National Cancer Control Programme

Hereditary Cancer Model of Care

HSE National Cancer Control Programme

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NCCP-COM-100 NCCP Hereditary Cancer Model of Care

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Foreword

Cancer remains a significant cause of illness and death in Ireland, with one in two people expected to develop cancer in their lifetime. Addressing hereditary cancer risk is a powerful tool, as it is estimated that inherited gene variants are responsible for between 5 and 10% of cancers.

The potential benefits of identification and management of an inherited cancer predisposition are far-reaching, with implications for both a person diagnosed with cancer and their family members. The benefits can include reducing the risk of cancer developing, identifying cancer at an earlier more treatable stage and informing the optimal choice of targeted therapy if a cancer develops.

I strongly welcome the development of this model of care, which provides the blueprint for the necessary development of hereditary cancer services in Ireland. I also look forward to ongoing collaboration with the National Genetics and Genomics Office, on shared priorities for hereditary cancer services in the *National Strategy for Accelerating Genetic and Genomic Medicine in Ireland.*

I would like to acknowledge members of the Advisory Group, who lent their time and expertise to the development of this model of care, with particular thanks to our four patient representatives. The broad membership of the Advisory Group highlights the range of health care disciplines involved in the area of hereditary cancer and who will be key to the implementation of this model of care.

Professor Risteárd Ó Laoide

National Director

National Cancer Control Programme

Purpose

This document has been developed on behalf of the NCCP Advisory Group on Hereditary Cancer (see Appendix B for membership). Its purpose is to inform development of services for the identification and management of people with an inherited cancer predisposition. The necessary laboratory testing and expertise required for such services is outside the scope of this document and will be considered elsewhere.

Abbreviations

Abbreviation	Definition
ANP	Advanced Nurse Practitioner
AYA	Adolescents and Young Adults
CAYA	Children, Adolescents and Young Adults
CHI	Children's Health Ireland
CNS	Clinical Nurse Specialist
DNA	Deoxyribonucleic acid
DOH	Department of Health
EHR	Electronic Health Record
ERN	European Reference Network
EU	European Union
GDPR	General Data Protection Regulation
GP	General Practitioner
HBOC	Hereditary Breast and Ovarian Cancer Syndrome
HSE	Health Service Executive
ICS	Irish Cancer Society
IT	Information Technology
KPI	Key Performance Indicators
MDT	Multi-Disciplinary Team
NCCP	National Cancer Control Programme
NCCS	National Children's Cancer Service
NCRI	National Cancer Registry Ireland
NGS	Next Generation Sequencing
NHS	National Health Service
NICE	National Institute for Health and Care Excellence
PPI	Public and Patient Involvement
PPPGs	Policy, Procedures, Protocols and Guidelines
RHA	Regional Health Area
SJH	St. James's Hospital
UKCGG	UK Cancer Genetics Group
VUS	Variant of Uncertain Significance
WGS	Whole Genome Sequencing
WHO	World Health Organization
WTE	Whole-Time Equivalent

Glossary

Name	Definition
Advanced Nurse Practitioner (ANP)	The advanced practice service is provided by nurses who practice at a higher level of capability as independent, autonomous and expert advanced practitioners. The overall purpose of the service is to provide safe, timely, evidenced based nurse-led care to patients at an advanced nursing level. This involves undertaking and documenting complete episodes of patient care, which include comprehensively assessing, diagnosing, planning, treating and discharging patients in accordance with collaboratively agreed local policies, procedures, protocols and guidelines and/or service level agreements/ memoranda of understanding.
Carrier	A person carrying a pathogenic variant or fault in a gene. For many cancer predisposition syndromes, a fault in one copy of the gene is enough to increase their risk of developing cancer. For other conditions, including some less common cancer predisposition syndromes, both copies of the gene must be affected for disease to occur. In this case, a carrier is not themselves at risk of the syndrome but they may still pass on the faulty gene conferring that risk to their children.
Cancer predisposition	Having a predictable increased risk of developing cancer compared to the general population.
Clinical Nurse Specialist (CNS)	A CNS is an expert practitioner who has attained, at a minimum, a post graduate qualification in their specialist area of practice. The role is defined under five core competencies - clinical, education, consultation, advocacy and audit. This specialist practice encompasses a major clinical focus of care to patients or clients and their families in hospital, community and outpatient settings.
CORU	CORU is Ireland's multi-profession health and social care regulator. The name CORU originates from an Irish word, "cóir" meaning fair, just and proper. CORU regulates individuals who provide health and social care services in Ireland.
Diagnostic testing	See *genetic testing*
Gene	The basic physical and functional unit of heredity. Genes, which are made up of DNA, act as instructions to make molecules called proteins.
Genetic testing	**Germline genetic testing** tests for the presence of germline genetic variants - A germline genetic variant (or germline mutation) is a gene change that is passed down from parent to offspring, then incorporated into the DNA of every cell in the body of the offspring. Germline testing is usually carried out on blood, saliva or skin. Germline gene variants are also known as hereditary gene variants.

Genetic testing	**Somatic genetic testing** tests for the presence of somatic mutations – A somatic mutation is an alteration in DNA that is not inherited. It may arise sporadically or environmental/ lifestyle factors may induce a somatic change. Somatic mutations can occur in any cell of the body except for germ cells and therefore are not passed on to any offspring. These alterations can (but do not always) cause cancer or other diseases. **Diagnostic testing** is testing offered to symptomatic people to determine whether they have a condition, e.g. a cancer predisposition syndrome, associated with the symptoms they already have, such as an existing personal diagnosis of cancer. **Cascade testing** is the genetic testing carried out in individuals within a family, looking for a pathogenic variant previously identified in a blood relative. This process is repeated as more individuals with the pathogenic variant are identified within the family. Note that the term **predictive testing** is commonly used in genetics when testing for a specific known pathogenic variant. Its use is avoided in this document due to the potential for confusion, as in medical oncology it is typically used to describe testing to predict likely response to a drug treatment.
Genetics	Genetics – the study of genes, genetic variation, and heredity in living organisms.
Genomics	Genomics - the application of genome-based knowledge through the study of genes and other genetic information, their functions and interrelationships for the benefit of human health.
Germline genetic testing	See *genetic testing*
Hereditary cancer services	Used in this document to refer to patient-facing services from appropriate identification of people who may have an inherited cancer predisposition (germline genetic variant/mutation), through assessment and testing, and appropriate clinical management and support to maximise health and wellbeing. Hereditary cancer services are reliant on laboratory services and expertise but the configuration and delivery of laboratory services is outside the scope of this model of care.
Mainstreamed testing/ main-streaming	Diagnostic genetic testing for patients outside of genetics services and as part of standardised cancer care and in line with nationally agreed criteria and protocols, including appropriate education and training.
Pathogenic variant	An alteration in a gene, which leads to an increased risk of disease, e.g. cancer. Used in preference over the term gene 'mutation'.
Predictive testing	See *genetic testing*
Somatic testing	See *genetic testing*
Variant of Uncertain Significance	A Variant of Uncertain Significance (VUS) is a genetic change that cannot be interpreted as either harmless (benign) or potentially disease-causing (pathogenic), as the meaning of this type of genetic change is not known at this point in time.

Executive Summary

Genetics is now recognised as a growing and integral part of cancer care, with a role across treatment, early detection and risk reduction. Identification of cancer predisposition will only lead to patient benefit, if necessary services are in place and appropriately resourced and monitored. The potential benefit to a patient - and their relatives - can include reducing the risk of cancer developing, identifying cancer at an earlier more treatable stage, informing the optimal choice of targeted therapy if a cancer develops and facilitating reproductive options for the individual.

- **Risk reduction** – Genetics offers a direct and cost-effective way of identifying people who are at high risk of developing cancer. These people can then be managed appropriately, including offering interventions to reduce the risk of cancer developing, e.g. risk-reducing surgery and chemoprevention.
- **Early detection** – People who are identified as having an inherited cancer predisposition can be closely monitored to facilitate early detection and treatment of disease. Diagnosing cancer early typically allows for less complex treatment and optimal outcomes.
- **Treatment** - Certain cancer therapies rely on genetic and genomic information to tailor treatment to give the best chance of cure. For cancer patients with an inherited cancer predisposition, treatment decision-making based on genetic testing is rapidly becoming an international standard of care. Genetic test results guide surgical decisions and direct choice of medication. Targeting medication according to genetic test results is at the forefront of precision medicine.
- **Reproductive counselling and care** – Additional benefits of cancer genetics include the ability to identify people with a cancer predisposition and offer them appropriate counselling and care in relation to their reproductive health and options.
- **Benefit to relatives** – Cascade testing of unaffected relatives identifies more individuals with an inherited cancer predisposition, who can then benefit from the above cancer risk reduction and early detection measures.

The identification of cancer predisposition allows for the implementation of these risk-reduction measures, such as prophylactic surgery and chemoprevention, and surveillance to ensure early detection. In this way, genetic testing - for individuals and subsequent cascade testing of relatives - and provision of comprehensive downstream services can effectively reduce both cancer incidence and cancer mortality.

The vision of this model of care is that it will provide clarity regarding the structure and governance of hereditary cancer services in Ireland. The model of care will ensure/support:

- Development of national guidance to underpin standardised best practice, including mainstreaming of cancer genetic testing
- Equitable access to hereditary cancer services for all
- Clear governance and clarity regarding the roles of national genetics and genomic structures, Specialist Cancer Genetics Services and Regional Cancer Predisposition Services, within an overarching model of care

- Commitment to the Sláintecare principles of right person, right place, right time – through the integration of genetic assessment and testing into regional services, with access to specialist genetic expertise as required
- Maximal use of technological solutions
- Timely access to genetics assessment and testing, including specialist cancer genetics expertise as required
- Timely access to clinical management of cancer predisposition, including risk reduction options
- Comprehensive and coordinated approaches to ongoing management
- Quality assurance of service
- Access to high quality patient information, psychological support and peer support for patients
- Education and training of health care professionals, including accredited training for those delivering mainstreamed pathways

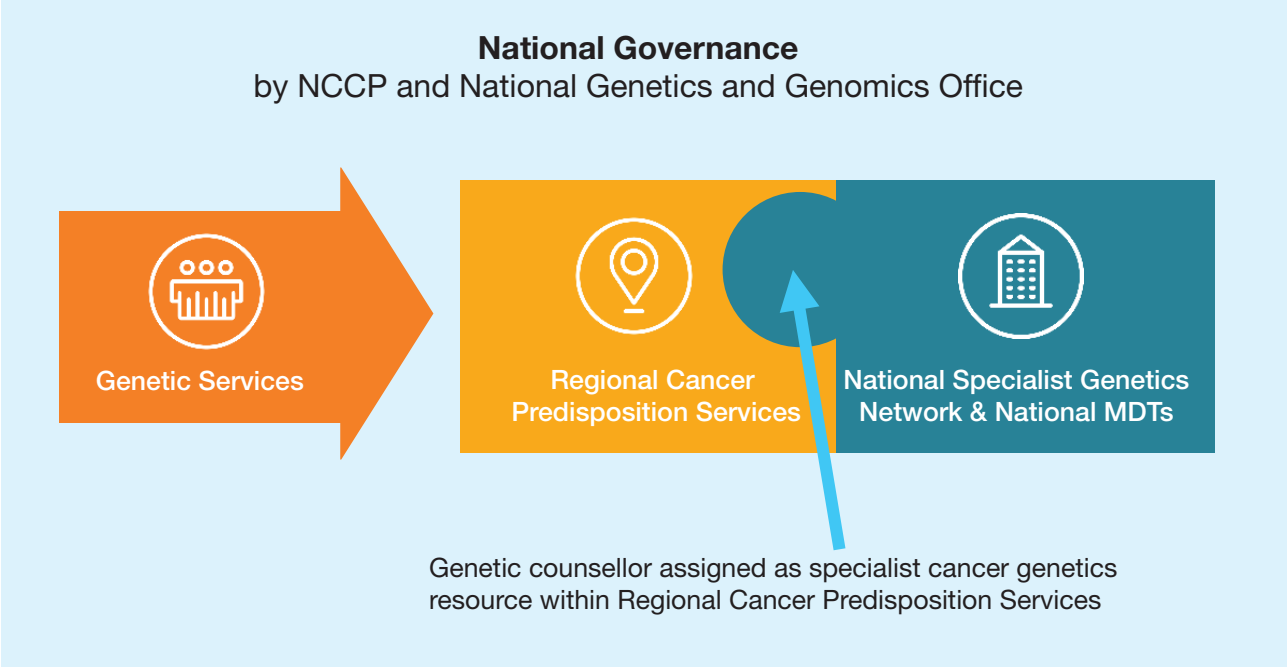
Three tier model of service delivery

A programmatic approach is required to ensure national agreement of guidance, including mainstreaming protocols, laboratory and clinical data infrastructure, specialist genetics workforce development, and education of the wider workforce. This programme will be a collaboration between the National Genetics and Genomics Office and the National Cancer Control Programme.

Delivery of hereditary cancer services will be organised across three 'tiers':

- Generalist level
- Regional Cancer Predisposition Services
- Specialist Cancer Genetics Services

This model of care outlines roles of each tier and the working relationships to be established between the 'tiers', e.g. the way in which the Specialist Cancer Genetics Service will support and enable delivery of care by Regional Cancer Predisposition Services. It is essential that patient care across the tiers is seamless, with clear communication and coordination of care.



