taken directly from the National Cancer Strategy 2017–2026 (unless otherwise stated) |
|---|---|
| **Implement a hub and spoke model with genetics expertise within the dispersed oncology system Genetics needs to be formally integrated into the cancer treatment pathway with uniform access to genetic testing, molecular tumour boards and access to genetics expertise and support at the point of care for both patients and their clinicians.** | To meet present demand, additional full-time consultant appointments will be required in the National Cancer Genetics Service to facilitate the delivery of a high-quality service, using a hub and spoke model involving the active participation of surgeons and/or physicians in individual cancer centres to generate a deeper engagement with cancer genetics at a local level.<br><br>National management protocols for common predisposition syndromes will enable decentralisation of care for common genetic disorders and facilitate nurse/counsellor-led clinics in cancer centres nationally, backed up by appropriate consultant-led clinical governance. Oncology care in all designated cancer centres will require input from the Clinical Cancer Genetics Service (pg. 76).<br><br>We have an opportunity now to develop an integrated cancer genetics service, which will provide an infrastructure for a time when genetics-based clinical care is commonplace, by appointing at least one cancer genetics nurse specialist/counsellor in each designated cancer centre (pg. 77). |
| **Build and further develop the genetics workforce and capability.** | --a directed effort to train Irish oncology graduates in genetics will be required. To meet present demand, additional full-time consultant appointments will be required (pg. 76).<br><br>Clinical cancer genetics in Ireland requires a strategic approach that will include increased infrastructural and financial support (pg. 75). |
| **Increase cancer genetics diagnostics capability and expertise in Ireland** | There is a lack of a co-ordinated genetic testing service in Ireland, due to funding issues in the main. A fully functioning national service cannot be accommodated. This has led to poor practice in terms of testing requests and also poor-quality foreign laboratories handling Irish samples (Review of The HSE and NDTP: Clinical Genetics Medical Workforce in Ireland, 2019). |
| **Use a data management system that tracks referrals, appointments, and receipt of diagnosis with associated key performance indicators in terms of time to appointments, time to receipt of genetic test results and time to receipt of follow-up interventions (if required).** | Waiting lists in the three hospitals are extensive, and patients often have prolonged waits for their results. Thus, results with therapeutic relevance for patients undergoing treatment are delayed and healthy individuals are not being informed of their inherited cancer risk in a timely way (pg. 75).<br><br>--- need for a coordinated national recording of genetic test results and an associated method of communication (pg. 77). |
| **Streamline the genetics pathway to optimise online data collection and processing of data ensuring that follow-up counselling and health promoting interventions for individuals with positive mutations is optimised.** | A clear strategy for dealing with patient information obtained from genetic testing will need to be developed (pg. 74). |

| Recommendations from this study | Quotes taken directly from the National Cancer Strategy 2017–2026 (unless otherwise stated) |
|---|---|
| **Increase knowledge and awareness of health care professionals, patients and the public of genetics and genetic services.** | A further major challenge is to ensure that the education and training of all those involved in this area keep pace with the scientific and technical developments (pg. 74). |
| **A dedicated pathway for individuals with specific syndromes or mutations with audited quality assured key performance indicators is required e.g. BRCA, Hereditary Breast and Ovarian Cancer Syndrome, Lynch Syndrome. Such pathways will ensure coordination of timely access to evidence-based surveillance, screening, surgery and treatments as needed for individuals with specific mutations.** | The NCCP will further develop the Programme for Hereditary Cancers to ensure that evaluation, counselling, testing and risk reduction interventions are available as appropriate, and that services are available to patients on the basis of need (pg. 76).<br>Regular multidisciplinary clinics should be established for less common cancer predisposition syndromes to ensure appropriate care for patients with these disorders, as well as to facilitate Ireland's participation in international efforts to develop a unified approach for such cases (pg. 77). |
| **Test interventions that support the communication of information relating to genetic mutations with family members.** | |
| **Explore and address the barriers to cascade testing of at-risk relatives** | Audits will ensure that equitable access is available irrespective of patients' age, geographic location, and socioeconomic status (pg. 76). |
| **Address concerns relating to the management of clinical samples and genetics data.** | A clear strategy for dealing with patient information obtained from genetic testing will need to be developed (pg. 74). |

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