Generalist Services

Healthcare services (including primary care and non-oncology services in secondary care) should be empowered to recognise those with a possible inherited predisposition to cancer and refer appropriately and to understand the management of those with identified cancer predisposition.

Referral pathways and protocols will be agreed nationally, and specific education initiatives should focus on those working in generalist services, to ensure appropriate knowledge of referral pathways/protocols and identification of patients for referral. The need for improved genetic literacy across all healthcare professionals is recognised in the *National Strategy for Accelerating Genetic and Genomic Medicine in Ireland*[1].

Regional Cancer Predisposition Services

Regional services have a role in the identification and assessment of those with a possible inherited predisposition to cancer, including mainstreamed diagnostic testing for inherited pathogenic gene variants in patients with cancer, according to national protocols.

Regional services also deliver the necessary healthcare to those identified with an inherited cancer predisposition, to determine role for targeted cancer therapy where relevant, to reduce cancer risk and/or facilitate early detection and to maximise patient health and wellbeing.

Multiple services including medical oncology, surgical oncology, haematology and benign services such as gastroenterology are involved in the identification and management of cancer predisposition.

The required multi-disciplinary service for people with the more common cancer predisposition syndromes (e.g. *BRCA* carriers, Lynch syndrome) should be coordinated regionally at cancer centre level and requires a structure to facilitate follow-up of patients. Certain rarer syndromes may follow a different model, with patient management and follow-up delivered in one centre, in close collaboration with the Specialist Cancer Genetics Service.

Regional Cancer Predisposition Services require designated input from Specialist Cancer Genetics Services, with an embedded genetic counsellor resource and within a clear clinical governance structure.

Regional Cancer Predisposition Services - Workforce

Clinical Lead Role

A **Clinical Lead for Cancer Predisposition** should be appointed with an identified sessional commitment to lead the development of each Regional Cancer Predisposition Service. This consultant role will include leading on:

- development of regional service (tailored to population size and specialisms)
- adherence to national protocols
- development of local pathways
- ensuring appropriate education and training of local workforce. All cancer services should be upskilled to ensure the provision of a high-quality service capable of accurately assessing family history risk (in patients affected/ unaffected by cancer) and to effectively implement mainstreamed testing pathways (related to drug treatment or other indications for treatment).
- clinical audit & service improvement
- service monitoring, including measuring and reporting KPIs

Requirement: *Each regional service will require an identified consultant clinical lead for this role, e.g. with a sessional commitment equivalent to 0.4WTE. A lesser time commitment may be feasible in smaller centres, or if sharing this role across two sites within a network/ future Regional Health Area.*

Administrative support

Appropriate administrative support will be integral to service delivery, monitoring and evaluation. These roles should include data management and clinic coordination. Genetics and genomics has a higher requirement for record retrieval and documentation than other specialties, due e.g. to the detailed information provided by patients on their family history, or the tracking of multiple pathology test results. Genomics Resource Associates will play an essential non-clinical support role in this area, including support for patients in navigating the assessment and testing process.

Administrative support in this area will allow skilled genetic counsellors to work at the top of their scope and allow more timely access to genetic counselling for patients. Technological solutions may alleviate some of the administrative burden associated with service delivery.

Genetic Counsellors

The role of **Genetic Counsellor** is a Specialist Cancer Genetics role, but will be delivered regionally. It will be possible for a Genetic Counsellor to be either appointed and employed locally by the Regional Cancer Predisposition Service, or centrally by the Specialist Cancer Genetics Service. Where appointed regionally, a professional reporting relationship to and sessional commitment to Specialist Cancer Genetics will be required.

A Genetic Counsellor will fulfil the following roles within each of the Regional Cancer Predisposition Services:

- act as point of contact for support for non-geneticists who are providing pre-test counselling
- develop and deliver educational initiatives to the wider workforce
- provide comprehensive pre or post-test genetic counselling as required, including reproductive information, on site or virtually
- act as point of contact/link to the Specialist Cancer Genetics tertiary service for complex cases, including children requiring cascade testing and consultation on variants of uncertain significance
- contribute to research and training

Requirement: *Each of the Regional Cancer Predisposition Services requires an identified genetic counsellor, appointed either locally or centrally, with a professional reporting relationship to a national lead cancer genetic counsellor in the Specialist Cancer Genetics Service.*

A cancer genetics counsellor acts as part of the specialist genetics resource embedded in the regional cancer predisposition service. Their role should include a sessional commitment to the national network, including national MDTs.

Specialty Lead

Within specific specialties (e.g. medical oncology, haemato-oncology, colorectal, gynaecology, breast), a **lead consultant** should lead on implementing Regional Cancer Predisposition Services, including ensuring access to mainstreamed testing where applicable and ongoing management of those with an identified cancer predisposition.

The exact configuration of specialties and their role in delivery of Regional Cancer Predisposition Services will vary according to services available in the region and may change over time – e.g. specific surgical oncology, medical oncology, haematology services. Collaborative working will be key, as patient needs frequently cross clinical specialties. Pathways for the management of those with an identified cancer predisposition may also need to incorporate access to certain services from outside the region.

Nursing

Clinical Nurse Specialists (CNSs) have considerable input in managing patient care within cancer predisposition services. They are expert practitioners who have attained, at a minimum, a post graduate qualification in their specialist area of practice. They coordinate patient care from the time of referral to their service. The CNS has the primary responsibility for educating and supporting the patient/carer in this service.

The Advanced Nurse Practitioner (ANP) role in cancer predisposition services provides autonomous caseload management with high levels of clinical decision-making. While working within the multidisciplinary team and having attained a masters qualification, they have a greater level of autonomy and can complete a full episode of care. The ANP plans and initiates care and treatment to achieve patient-centred outcomes and evaluates their effectiveness, initiating and completing a care episode. The ANP can initiate and implement innovation and change in their healthcare service in response to patient/client need and service demand.

CNS and ANP members of teams are key to the delivery of the Regional Cancer Predisposition Services, with roles including assessment of family history, patient pre-test counselling and provision of continuity of care and coordinated care for those identified with a cancer predisposition. Administrative support in this area will allow skilled nursing staff to work at the top of their licence and will support the coordination of care across different clinical areas.

<table>
<tr><td colspan="7"><u>Sample</u> staff in Regional Cancer Predisposition Service</td></tr>
<tr><td colspan="7">Clinical Lead for Cancer Predisposition Services</td></tr>
<tr><td>Medical oncology</td><td>Haemato-oncology</td><td>Colorectal service</td><td>Gynaecology</td><td>Breast service</td><td rowspan="2">Admin staff</td><td rowspan="2">Genetic Counsellor*</td></tr>
<tr><td>Consultant</td><td>Consultant</td><td>Consultant</td><td>Consultant</td><td>Consultant</td></tr>
<tr><td>CNS</td><td>CNS</td><td>CNS</td><td>CNS</td><td>CNS</td><td colspan="2" rowspan="2">Genomic Resource Associate</td></tr>
<tr><td>ANP</td><td>ANP</td><td>ANP</td><td>ANP</td><td>ANP</td></tr>
<tr><td colspan="7">Pathology, laboratory, radiology, endoscopy, psychooncology services</td></tr>
</table>

**may be employed by cancer centre or Specialist Cancer Genetics Service but professional reporting relationship to specialist cancer genetics*

Specialist Cancer Genetics Service

The Specialist Cancer Genetics Service will function as the national centre for clinical expertise in Ireland and will provide support and advice to Regional Cancer Predisposition Service sites via a network of designated genetic counsellors, a tertiary referral service and national MDT(s).

The role of the Specialist Cancer Genetics Service will include:

- Develop national guidance, including protocols for mainstreamed services as appropriate. All standards, protocols and guidelines will be agreed nationally, and founded on the best available evidence and best international practice, appropriately applied in the Irish context.
- Horizon scanning - as cancer genetics and genomic services and techniques continue to advance, it is important to ensure that the introduction of new tests

and phasing out of others is informed by specific guidelines, and that this will be audited.

- Develop and deliver educational initiatives to the wider workforce. Guidance is specifically required to support the implementation of mainstreamed pathways.
- Support non-geneticists who are providing pre-test counselling.
- Provide a regional cascade testing service via the networked genetic counsellor resource.
- Provide comprehensive pre or post-test genetic counselling as required, including reproductive information, on site or virtually.
- Provide a tertiary referral service for complex cases and consultation on variants of uncertain significance (VUS) – referral pathways will be agreed to facilitate referral of patients to the Specialist Cancer Genetics Service by a clinical team based at the Regional Cancer Predisposition Services.
- Support in the management of complex cases may be provided remotely or in person at a tertiary referral centre (+/- discussion at a national MDT).

Adult and paediatric patients have differing needs and should be assessed and managed in an appropriate environment (see below).

Specialist Cancer Genetics Services - Workforce

Consultants

Consultant expertise in specialist cancer genetics is provided by (i) those trained as Consultants in Clinical Genetics and (ii) those trained as oncology consultants (medical, surgical, radiation or haemato-oncology), who have developed expertise in the area of cancer genetics, specifically cancer predisposition. Those trained in oncology have a greater understanding of the overlap with current or future cancer treatments, whereas clinical geneticists have a broader role in areas such as syndrome overlap with non-cancer areas. Both areas of expertise are valued and should be maintained.

Of note, the oncology consultant role referred to here is different to that defined within Regional Cancer Predisposition Services. The lead role regionally may be held by someone with an interest in service development in this area but would not require the clinical expertise to provide a tertiary referral Specialist Cancer Genetics Service. However, regional expertise can be expected to develop further over time, particularly in the area of more common hereditary cancer syndromes.

Genetic Counsellors

As previously outlined, some genetic counsellors will provide a dedicated service to Regional Cancer Predisposition Services but will maintain both a sessional commitment and a professional reporting relationship to the Specialist Cancer Genetics Service.

Genetic Counsellors will also operate as part of the tertiary referral service in cancer specialist genetics, will have a role in education, training and guidance development and provide a clinical governance structure for genetic counsellors working regionally.

Challenges in Ireland have been noted in relation to the lack of career pathway, HSE grade codes, Health & Social Care Professionals Council (CORU) recognition, and registration for genetic counsellors. This has been highlighted as a priority issue for the national genetics and genomics office.

Administrative and IT support

As outlined for regional services, the administrative workload for genetics is high compared to other specialties. The team will also benefit from Genomic Resources Associates and support to ensure coordination of national MDT(s) and the genetic counsellor network.

Laboratory expertise

Molecular diagnostic expertise is required as part of the specialist cancer genetics MDT, particularly in relation to the interpretation of Variants of Uncertain Significance. Of note, the broader laboratory infrastructure and clinical and scientific expertise required for germline genetic testing is outside the scope of this document and will be considered elsewhere.

Paediatric and AYA Cancer Predisposition Service

Childhood, adolescent and young adult (CAYA) cancers are rare and complex disorders, with differing cancer types and different disease biologies to those seen in adults. In addition, these cancers are associated with a higher rate of cancer predisposition, with less influence of environmental factors. Currently, it is estimated that up to 30% of CAYAs with cancer require genetic evaluation, with at least 10% of CAYAs with cancer having a confirmed cancer predisposition syndrome.

A comprehensive service is therefore required that incorporates cancer predisposition risk assessment for all CAYA patients. In addition, as the standard of care is moving internationally towards somatic testing of all tumours diagnosed in the CAYA population, germline testing for all will likely be required, either in sequence or parallel, necessitating expertise in relation to interpretation and management.

Children's cancer services are already centralised at the National Children's Cancer Service (NCCS), based at Children's Health Ireland at Crumlin. The development of a formal national network for AYA services is currently underway involving CHI, St James's Hospital, Cork University Hospital and University Hospital Galway (Framework for the Care and Support of Adolescent and Young Adults (AYA) with cancer in Ireland[2]). The provision of a single national specialist paediatric MDT for cancer genetics fits with this model and such a service has already commenced at CHI at Crumlin. The multidisciplinary input required crosses specialist genetics, oncology/ haematology, molecular pathology and the wider CAYA cancer team. Collaboration will be required between this MDT and that at the AYA service in St James' Hospital, to meet the needs of this age group.

In addition to providing access from regional paediatric services to/from this specialist service, consideration should also be given to cross over with Regional Cancer Predisposition Services who are dealing with adults. For example, it may be appropriate for genetic testing to be carried out in children of adults diagnosed with certain cancer predisposition syndromes. These children should be referred to specialist genetics services associated with paediatric cancer predisposition services, for age-appropriate genetic counselling and management. The benefit of consultant clinical geneticists and genetic counsellors is that they are skilled to deal with all ages and all types of clinical presentations (syndromic/ non-syndromic), cancer risks and risks of other medical conditions, across all age spectrums.

Similarly, when cancer predisposition is identified in a child, testing of adults may identify those with the same predisposition and who will benefit from ongoing management in their regional cancer predisposition service.

Challenges specific to the Children, Adolescent and Young Adult population

Challenges relating to cancer predisposition in the CAYA population will vary according to the age of the child or young person, with some diagnoses of cancer predisposition as early as ante-natally or even pre-natally. Reproductive advice and referral to services is required for many parents of children with identified cancer predisposition, as the parents are themselves still of reproductive age. For children themselves, it is important that this information regarding reproduction and fertility is raised at an age-appropriate time. This can be incorporated into genetics review during the AYA transition period. Other issues for consideration at this time include any need

for further genetic testing, any changes in surveillance recommendations, and ownership of the diagnosis going forward.

Specific challenges can also arise in relation to disease surveillance in this population, particularly younger children. These include greater reliance on ultrasound and MRI techniques to minimise exposure to radiation, the need for hospital day ward admission for young patients in order to access surveillance imaging procedures under general anaesthesia with appropriate medical and nursing supports needed to provide this level of care, and frequent clinical review and examination by experienced health professionals.

Implementation

The implementation of this Model of Care will be a multi-year project. An implementation group will be established to oversee this work, encompassing the following areas:

Thematic Area 1: **Establish oversight for the implementation of the Hereditary Cancer Model of Care**
1.1 Establish a governance and reporting structure aligned to the National Cancer Strategy and National Cancer Control Programme
1.2 Agree areas for collaboration with the National Genetics and Genomics Office once in place
1.3 Convene a multi-disciplinary implementation steering group, with clinical leadership and patient involvement
1.4 Establish relevant working groups for thematic priorities
1.5 Agree an overarching governance framework for hereditary cancer service
1.6 Appoint Clinical Leads for Cancer Predisposition within regional services
1.7 Report at minimum annually on progress in implementation
Thematic Area 2 : **Development of networked services and MDTs**
2.1 Define the process(es) by which genetic counselling services can be networked and made accessible regionally
2.2 Develop a standardised pathway to a national specialist cancer genetics MDT (adults)
2.3 Develop a standardised pathway to a national specialist cancer genetics MDT (CAYA)
2.4 Establish a network of regional cancer predisposition leads
Thematic Area 3 : **Healthcare professional education and training; workforce development**
3.1 Identify the training needs of staff working in generalist settings
3.2 Identify the training needs of staff working in Regional Cancer Predisposition Services
3.3 Develop an accredited online programme to meet the minimal learning needs of staff working in regional services
3.4 Identify or develop further training courses to meet identified needs of staff

3.5 Work with the National Genetics and Genomics Office, professional training bodies and regulators to develop career pathways in specialist cancer genetics
Thematic Area 4: **Clinical guidance and pathway development**
4.1 Agree the guidance development and approval process, in line with HSE policy, including guidance on laboratory testing
4.2 Establish guidance development groups for common presentations and cancer predispositions
4.3 Ensure patients are involved in the co-design of clinical pathways
4.4 Agree the process by which international guidance relating to rarer conditions will be approved and disseminated for use in Ireland
4.5 Agree the platform(s) through which current guidance will be made available to all
4.6 Highlight critical points in clinical pathways, which should be subject to regular audit
4.7 Describe resource requirements for implementation of pathways
Thematic Area 5: **Patient information and supports**
5.1 Establish subgroups for development of patient information, according to common presentations and cancer predispositions
5.2 Ensure patients are engaged in the co-design of patient information materials
5.3 Consider how best to meet the information needs of underserved populations and those with health literacy challenges
5.4 Agree or develop platform(s) through which patient information can be most easily accessed
5.5 Ensure evaluation of information materials and supports provided
Thematic Area 6: **Data requirements and IT infrastructure**
6.1 Explore the role of virtual patient assessment within hereditary cancer services
6.2 Identify the IT infrastructure required to enable the implementation of this model of care, including a genetic counselling network and referral to a national specialist genetics MDT
6.3 Examine potential data capture options with the National Cancer Information System
6.4 Engage with the National Genetics and Genomics Office in relation to development of registries for cancer predisposition
6.5 Agree minimum datasets for common presentations and cancer predispositions
6.6 Agree key performance indicators for the delivery of specialist cancer genetics and Regional Cancer Predisposition Services

1. Introduction

Cancer is a leading cause of morbidity and mortality in Ireland, giving rise to approximately 43,600 new cases and 9,400 deaths annually (NCRI). Cancer is caused by changes in the genes that regulate how cells grow and multiply. These genetic changes can result from:

- random errors that occur in DNA as cells multiply
- damage to cellular DNA arising from exposure to carcinogens (e.g. tobacco smoke, UV rays from the sun, and the human papillomavirus (HPV))
- the presence of a pathogenic gene variant, which confers an increased predisposition to cancer.

The majority of cancer cases (approximately 90%) are 'sporadic' and arise as a result of an accumulation of damage to DNA, resulting from the first two processes outlined above – random errors that occur in DNA as cells multiply, and/or DNA damage arising from exposure to carcinogens.

Approximately 5-10% of cancer cases are associated with the presence of a germline pathogenic gene variant. These are genetic variants that were present in the egg or sperm cell at the time of fertilisation and can be passed down from parents to offspring. A pathogenic gene variant is one that is known to cause disease, such as conferring an increased risk of developing one or more types of cancer.

Some pathogenic gene variants increase risk of more than one cancer type. These gene variants can cause hereditary cancer 'syndromes' – i.e. collections of signs and symptoms with a common cause. The two most common hereditary cancer syndromes are:

- Hereditary Breast and Ovarian Cancer (HBOC) Syndrome - caused by a pathogenic *BRCA1* or *BRCA2* gene variant and characterised by increased risk for female and male breast cancer, ovarian cancer, prostate cancer, pancreatic cancer, melanoma and other cancers.
- Lynch syndrome is caused by a pathogenic gene variant of one of five genes – *MLH1, MSH2, MSH6, PMS2 and EPCAM*. Lynch syndrome is characterised by an increased risk for colorectal, endometrial and other cancers.

The likely cancer risk for people with hereditary cancer syndromes varies by the gene variant (e.g. people with a *BRCA1* variant have a different risk profile to those with a *BRCA2* variant). It is important for patients and clinicians to know which variant an individual has in order to understand the associated risks and plan appropriate management.

1.1 Indicators of possible cancer predisposition

The first step in harnessing the power of genetics in cancer risk reduction is to identify people who are at increased risk of having an inherited predisposition to cancer, and who should therefore be considered for genetic testing. For example, a suspicion of inherited cancer predisposition may be raised by a family history of cancers occurring at a young age, unusual types of cancer (e.g. male breast cancer), or patterns of cancers that suggest a hereditary cancer syndrome. Other clinical features or laboratory test results may indicate a likelihood, as outlined in Figure 1 below.

Figure 1: **Examples of clinical scenarios relating to hereditary cancer and the identification of a possible inherited cancer predisposition through genetic testing**

- **Individuals with a cancer diagnosis**
  - ➢ Genetic testing related to suitability for drug treatments
  - ➢ Features suggesting inherited risk, e.g. tumour pathology, many paediatric cancers, tumour genetic profiling carried out (e.g. in research context or prognostic)
  - ➢ Family history which suggests an inherited risk
- **Individuals without a cancer diagnosis/personal history of cancer**
  - ➢ Family history which suggests an inherited risk
  - ➢ Relative of a known carrier, i.e. cascade testing
  - ➢ Clinical syndrome or phenotype associated with a high risk of cancer (examples in both paediatrics and adult presentation)

1.2. Genetics in cancer care and risk reduction

Genetics is now recognised as a growing and integral part of cancer care, with a role across treatment, early detection and risk reduction. Identification of cancer predisposition will only lead to patient benefit, if necessary services are in place and appropriately resourced and monitored. The potential benefit to a patient - and their relatives - can include reducing the risk of cancer developing, identifying cancer at an earlier more treatable stage, informing the optimal choice of targeted therapy if a cancer develops and facilitating reproductive options for the individual.

- ❖ **Risk reduction** – Genetics offers a direct and cost-effective way of identifying people who are at high risk of developing cancer. These people can then be managed appropriately, including offering interventions to reduce the risk of cancer developing, e.g. risk-reducing surgery and chemoprevention.
- ❖ **Early detection** – People who are identified as having an inherited cancer predisposition can be closely monitored to facilitate early detection and treatment of disease. Diagnosing cancer early typically allows for less complex treatment and optimal outcomes.
- ❖ **Treatment** - Certain cancer therapies rely on genetic and genomic information to tailor treatment to give the best chance of cure. For cancer patients with an inherited cancer predisposition, treatment decision-making based on timely genetic testing is rapidly becoming an international standard of care. Genetic test results guide surgical decisions and direct choice of medication. Targeting medication according to genetic test results is at the forefront of precision medicine.
- ❖ **Reproductive counselling and care** – Additional benefits of cancer genetics include the ability to identify people with a cancer predisposition and offer them appropriate counselling and care in relation to their reproductive health and options.
- ❖ **Benefit to relatives** – Cascade testing of unaffected relatives identifies more individuals with an inherited cancer predisposition, who can then benefit from the above cancer risk reduction and early detection measures.

The identification of cancer predisposition allows for the implementation of these risk-reduction measures, such as prophylactic surgery and chemoprevention, and surveillance to ensure early detection. In this way, genetic testing - for individuals and subsequent cascade testing of relatives - and provision of comprehensive downstream services can effectively reduce both cancer incidence and cancer mortality.

2. Context

2.1 Strategic context

2.1.1 National Cancer Strategy 2017-2026

The National Cancer Strategy 2017-2026[3] highlighted the need for further development of hereditary cancer services in Ireland, with the aim of providing equitable access to services for all patients on a national basis. The strategy acknowledged the need for a strategic approach, to include increased infrastructural and financial support.

Relevant recommendations of the strategy include recommendations 6, 19 and 20:

Recommendation 6 - The NCCP will draw up a plan by end-2017 for the development of an integrated cancer control and surveillance service for defined population subgroups with an inherited familial predisposition to cancer (e.g. breast, ovarian and colorectal).

Recommendation 19 - 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.

Recommendation 20 - The HSE will ensure that the existing cancer genetics services are amalgamated into one National Cancer Genetics Service and will identify the most appropriate site for its location.

Priorities highlighted in the strategy include cancer control and surveillance services for those with an inherited familial predisposition to breast, ovarian and colorectal cancer.

2.1.2 National Strategy for Accelerating Genetic and Genomic Medicine in Ireland

Development of genetics and genomics services in Ireland, to benefit all relevant clinical disciplines, is a current priority. The *National Strategy for Accelerating Genetic and Genomic Medicine in Ireland*[1] was published in December 2022.

The strategy addresses a number of core issues common to specialist genetics across multiple specialty areas. These include workforce challenges, in particular the need for a training and registration pathway for genetic counsellors in Ireland and recruitment and retention challenges across the services as a whole.

The Specialist Cancer Genetics Service represents a core component of overall clinical genetics services in Ireland. The planned National Office for Genetics and Genomics will provide governance for the Specialist Cancer Genetics Service in collaboration with the NCCP.

2.2 Current hereditary cancer services in Ireland

At present, hereditary cancer services in Ireland are underdeveloped and under-resourced to meet the rapid growth in service demand[3]. This is not unique to cancer, hence the need identified for a national genetic and genomic strategy for Ireland, to drive the necessary investment and development in genetics, to support a broad range of specialties.

Currently, specialist genetics services for hereditary cancer are provided in St James's Hospital (SJH) Cancer Genetics Service (adults only) and at the Department of Clinical Genetics, Children's Health Ireland (CHI) at Crumlin (adult and paediatric). Referrals are received from both primary and secondary

care. Services engage with patients via a range of in-person, virtual and telephone clinics and postal methods.

Services provided include:

- Genetic/family risk assessment, education and genetic counselling
- Germline testing
- Interpretation of results, including variants of uncertain significance
- Recommendations in relation to appropriate follow-up and ongoing management
- Cascade germline testing for relatives
- Reproductive options and information

Patients are typically discharged to the referring clinician, with follow-up recommendations. Given the absence of a structured system of referral for ongoing care, patients may be directed towards a known service in their geographical area or this may be left to the referring clinician to coordinate.

A number of established services exist, with considerable expertise in relation to certain cancer predisposition syndromes, e.g. specifically in relation to breast cancer predisposition, or colorectal cancer predisposition. However, these currently operate independently of each other and without an agreed pathway to/from specialist cancer genetics.

2.3 Defining the requirements for hereditary cancer services in Ireland

Deficiencies and challenges exist across all elements of the hereditary cancer services in Ireland. These challenges are defined in the National Cancer Strategy 2017-2026[3], and in various other reports and assessments, including an environmental scan of the cancer genetics services published by the Irish Cancer Society in April 2021[4], and a health needs assessment for people with cancer predisposing *BRCA* gene variants, carried out by the National Cancer Control Programme in 2022[5].

2.3.1 Health needs assessment for persons diagnosed with a cancer-predisposing variant of BRCA1 and BRCA2 in Ireland

A health needs assessment published by the NCCP in 2022 highlighted the improvements required to ensure a high quality, comprehensive service for carriers of pathogenic *BRCA* gene variants. The needs assessment highlighted deficiencies across eight key themes, and made associated recommendations for service improvement (Appendix C). Many of the issues raised are challenges common to other inherited cancer predisposition syndromes.

- The need for accessible, inclusive and jargon-free information resources for carriers and information resources for health care professionals working in areas other than specialist cancer genetics.
- A system required to gather data to support national planning and co-ordination of services for the affected population and facilitate local follow-up of patients.
- Need for improved access to suitably qualified specialist genetics healthcare professionals after diagnosis, to include family planning supports and specialist fertility services.
- A structured care pathway is required to ensure consistency in and access to optimal standards of care in all geographic areas and improved co-ordination of care.
- Adequate access to risk-reducing measures, such as prophylactic surgery and surveillance, require dedicated resources, protected from the demands of the symptomatic service.
- The need for access to psychological support throughout the patient journey.
- Specialist services relating to the adverse effects of risk-reducing measures, in this case the management of surgical menopause.

2.3.2 The unmet need in cancer genetic services: conducting an environmental scan of the cancer genetics service in an Irish context underpinned by a mixed methods approach - Report prepared for the Irish Cancer Society[4]

This report provided an overview of current cancer genetic services in Ireland, highlighted deficiencies and made a suite of recommendations to optimise services (direct transcript in Table 1).

Table 1: Recommendations of the Irish Cancer Society report[4]

Number	Recommendation
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 pathogenic variants 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.

2.4 Summary - challenges facing hereditary cancer services in Ireland

As highlighted through these various needs assessments and reports, the challenges facing hereditary cancer services in Ireland include, but are not limited to, the following:

- There is no nationally agreed model of care for patients with inherited cancer predispositions.
- There are no nationally agreed guidelines for genetic assessment, testing or management in Ireland (a number of international guidance documents are followed, e.g. European Refence Network (ERN) Genturis, NICE Familial Breast Cancer guidelines[6] and UK Cancer Genetics Group (UKCGG) guidelines).
- Existing hereditary cancer services are underdeveloped and under-resourced. There is a lack of capacity to deliver care across all elements of the pathway - including genetic assessment, testing and counselling, but also for timely ongoing clinical follow up for cancer predisposition and access to surveillance imaging/ endoscopy, risk reducing surgery, related health needs (e.g. fertility/reproductive care) and follow-up care.
- Waiting lists for specialist cancer genetic services 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.
- There is a lack of sufficient integration and coordination of care between existing specialist cancer genetic services, regional cancer services and primary care.
- There is inequitable access to existing services.
- Many healthcare professionals lack sufficient training/genetic literacy to advise and inform service users in relation to hereditary cancer services.
- There is a lack of appropriate data sources and IT infrastructure to support the planning, delivery, monitoring and improvement of hereditary cancer services, including the absence of a registry of patients with inherited cancer predispositions.

2.5 Optimising hereditary cancer services in Ireland

2.5.1 Hereditary cancer services model of care

This model of care defines the vision for an optimal Hereditary Cancer Service for the population of Ireland, guided by the needs identified above.

Implementation of the model will be a complex, multi-year project.

A core concept which will be included in the proposed framework is that of 'mainstreaming' genetics and how this can be implemented in a safe and efficient manner in cancer services in Ireland.

2.5.2 The concept of mainstreaming genetics

Mainstreaming in cancer genetics refers to a model whereby the provision of cancer genetic testing for those with a personal cancer diagnosis is integrated into oncology services. This approach is also being adopted across many other non-cancer specialties, as a means of increasing access to genetic testing for patients.

Mainstreamed pathways enable access to diagnostic genetic testing for patients with cancer as part of the standardised management of cancer care, in line with nationally agreed criteria and protocols.

A member of the team treating the patient, e.g. consultant or clinical nurse specialist, carries

out the pre-test counselling, in accordance with agreed protocols and supported by necessary training. The identification of a pathogenic gene variant then triggers referral to a genetic counsellor for post-test counselling and further information in relation to cascade testing of family members.

Referral onward to specialist genetics is also required when a variant of uncertain significance (VUS) is identified, or where no variant is identified but a significant family history warrants genetics input.

A number of healthcare care institutions internationally have implemented such a model, likely in response to such factors as:

- Increased clinical utility of genetic testing, including in selection of appropriate drug treatment, necessitating quicker access to testing and results
- Improving genetic literacy among both the public and non-genetics healthcare professionals
- A recognition of the need to increase identification of those with inherited cancer predisposition where action can be taken to reduce cancer risk
- A recognition worldwide of the limited specialist genetics workforce capacity to meet this growing demand.

The literature on this area (see Appendix A for bibliography) focusses most commonly on mainstreaming of *BRCA* testing for those with a diagnosis of ovarian cancer or breast cancer. The Royal Marsden Hospital is widely regarded as a leader in this area, with many centres, particularly other centres in the UK, using the Royal Marsden materials to train staff as part of implementing mainstreaming in their services.

Overall, patient experience and feedback on pre-test counselling and testing within their oncology appointments has been positive. Where compared to traditional clinical genetic service testing models, generally the mainstreamed approach did not impact on satisfaction, knowledge and/or distress.

Importantly, several studies report substantially reduced waiting times for genetic testing following implementation of a mainstreamed model, and a small number of studies reported cost-savings within their service.

One Australian study reported on the integration of a genetic counsellor into oncology clinics; however, this approach would be limited by existing staff resources in a given healthcare system and is much less feasible compared to the mainstreamed approach using oncology staff.

Overall, the majority of oncologists and clinical nurse specialists studied indicated positive feedback and acceptance of the mainstreamed approach. One UK study reported that most of the breast surgeons surveyed were not in favour of the mainstreamed approach due to existing constraints on their time and perceived lack of expertise, preferring to maintain their role in triaging patients by referral to clinical genetics.

Genetics professionals studied were overall supportive, although in one study less than 50% of the geneticists or genetic counsellors surveyed responded that patients seemed to receive accurate information about the *BRCA* test in the pre-test counselling session.

These findings highlight the potential of mainstreaming to improve patient care, if planned for and supported appropriately. Health system interventions shown to assist in overcoming mainstreaming implementation challenges include integration of genetic testing, follow up and tracking into an electronic patient record; genetic counsellors as a point of contact for services or embedded into oncology services; champions in cancer centres; ongoing education and training and a standardised care pathway. Education and training of healthcare professionals involved in mainstreamed pathways appears to have positive impacts on confidence and knowledge of healthcare professionals to engage in pre-genetic test discussions with patients and

address questions. Communication between non-genetics and genetics services in mainstreamed pathways has been highlighted as a priority, both in the pre-implementation and operational phases of these services. It is therefore key that a move towards a mainstreamed model for cancer genetics in Ireland would happen in conjunction with further development of the Specialist Cancer Genetics Services, to ensure a safe and supported mainstreamed service, in addition to the ongoing specialist cancer genetics role.

3. Vision for the model of care

The vision of this model of care is that it will provide clarity regarding the structure and governance of hereditary cancer services in Ireland. The model of care will ensure/ support:

- Development of national guidance to underpin standardised best practice, including mainstreaming of cancer genetic testing
- Equitable access to hereditary cancer services for all
- Clear governance and clarity regarding the roles of national genetics and genomic structures, Specialist Cancer Genetics Services and Regional Cancer Predisposition Services, within an overarching model of care
- Commitment to the Sláintecare principles of right person, right place, right time – through the integration of genetic assessment and testing into regional services, with access to specialist genetic expertise as required
- Maximal use of technological solutions
- Timely access to genetics assessment and testing, including specialist cancer genetics expertise as required
- Timely access to clinical management of cancer predisposition, including risk reduction options
- Comprehensive and coordinated approach to ongoing management
- Quality assurance of service
- Access to high quality patient information, psychological support and peer support for patients
- Education and training of health care professionals, including accredited training for those delivering mainstreamed pathways

3.1 Evidence-based national guidance

Standards, protocols and guidelines should be agreed nationally, in accordance with best available evidence, best international practice, and as applied to the Irish context. Such guidance should be agreed under the governance of the NCCP, in collaboration with the national genetics and genomics office.

The full patient pathway should be addressed, from identification of those requiring family risk/ genetic assessment, use of assessment tools, criteria for genetic testing and types of testing employed (e.g. a national test directory similar to NHS) and recommendations on surveillance, follow up and risk reduction. Guidance should be easily available to clinicians, e.g. maintained online.

Guidance is specifically required to support the implementation of mainstreamed pathways. Mainstreamed pathways enable access to diagnostic genetic testing for patients with cancer as part of the standardised management of cancer care, in line with nationally agreed criteria and protocols.

3.2 Equity of access

The service should ensure equitable access to optimal care for all, regardless of area of residence. This does not mean all aspects of care are best delivered locally. Certain services will be most appropriately delivered supraregionally/ nationally, for reasons of quality and efficiency, but equity of access must be assured for all.

3.3 Clarity of roles within an overarching model of care

A clear governance framework is required which will outline the role of the new national genetics and genomic programme in relation to the agreement of national standards, practices and protocols and management guidelines; education and training; specialised services e.g. in relation to family planning and how this will operate in partnership with the NCCP in relation to cancer services.

Clarity is also required in relation to role and governance of both specialist genetics services and Regional Cancer Predisposition Services.

3.4 Right person, right place, right time – assessment and testing

Cancer services should be upskilled to ensure ability to assess family history risk (in patients affected/ unaffected by cancer) and to implement mainstreamed testing pathways (related to drug treatment or other treatment indications).

For a person with a diagnosis of cancer, mainstreamed pathways ensure that genetic assessment (and testing where appropriate) is part of standard management. Age-appropriate post-test counselling and management recommendations for those with a pathogenic variant should then be provided in line with national guidelines and delivered with specialist cancer genetics input as required. Specialist input will also be required in relation to counselling and management recommendations for those with variants of uncertain significance (VUS).

Where testing is considered in an unaffected individual (e.g. relative as part of cascade testing), that is most appropriately managed by specialist cancer genetics, i.e. a trained genetic counsellor, but the patient can be seen regionally, for either an in-person or virtual appointment. As regional access to specialist cancer genetics input improves, collaborative approaches to cascade testing can be developed which utilise the experience of a skilled regional staff member. A GP referral should not be required for relatives to avail of cascade testing.

Specialist cancer genetics services are also required for support for complex cases, either for remote advice, as a tertiary referral point +/- discussion at a national MDT and for management of paediatric patients, including those identified for cascade testing.

3.5 Maximal use of technological solutions

The use of technology, such as virtual assessment and decision aid tools, should be employed appropriately to maximise efficiency and support use of protocols.

Consideration should be given to patient preference, particularly if there is potential for receiving an initial diagnosis virtually, while being mindful of the potential for issues with digital literacy, healthcare literacy, language barriers, education level and poverty. Virtual appointments can be easier and less costly for patients. The patient can still attend a more local hospital for the virtual appointment.

Group genetic counselling, including digitised formats such as website information, self-paced learning for informed consent, and genetic testing to improve access in pre-test context are also possible and could improve efficiency. Examples of such programs are the Jenescreen[7] and BRCA DIRECT in the UK[8].

3.6 Timely access to genetics assessment +/- testing and results

As highlighted above, employment of mainstreaming pathways as appropriate will improve timely access to genetic testing for those with a cancer diagnosis.

For all patients, there should be an agreed

timeframe for patients to be offered genetic testing and for a specialist cancer genetics consultation when required (before or after test result), to reduce anxiety and to allow for timely surveillance/ chemoprevention/ risk reducing surgery. These timeframes should be incorporated into national guidance and a monitoring framework put in place.

The turnaround time for testing should facilitate timely treatment decisions and risk reduction measures, and be acceptable to patients.

3.7 Timely access to risk-reduction options

A dedicated pathway with protected resources is required for management of those identified with a pathogenic variant/ inherited cancer predisposition – across prevention, surveillance, risk-reducing surgery, chemoprevention and wider supportive management (e.g. psychological support, management of adverse effects of risk reduction measures, reproduction and fertility). The most appropriate location for services will vary depending on the prevalence of the condition and the complexity of the intervention.

In particular, the appropriate configuration of services for surveillance and risk-reducing surgery should be examined, to identify those that would result in low wait times and the best clinical outcomes for patients. Consideration should also be given to the specialist pathology input required.

A sufficiently resourced system, supported by necessary IT platform(s), is required to follow patients and ensure interventions can be offered and delivered in a timely fashion at the appropriate age. Relevant timeframes should be agreed and incorporated into national guidance and a monitoring framework put in place.

3.8 Patient access to specialist genetics expertise

Those with an identified cancer predisposition/ pathogenic gene variant should have access to suitably qualified healthcare professionals after they receive their results, to ask any questions that arise after they have had a chance to process their diagnosis.

Some of these clinical queries could be addressed by designated genetics-trained healthcare professional(s) (e.g. Advanced Nurse Practitioners (ANPs)) in cancer centres and relate to more common hereditary cancer syndromes, while other queries, including reproductive counselling, would fall solely within the remit of specialist genetics services (e.g. Genetic Counsellors or genetic consultants). Paediatric patients will be managed by specialist genetics and the paediatric cancer predisposition service. AYA patients will have unique needs - specialist expertise for the AYA age group should also be available to those who were identified with a cancer predisposition as a child.

Individuals with a pathogenic gene variant should be provided with information regarding specialist advice on reproduction and fertility. Fertility services in this context should be made available in the public sector and in a timely fashion.

3.9 Comprehensive and coordinated approach to ongoing management

Improved coordination of care is required, including between Specialist Cancer Genetics Services and regional services/cancer centres. This can be supported via the agreement of a national model of care, including patient pathways.

Comprehensive services require access to wider supportive management (e.g. psychological support, management of adverse effects of risk reduction measures, reproduction and fertility, health and wellbeing supports, including lifestyle behaviours), in addition to risk reduction measures.

Ongoing clinical care of those with an identified

inherited cancer predisposition could be delivered by a nurse-led service, with the CNS/ANP also acting as the dedicated point of contact. There is benefit to the autonomous clinical role of an ANP in this context.

The coordinated approach to management at regional level, including adherence to protocols and regular audit, should sit within an overarching framework for governance of clinical care of hereditary cancer services.

3.10 Quality assurance of service

All elements of the patient care pathway (structures, processes and outcomes) should have defined quality standards and a subset of specific key performance indicators to facilitate performance measurement.

Patient or family-reported experience measures should be an embedded quality indicator.

3.11 Access to high quality patient information

Accessible, inclusive (across age, language and ethnicity) and jargon-free informational resources should be developed for and available as part of routine care, to those undergoing genetic testing and those identified as carriers.

Any such resources should be co-designed with patient stakeholders.

3.12 Psychological support and peer support

Psychological support including professional support should be offered (but not mandatory) to all carriers following a diagnosis of a pathogenic variant. This should be available if needed throughout the patient journey, including but not only at times such as undergoing risk-reducing surgery and in the event of a cancer diagnosis.

Education initiatives should meet the education and training needs of the psychology profession specific to inherited cancer predisposition.

A pathway should be established for carriers to access psychological supports (both professional and peer-based) as required, including those external to cancer centres.

3.13 Education and training of health care professionals

Development of information resources specifically for healthcare professionals working in primary care and cancer services in Ireland is required. This should be informed by consultation with clinical subject matter experts.

Education of the cancer services workforce is required, to support implementation of guidelines, pathways and protocols, including accredited training for those delivering mainstreamed pathways. In addition to knowledge of guidance and pathways, healthcare professionals should be trained in the discussion of genetic risk with patients and in the communication of test results.

4. Service delivery/structure

4.1 Three tier model of service delivery

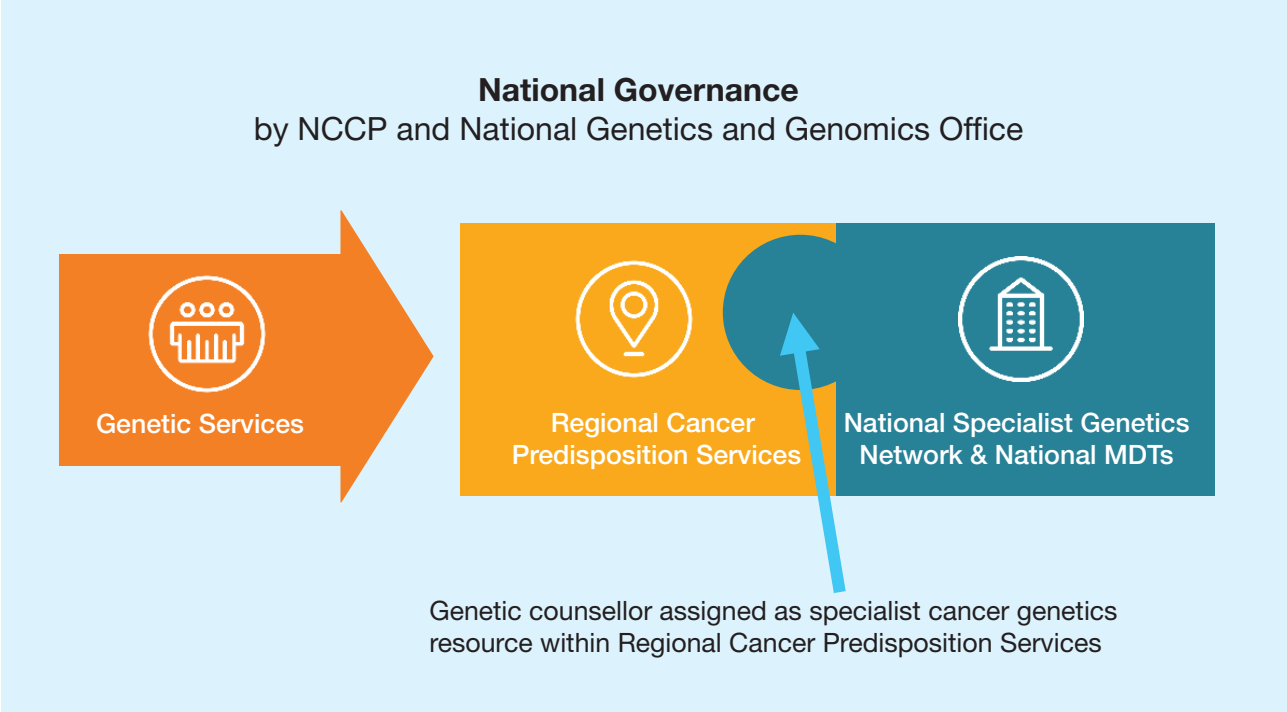
A programmatic approach is required to ensure national agreement of guidance, including mainstreaming protocols, laboratory and clinical data infrastructure, specialist genetics workforce development, and education of the wider workforce. This programme will be a collaboration between the National Genetics and Genomics Office and the National Cancer Control Programme.

Delivery of hereditary cancer services will be organised across three 'tiers':

- Generalist level
- Regional Cancer Predisposition Services
- Specialist Cancer Genetics Services

This model of care outlines roles of each tier and the working relationships to be established between the 'tiers', e.g. the way in which the Specialist Cancer Genetics Service will support and enable delivery of care by Regional Cancer Predisposition Services. It is essential that patient care across the tiers is seamless, with clear communication and coordination of care.

Figure 2: **Hereditary cancer services - model of service delivery**



4.2 Generalist Services

Healthcare services (including primary care and non-oncology services in secondary care) should be empowered to recognise those with a possible inherited predisposition to cancer and refer appropriately and to understand the management of those with identified cancer predisposition.

Referral pathways and protocols will be agreed nationally, and specific education initiatives should focus on those working in generalist services, to ensure appropriate knowledge of referral pathways/protocols and identification of patients for referral. The need for improved genetic literacy across all healthcare professionals is recognised in the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland.[1]

4.3 Regional Cancer Predisposition Services

Regional services have a role in the identification and assessment of those with a possible inherited predisposition to cancer, including **mainstreamed diagnostic testing** for inherited pathogenic gene variants in patients with cancer, according to national protocols.

Regional services also deliver the necessary healthcare to those identified with an inherited cancer predisposition, to determine the role for targeted cancer therapy where relevant, to reduce cancer risk and/or facilitate early detection and to maximise patient health and wellbeing.

Multiple services including medical oncology, surgical oncology, haematology and benign services such as gastroenterology are involved in the identification and management of cancer predisposition.

The required multi-disciplinary service for people with the more common cancer predisposition syndromes (e.g. *BRCA* carriers, Lynch syndrome) should be coordinated regionally at cancer centre level and requires a structure to facilitate follow-up of patients. Certain rarer syndromes may follow a different model, with patient management and follow-up delivered in one centre, in close collaboration with the Specialist Cancer Genetics Service.

Regional Cancer Predisposition Services require designated input from Specialist Cancer Genetics Services, with an embedded genetic counsellor resource and within a clear clinical governance structure.

4.3.1 Regional Cancer Predisposition Services - Workforce

Clinical Lead Role

A **Clinical Lead for Cancer Predisposition** should be appointed with an identified sessional commitment to lead the development of each Regional Cancer Predisposition Service. This consultant role will include leading on:

- development of regional service (tailored to population size and specialisms)
- adherence to national protocols
- development of local pathways
- ensuring appropriate education and training of local workforce. All cancer services should be upskilled to ensure the provision of a high-quality service capable of accurately assessing family history risk (in patients affected/ unaffected by cancer) and to effectively implement mainstreamed testing pathways (related to drug treatment or other indications for treatment).
- clinical audit & service improvement
- service monitoring, including measuring and reporting KPIs

Requirement: *Each regional service will require an identified consultant clinical lead for this role, e.g. with a sessional commitment equivalent to 0.4WTE. A lesser time commitment may be feasible in smaller centres, or if sharing this role across two sites within a network/ future Regional Health Area.*

Administrative support

Appropriate administrative support will be integral to service delivery, monitoring and evaluation. These roles should include data management and clinic coordination. Genetics and genomics has a higher requirement for record retrieval and documentation than other specialties, e.g. due to the detailed information provided by patients on their family history, or the tracking of multiple pathology test results. **Genomics Resource Associates** will play an essential non-clinical support role in this area, including support for patients in navigating the assessment and testing process.

Administrative support in this area will allow skilled genetic counsellors to work at the top of their scope and allow more timely access to genetic counselling for patients. Technological solutions may alleviate some of the administrative burden associated with service delivery.

Genetic Counsellors

The role of **Genetic Counsellor** is a Specialist Cancer Genetics role, but will be delivered regionally. It will be possible for a Genetic Counsellor to be either appointed and employed locally by the Regional Cancer Predisposition Service, or centrally by the Specialist Cancer Genetics Service. Where appointed regionally, a professional reporting relationship to and sessional commitment to Specialist Cancer Genetics will be required.

A Genetic Counsellor will fulfil the following roles within each of the Regional Cancer Predisposition Services:

- act as point of contact for support for non-geneticists who are providing pre-test counselling
- develop and deliver educational initiatives to the wider workforce
- provide comprehensive pre or post-test genetic counselling as required, including reproductive information, on site or virtually
- act as point of contact/link to the Specialist Cancer Genetics tertiary service for complex cases, including children requiring cascade testing and consultation on variants of uncertain significance
- contribute to research and training

Requirement: *Each of the Regional Cancer Predisposition Services requires an identified genetic counsellor, appointed either locally or centrally, with a professional reporting relationship to a national lead cancer genetic counsellor in the Specialist Cancer Genetics Service.*

A cancer genetics counsellor acts as part of the specialist genetics resource embedded in the regional cancer predisposition service. Their role should include a sessional commitment to the national network, including national MDTs.

Specialty Lead

Within specific specialties (e.g. medical oncology, haemato-oncology, colorectal, gynaecology, breast), a **lead consultant** should lead on implementing Regional Cancer Predisposition Services, including ensuring access to mainstreamed testing where applicable and ongoing management of those with an identified cancer predisposition.

The exact configuration of specialties and their role in delivery of Regional Cancer Predisposition Services will vary according to services available in the region and may change over time – e.g. specific surgical oncology, medical oncology, haematology services. Collaborative working will be key, as patient needs frequently cross clinical specialties. Pathways for the management of those with an identified cancer predisposition may also need to incorporate access to certain services from outside the region.

Nursing

Clinical Nurse Specialists (CNSs) have considerable input in managing patient care within cancer predisposition services. They are expert practitioners who have attained, at a minimum, a post graduate qualification in their specialist area of practice. They coordinate patient care from the time of referral to their service. The CNS has the primary responsibility for educating and supporting the patient/carer in this service.

The Advanced Nurse Practitioner (ANP) role in cancer predisposition services provides autonomous caseload management with high levels of clinical decision-making. While working within the multidisciplinary team and having attained a masters qualification, they have a greater level of autonomy and can complete a full episode of care. The ANP plans and initiates care and treatment to achieve patient-centred outcomes and evaluates their effectiveness, initiating and completing a care episode. The ANP can initiate and implement innovation and change in their healthcare service in response to patient/client need and service demand.

CNS and ANP members of teams are key to the delivery of the Regional Cancer Predisposition Services, with roles including assessment of family history, patient pre-test counselling and provision of continuity of care and coordinated care for those identified with a cancer predisposition. Administrative support in this area will allow skilled nursing staff to work at the top of their licence and will support the coordination of care across different clinical areas.

Table 2: Sample Staff in Regional Cancer Predisposition Service

<table>
<tr><td colspan="7"><u>Sample</u> staff in Regional Cancer Predisposition Service</td></tr>
<tr><td colspan="7">Clinical Lead for Cancer Predisposition Services</td></tr>
<tr><td>Medical oncology</td><td>Haemato-oncology</td><td>Colorectal service</td><td>Gynaecology</td><td>Breast service</td><td rowspan="2">Admin staff</td><td rowspan="2">Genetic Counsellor*</td></tr>
<tr><td>Consultant</td><td>Consultant</td><td>Consultant</td><td>Consultant</td><td>Consultant</td></tr>
<tr><td>CNS</td><td>CNS</td><td>CNS</td><td>CNS</td><td>CNS</td><td colspan="2" rowspan="2">Genomic Resource Associate</td></tr>
<tr><td>ANP</td><td>ANP</td><td>ANP</td><td>ANP</td><td>ANP</td></tr>
<tr><td colspan="7">Pathology, laboratory, radiology, endoscopy, psychooncology services</td></tr>
</table>

**may be employed by cancer centre or Specialist Cancer Genetics Service but professional reporting relationship to specialist cancer genetics*

4.4 Specialist Cancer Genetics Service

The Specialist Cancer Genetics Service will function as the national centre for clinical expertise in Ireland and will provide support and advice to Regional Cancer Predisposition Service sites via a network of designated genetic counsellors, a tertiary referral service and national MDT(s).

The role of the Specialist Cancer Genetics Service will include:

- developing national guidance, including protocols for mainstreamed services as appropriate. All standards, protocols and guidelines will be agreed nationally, and founded on the best available evidence and best international practice, appropriately applied in the Irish context.

- horizon scanning - as cancer genetics and genomic services and techniques continue to advance, it is important to ensure that the introduction of new tests and phasing out of others is informed by specific guidelines, and that this will be audited.
- developing and delivering educational initiatives to the wider workforce. Guidance is specifically required to support the implementation of mainstreamed pathways.
- supporting non-geneticists who are providing pre-test counselling.
- providing a regional cascade testing service via the networked genetic counsellor resource.
- providing comprehensive pre or post-test genetic counselling as required, including reproductive information, on site or virtually.
- providing a tertiary referral service for complex cases and consultation on variants of uncertain significance (VUS) – referral pathways will be agreed to facilitate referral of patients to the Specialist Cancer Genetics Service by a clinical team based at the Regional Cancer Predisposition Services.
- Support in the management of complex cases may be provided remotely or in person at a tertiary referral centre (+/- discussion at a national MDT).

Adult and paediatric patients have differing needs and should be assessed and managed in an appropriate environment (see below).

4.4.1 Specialist Cancer Genetics Services - Workforce

Consultants

Consultant expertise in specialist cancer genetics is provided by (i) those trained as Consultants in Clinical Genetics and (ii) those trained as oncology consultants (medical, surgical, radiation or haemato-oncology), who have developed expertise in the area of cancer genetics, specifically cancer predisposition. Those trained in oncology have a greater understanding of the overlap with current or future cancer treatments, whereas clinical geneticists have a broader role in areas such as syndrome overlap with non-cancer areas. Both areas of expertise are valued and should be maintained.

Of note, the oncology consultant role referred to here is different to that defined within Regional Cancer Predisposition Services. The lead role regionally may be held by someone with an interest in service development in this area but would not require the clinical expertise to provide a tertiary referral Specialist Cancer Genetics Service. However, regional expertise can be expected to develop further over time, particularly in the area of more common hereditary cancer syndromes.

Genetic Counsellors

As previously outlined, some genetic counsellors will provide a dedicated service to Regional Cancer Predisposition Services but will maintain both a sessional commitment and a professional reporting relationship to the Specialist Cancer Genetics Service.

Genetic Counsellors will also operate as part of the tertiary referral service in cancer specialist genetics, will have a role in education, training and guidance development and provide a clinical governance structure for genetic counsellors working regionally.

Challenges in Ireland have been noted in relation to the lack of career pathway, HSE grade codes, Health & Social Care

Professionals Council (CORU) recognition, and registration for genetic counsellors. This has been highlighted as a priority issue for the national genetics and genomics office.

Administrative and IT support

As outlined for regional services, the administrative workload for genetics is high compared to other specialties. The team will also benefit from Genomic Resources Associates and support to ensure coordination of national MDT(s) and the genetic counsellor network.

Laboratory expertise

Molecular diagnostic expertise is required as part of the specialist cancer genetics MDT, particularly in relation to the interpretation of Variants of Uncertain Significance. Of note, the broader laboratory infrastructure and clinical and scientific expertise required for germline genetic testing is outside the scope of this document and will be considered elsewhere.

4.5 Paediatric and AYA Cancer Predisposition Service

Childhood, adolescent and young adult (CAYA) cancers are rare and complex disorders, with differing cancer types and different disease biologies to those seen in adults. In addition, these cancers are associated with a higher rate of cancer predisposition, with less influence of environmental factors. Currently, it is estimated that up to 30% of CAYAs with cancer require genetic evaluation, with at least 10% of CAYAs with cancer having a confirmed cancer predisposition syndrome.

A comprehensive service is therefore required that incorporates cancer predisposition risk assessment for all CAYA patients. In addition, as the standard of care is moving internationally towards somatic testing of all tumours diagnosed in the CAYA population, germline testing for all will likely be required, either in sequence or parallel, necessitating expertise in relation to interpretation and management.

Children's cancer services are already centralised at the National Children's Cancer Service (NCCS), based at Children's Health Ireland at Crumlin. The development of a formal national network for AYA services is currently underway involving CHI, St James's Hospital, Cork University Hospital and University Hospital Galway (Framework for the Care and Support of Adolescent and Young Adults (AYA) with cancer in Ireland[2]). The provision of a single national specialist paediatric MDT for cancer genetics fits with this model and such a service has already commenced at CHI at Crumlin. The multidisciplinary input required crosses specialist genetics, oncology/ haematology, molecular pathology and the wider CAYA cancer team. Collaboration will be required between this MDT and that at the AYA service in St James' Hospital, to meet the needs of this age group.

In addition to providing access from regional paediatrics services to/from this specialist service, consideration should also be given to cross over with Regional Cancer Predisposition Services who are dealing with adults. For example, it may be appropriate for genetic testing to be carried out in children of adults diagnosed with certain cancer predisposition syndromes. These children should be referred to specialist genetics services associated with paediatric cancer predisposition services, for age-appropriate genetic counselling and management. The benefit of consultant clinical geneticists and genetic counsellors is that they are skilled to deal with all ages and all types of clinical presentations (syndromic/ non-syndromic), cancer risks and risks of other medical conditions, across all age spectrums.

Similarly, when cancer predisposition is identified in a child, testing of adults may identify those with the same predisposition and who will benefit from ongoing management in their regional cancer predisposition service.

4.5.1 Challenges specific to the Children, Adolescent and Young Adult population

Challenges relating to cancer predisposition in the CAYA population will vary according to the age of the child or young person, with some diagnoses of cancer predisposition as early as ante-natally or even pre-natally. Reproductive advice and referral to services is required for many parents of children with identified cancer predisposition, as the parents are themselves still in reproductive age. For children themselves, it is important that this information regarding reproduction and fertility is raised at an age-appropriate time. This can be incorporated into genetics review during the AYA transition period. Other issues for consideration at this time include any need for further genetic testing, any changes in surveillance recommendations, and ownership of the diagnosis going forward.

Specific challenges can also arise in relation to disease surveillance in this population, particularly younger children. These include greater reliance on ultrasound and MRI techniques to minimise exposure to radiation, the need for hospital day ward admission for young patients in order to access surveillance imaging procedures under general anaesthesia with appropriate medical and nursing supports needed to provide this level of care, and frequent clinical review and examination by experienced health professionals.

5. Clinical Pathways

Detailed clinical pathways will need to be developed, or international guidelines adopted, in relation to a broad range of possible cancer predisposition.

Patient pathways should include the following:

- identification of those requiring family risk/ genetic assessment
- details of assessment tools to be used
- formal criteria for genetic testing
- types of testing to be employed (e.g. a national test directory similar to NHS) and
- recommendations on follow-up and risk reduction.

Such pathways and guidance will be agreed under the governance of the National Genetics and Genomics Office (once established) and the National Cancer Control Programme and should be easily available to clinicians, e.g. maintained online.

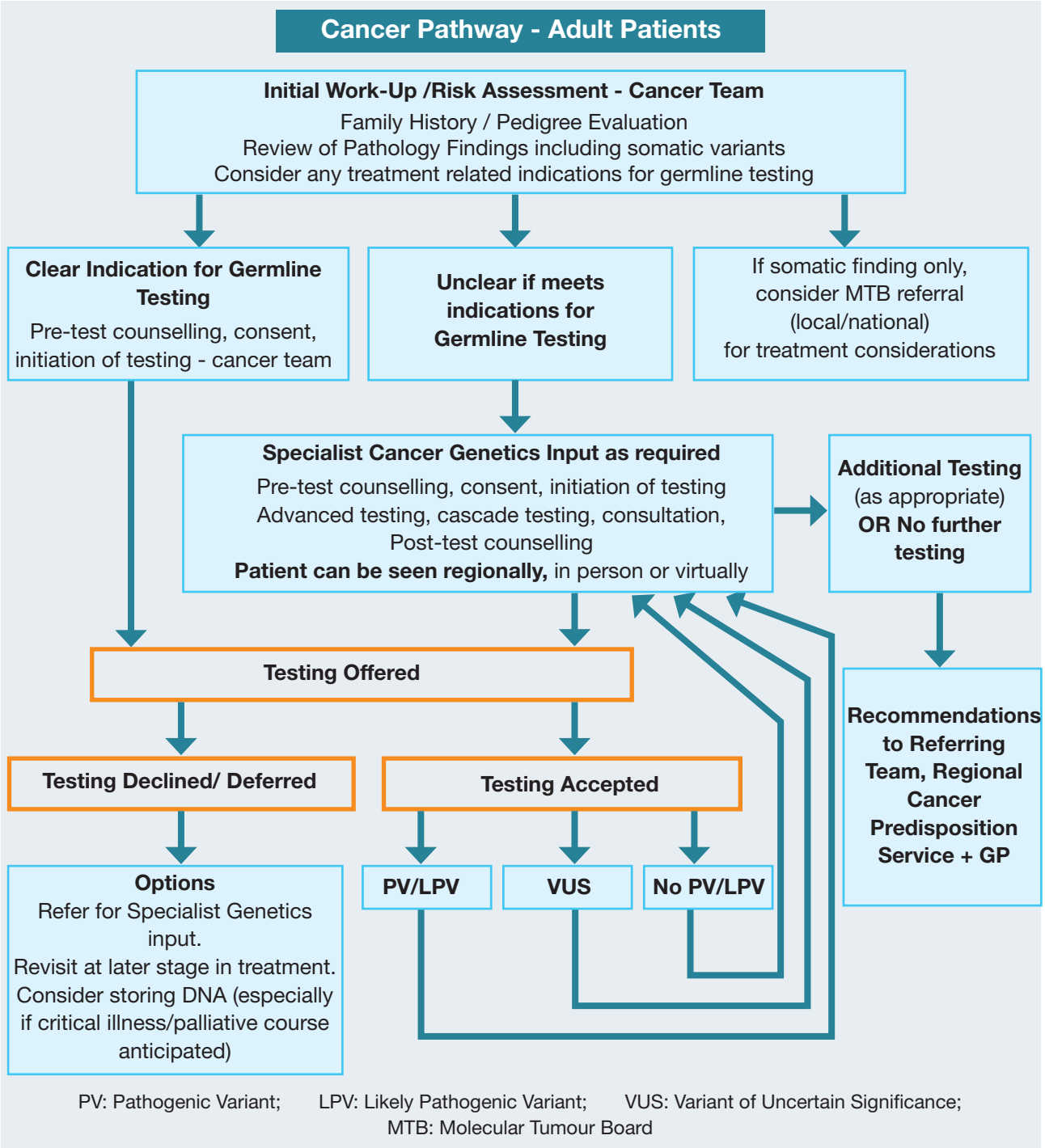
Generic pathways are presented here to outline the proposed patient flow across Regional Cancer Predisposition Services and specialist cancer genetics.

5.1 Pathways for referral, risk assessment and testing of people who may have a cancer predisposition

5.1.1 Adults with a cancer diagnosis

The implementation of "mainstreamed" pathways, as appropriate, will ensure timely access to genetic testing for those with a cancer diagnosis. A national timeline to provide patients with genetic consultations, and testing (if required) will need to be developed and agreed upon as part of a standard management programme. This is key to effectively managing patient anxiety and allowing for timely surveillance/ chemoprevention/ risk-reducing surgery options to be explored.

Pathway A: Adult Patients with a Cancer Diagnosis



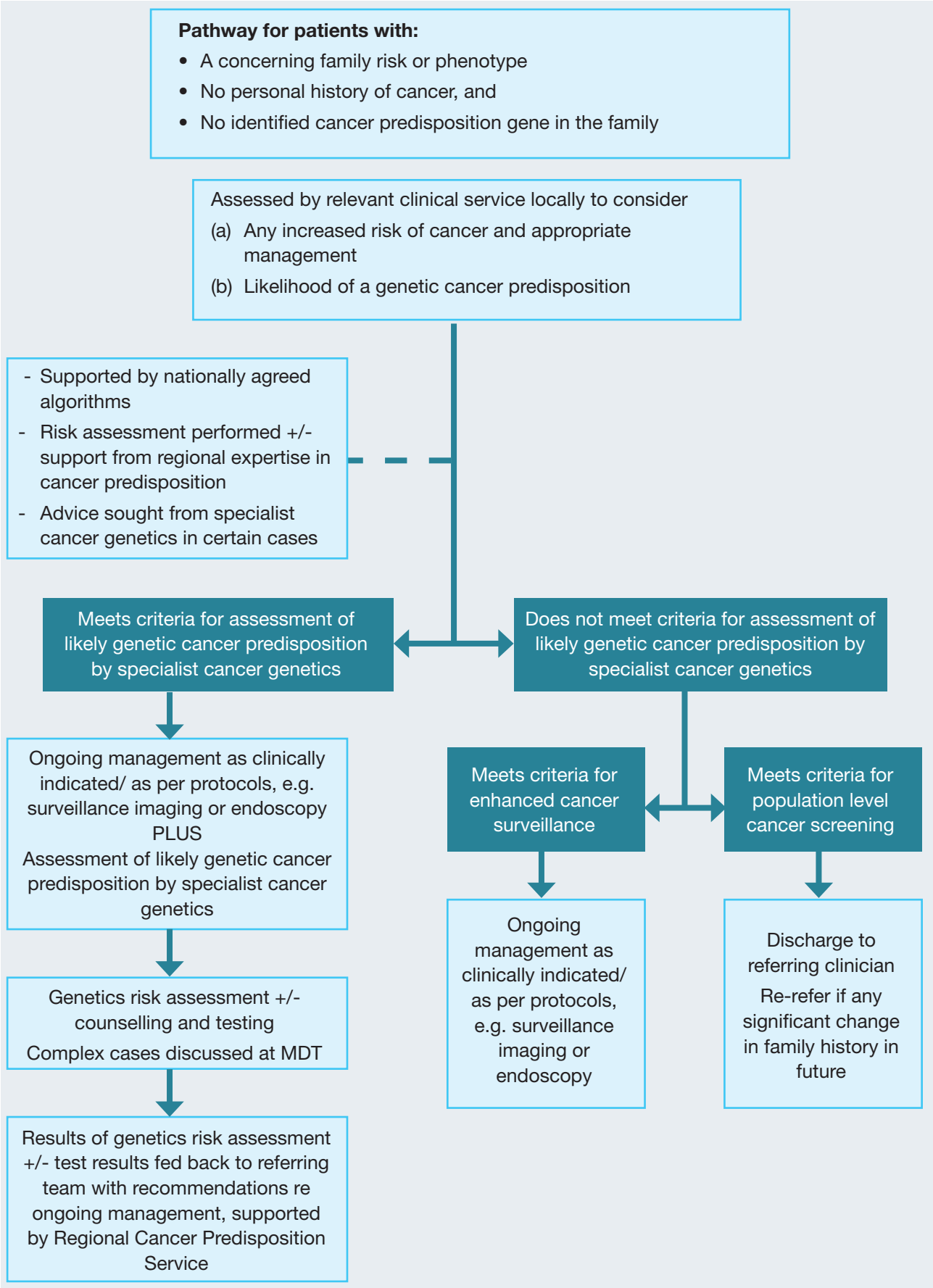
PV: Pathogenic Variant; LPV: Likely Pathogenic Variant; VUS: Variant of Uncertain Significance; MTB: Molecular Tumour Board

5.1.2 Adults without a cancer diagnosis

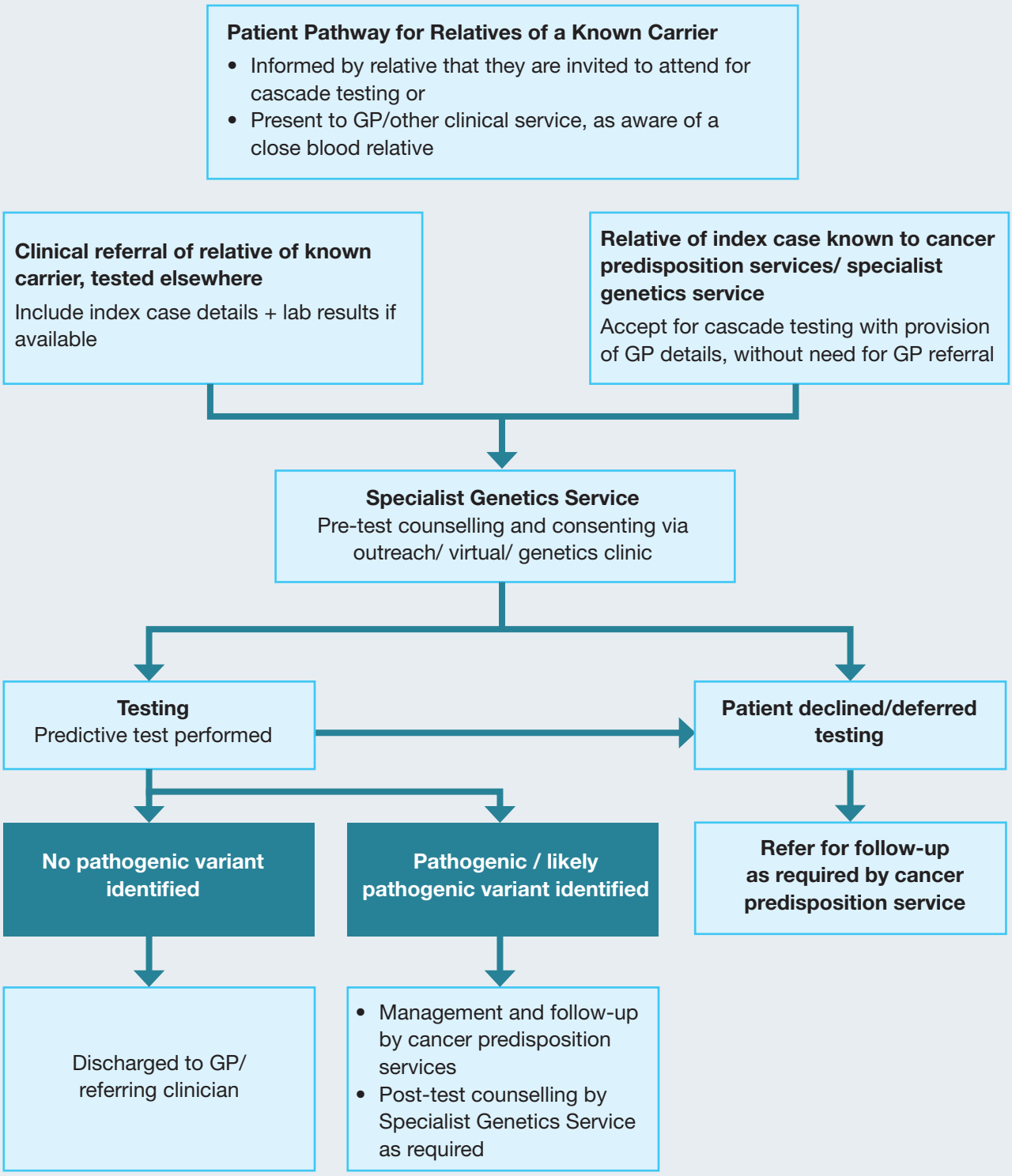
Where testing is undertaken in an individual who does not have a cancer diagnosis (e.g. a relative as part of cascade testing), this will be managed by a trained Genetic Counsellor. This can be service can be delivered regionally, whether by a genetic counsellor embedded within the regional service, or via outreach clinic or virtual appointments.

Obtaining a GP referral should not be a barrier to relatives availing of cascade testing. However, a relative should be asked to provide GP details, so that their GP can receive information on outcome of testing and any follow-up recommendations. Referral for ongoing management and follow up, if required, should be to the relevant regional cancer predisposition service.

Pathway B: Patients without a personal history of cancer, other than cascade testing



Pathway C: Cascade testing for unaffected relatives of those with an identified cancer predisposition gene



*Scope for virtual genetics clinics will be included (with potential for the expansion of local services, as expertise grow; with the support of specialist genetics service).

**Where testing is indicated for paediatric relatives, this should be carried out by the Specialist Cancer Genetics Service (see Pathway D). Where testing is not clinically indicated until a child reaches adulthood, information systems should capture the recommendation for future testing.

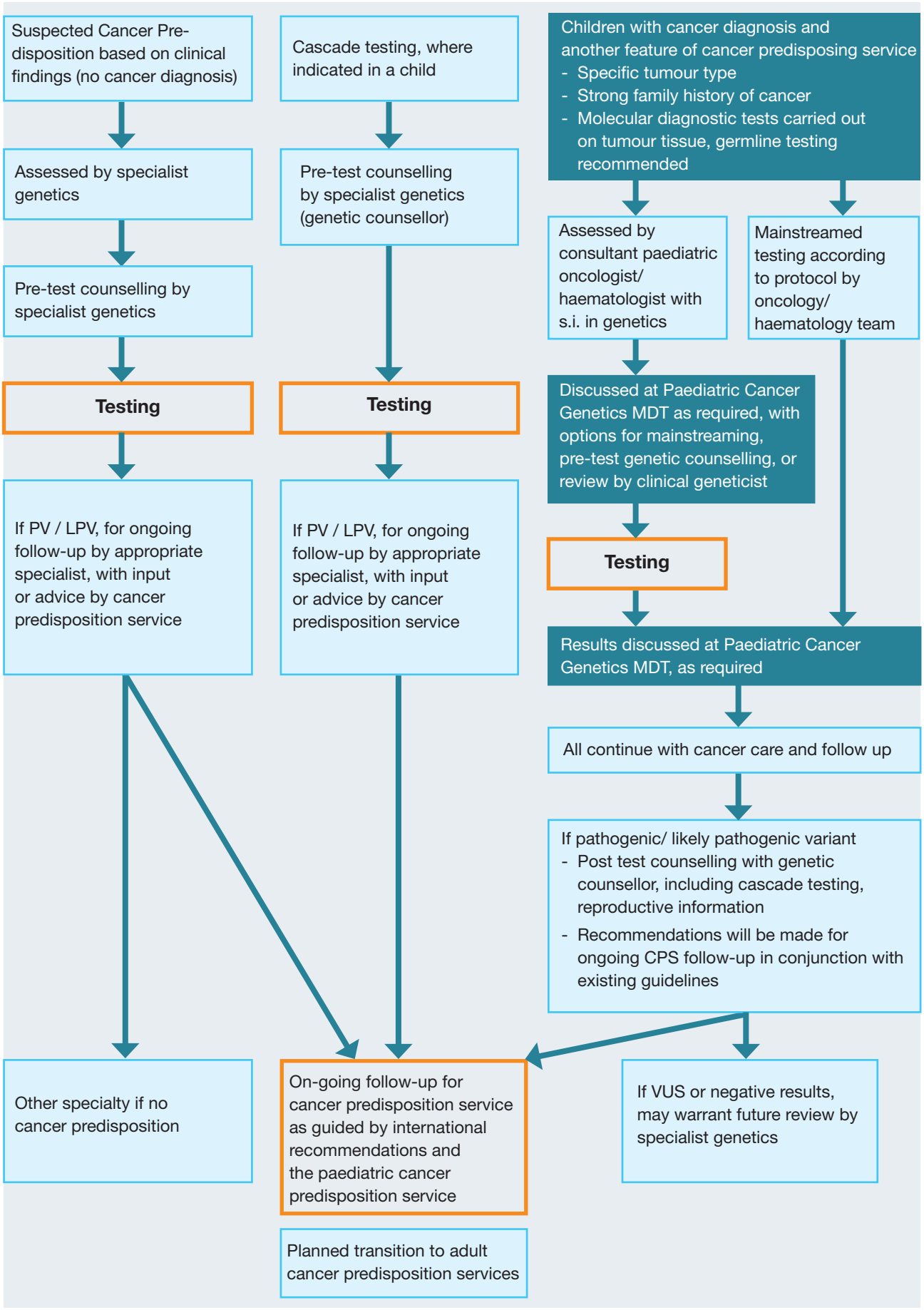
5.1.3 Assessment, testing and counselling of paediatric patients

Pathway D gives an overview of the pathway for paediatric patients, including those with a personal cancer diagnosis, those who have been identified for cascade testing (where it is appropriate at this age) and those without a cancer diagnosis but where there is a suspicion clinically of a cancer predisposition syndrome.

In a child with a personal history of cancer, the suspicion of cancer predisposition may arise due e.g. to the particular tumour type or tumour pathology, results of molecular diagnostic tests, the occurrence of multiple cancers in an individual or a significant family history of cancer.

A pathway specific to Adolescents and Young Adults is to be developed, in line with the principles of the AYA Model of Care[2] to ensure age-appropriate care. This is to be done while harnessing the expertise of both paediatric and adult oncology/ haematology specialists, in conjunction with clinical genetics services, who cover all age groups as consultant clinical geneticists/ genetic counsellors jointly trained in paediatric and adult genetics.

Pathway D: Paediatric Pathway



5.2 Clinical pathways for management of people with identified cancer predisposition

A dedicated pathway with protected resources is required for the management of those identified with a pathogenic variant/ inherited cancer predisposition – across clinical follow up, surveillance imaging/ endoscopy, risk-reducing surgery, chemoprevention and wider supportive management (e.g. psychological support, health and wellbeing advice, management of adverse effects of risk reduction measures, reproduction and fertility). This will be required, whether or not the patient has an existing diagnosis of cancer.

Pathway E outlines generic components of care for those with an identified cancer predisposition. Condition-specific pathways will be need to be agreed, including consideration of international best practice.

5.2.1 Key components of care for people diagnosed with a cancer predisposing genetic variant

Access to Specialist Cancer Genetics Services/expertise

Those with a newly identified cancer predisposition/ pathogenic gene variant should have access to a suitably qualified healthcare professional after they receive their diagnosis, to ask any questions that arise after they have had a chance to process their diagnosis.

A designated genetics-trained healthcare professional, e.g Advanced Nurse Practitioners (ANPs) in the Regional Cancer Predisposition Service/ cancer centres, could address some of these clinical queries, where related to the more common hereditary predisposition syndromes. Other requirements will fall within the remit of Specialist Cancer Genetic Services, e.g. Genetic Counsellors providing familial and reproductive counselling, noting that this specialist genetics resource will be delivered at regional level.

Timely access to risk-reducing interventions

Sufficient resourcing is required to ensure timely ongoing clinical follow-up and access to necessary interventions, including surveillance imaging/ endoscopy, chemoprevention and risk-reducing surgeries. A system is required to ensure interventions can be offered and delivered in a timely fashion at the appropriate age.

In particular, the appropriate configuration of services for surveillance and risk-reducing surgery must be addressed, to minimise wait times and ensure the best clinical outcomes for patients. Condition-specific pathways must also address the need for specialist pathology input, when dealing with specimens from prophylactic surgeries in those with a known pathogenic gene variant.

Psycho-oncology

Psychological support (including professional and peer-based) should be offered (but not mandatory) to all carriers following a diagnosis of a pathogenic variant. This should be available if needed throughout the patient journey, including but not only at times such as pre/ post risk-reducing surgery and in the event of a cancer diagnosis.

Additional education initiatives will be required to meet the training needs of the psychology profession, in relation to the area of inherited cancer predisposition.

Reproduction and Fertility

Carriers of pathogenic gene variants should be provided with information regarding specialist advice on reproduction and fertility. Fertility services in this context should be made available free at point of care in the public sector and in a timely fashion.

5.2.2 Paediatric and AYA patient considerations

As outlined in Pathway D, paediatric patients with inherited cancer predisposition should have their follow-up care led by a single national service at CHI. The need for general anaesthetic for certain surveillance procedures at a young age is one example of the additional complexity of surveillance in this age group. Other specific considerations include access to age-appropriate psycho-oncology services and access to reproductive/ fertility services at age of transition.

Where appropriate for more common conditions, services may be developed to include coordination of care by the national paediatric cancer predisposition service, with clinical follow up and surveillance procedures delivered in an age-appropriate setting regionally.

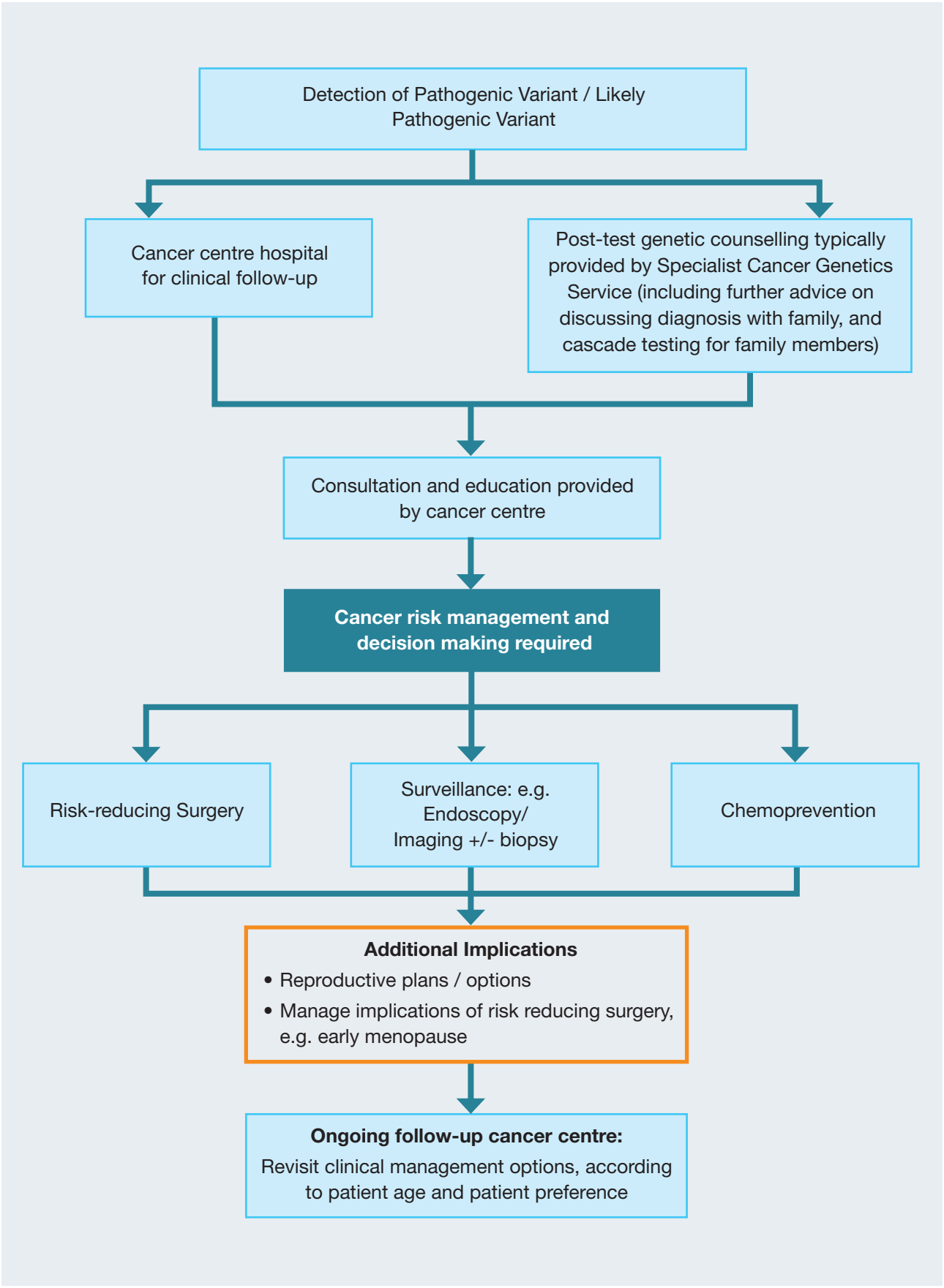
A pathway specific to Adolescents and Young Adults is also to be developed, in line with principles of the AYA Model of Care[2] to ensure age-appropriate care.

For most cancer predisposition syndromes diagnosed in adulthood, genetic testing would not be clinically indicated for paediatric family members. Condition-specific pathways should include the appropriate age for testing. Information systems should be used to capture and communicate the recommendation for testing at the appropriate time.

5.2.3 Pathway for management of people with inherited cancer predisposition

Regardless of the original pathway to identification, a comprehensive service is then required for those with a pathogenic/ likely pathogenic variant (Pathway E).

Pathway E: Management Algorithm for those with a pathogenic/ likely pathogenic cancer predisposing genetic variant



6. Key enablers of service delivery

Key enablers of effective delivery of cancer genetics services in Ireland include:

- Sufficient resources and funding to meet current and future service demand
- Clearly defined patient care pathways
- Governance
- Quality assurance, service monitoring and evaluation
- Appropriate IT infrastructure and data management
- Appropriate use of technology
- Education

6.1 Sufficient resources and funding

Guaranteed long-term funding is required for the Specialist Cancer Genetics Service and the Regional Cancer Predisposition Services in order to meet current and future demand.

The services should be staffed by sufficient numbers of appropriately trained staff with clearly defined roles and responsibilities. Sufficient resources are required right across the pathway, e.g. for assessment and counselling, laboratory testing; ongoing clinical follow-up; surveillance imaging and endoscopy; risk-reduction surgeries; comprehensive patient services including psycho-oncology, fertility and health and wellbeing supports; and service enablers such as data management and education.

6.2 Governance

A clear governance framework is required for hereditary cancer services in Ireland.

A National Genetics and Genomics Office being established, in line with the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland[1]. It will have responsibility for agreement of national standards (practices and protocols), guidelines, education and training, and delivery of specialised genetic services e.g. family planning, and workforce development.

The National Cancer Control Programme should be closely engaged with in relation to the development of services related to an inherited predisposition to cancer and a process agreed for the development and agreement of guidance and pathways relevant to cancer predisposition.

The Specialist Cancer Genetics Services will have clinical responsibility for development and agreement of national guidance for their speciality and have professional oversight of specialist genetics services (such as genetic counselling) delivered within Regional Cancer Predisposition Services.

Within the Regional Cancer Predisposition Services, the Clinical Lead for Cancer Predisposition will ensure that staff are appropriately trained for and support in the tasks they are undertaking and that as a service, there is

- A coordinated approach to service management
- Adherence to national protocols and
- Regular audits are undertaken.

6.3 Quality assurance, service monitoring and evaluation

All elements of the patient care pathway (structures, processes and outcomes) should have defined quality standards and a subset of specific key performance indicators to facilitate service monitoring, quality assurance and service improvement.

Patient or family-reported experience measures should be embedded as a quality indicator. Experience of patients who have received/are receiving care from the Specialist Cancer Genetics Service and/or Regional Cancer Predisposition Services should be included in the cancer patients experience survey (Recommendation 35 of the National Cancer Strategy[3] - The NCCP will define focused cancer patient experience surveys to incorporate treatment and survivorship in line with HIQA's standard approach for the National In-Patient Acute Care Patient Experience Survey).

Audits should be undertaken to ensure equitable access to services irrespective of patients' age, geographic location and socioeconomic status.

6.4 Appropriate IT Infrastructure and data management

A well-integrated IT system, linked to GP services and hospitals is required to enable effective delivery of the proposed distributed model of care.

A national database for inherited cancer predisposition, based on the use of a unique patient identifier, would support national planning and coordination of services for the population. Moreover, the development of a national database would allow health information to be accessed as required across different sites. Ideally, this would include data from public and private clinical services, while acknowledging that the HSE only has jurisdiction over/ responsibility for publicly delivered health services in Ireland. Commercial solutions with integrated patient support packages should be considered.

The approach taken to IT infrastructure will need to align with that taken by the National Genetics and Genomics Office, regarding wider services, laboratory test results and patient registries.

Options include:

- Development of a national database for Specialist Cancer Genetics and Regional Cancer Predisposition Services
- Each Regional Cancer Predisposition Site to host an individual system.

Genetic and genomic information (test results, as opposed to raw data) should be an integral component of clinical data available to patients with a cancer diagnosis. A formal process will be required to record patient consent digitally, including consent to the sharing of genetic information with family members.

At a minimum, an agreed minimum dataset should be established, to accurately record data on patients with a pathogenic gene variant. This would support Regional Cancer Predisposition Services to coordinate patient follow-up and actively participate in audit.

Genetic and genomic information (test results, as opposed to raw data) should be an integral component of clinical data available for an individual with a cancer diagnosis.

Each Regional Cancer Predisposition Service will require a data manager to facilitate reporting on service activity, and KPI reporting.

6.5 Technological aids

The use of technological aids, such as virtual assessment and decision aid tools, should be employed appropriately to maximise efficiency and support the use of protocols.

Guidance for clinicians should be maintained online for ease of access and resourced to ensure reflecting current best practice.

Virtual appointments can be more convenient and less financially costly for patients and can also be facilitated in a more local hospital. Consideration should be given to equity of access (barriers may include digital poverty or insufficient digital literacy to enable virtual

engagement with the services), and to patient preference, particularly if there is potential for receiving an initial diagnosis virtually.

Group genetic counselling, including digitised formats such as website information, self-paced learning for informed consent, and genetic testing to improve access in pre-test context are also possible and could improve.

6.6 Education

6.6.1 Education of the workforce

Information resources and education initiatives are required for healthcare professionals working in primary care, non-oncology secondary care and cancer services in Ireland. These will support the implementation of guidelines, pathways and protocols, and should include accredited training for those delivering mainstreamed pathways (currently under development). These resources should be developed in consultation with clinical subject matter experts.

A genetic counselling training pathway should be established through existing academic and clinical support programmes to increase workforce capacity, as part of the implementation of the National Strategy for Accelerating Genetic and Genomic Medicine in Ireland.

6.6.2 Patient Education

Patient stakeholders will be integral partners in the co-design of patient-facing educational resources.

7. Implementation

The implementation of this Model of Care will be a multi-year project. An implementation group will be established to oversee this work, encompassing the following areas:

- Guidance and clinical pathway development (*BRCA*, Lynch, other)
- Workforce
- Healthcare professional education and training
- Data requirements and Quality Assurance
- Patient Information and supports (general, and specific to cancer predisposition syndromes, e.g. *BRCA*, Lynch syndrome etc.)
- Synergies with the National Genetics and Genomics Office, once established.

Recommendations for implementation are outlined here, for detailed population and refinement by the implementation steering group once established.

Thematic Area 1: **Establish oversight for the implementation of the Hereditary Cancer Model of Care**
1.1 Establish a governance and reporting structure aligned to the National Cancer Strategy and National Cancer Control Programme
1.2 Agree areas for collaboration with the National Genetics and Genomics Office once in place
1.3 Convene a multi-disciplinary implementation steering group, with clinical leadership and patient involvement
1.4 Establish relevant working groups for thematic priorities
1.5 Agree an overarching governance framework for hereditary cancer service
1.6 Appoint Clinical Leads for Cancer Predisposition within regional services
1.7 Report at minimum annually on progress in implementation
Thematic Area 2 : **Development of networked services and MDTs**
2.1 Define the process(es) by which genetic counselling services can be networked and made accessible regionally
2.2 Develop a standardised pathway to a national specialist cancer genetics MDT (adults)
2.3 Develop a standardised pathway to a national specialist cancer genetics MDT (CAYA)
2.4 Establish a network of regional cancer predisposition leads

Thematic Area 3 : Healthcare professional education and training; workforce development
3.1 Identify the training needs of staff working in generalist settings
3.2 Identify the training needs of staff working in Regional Cancer Predisposition Services
3.3 Develop an accredited online programme to meet the minimal learning needs of staff working in regional services
3.4 Identify or develop further training courses to meet identified needs of staff
3.5 Work with the National Genetics and Genomics Office, professional training bodies and regulators to develop career pathways in specialist cancer genetics
Thematic Area 4: **Clinical guidance and pathway development**
4.1 Agree the guidance development and approval process, in line with HSE policy, including guidance on laboratory testing
4.2 Establish guidance development groups for common presentations and cancer predispositions
4.3 Ensure patients are involved in the co-design of clinical pathways
4.4 Agree the process by which international guidance relating to rarer conditions will be approved and disseminated for use in Ireland
4.5 Agree the platform(s) through which current guidance will be made available to all
4.6 Highlight critical points in clinical pathways, which should be subject to regular audit
4.7 Describe resource requirements for implementation of pathways
Thematic Area 5: **Patient Information and supports**
5.1 Establish subgroups for development of patient information, according to common presentations and cancer predispositions
5.2 Ensure patients are engaged in the co-design of patient information materials
5.3 Consider how best to meet the information needs of underserved populations and those with health literacy challenges
5.4 Agree or develop platform(s) through which patient information can be most easily accessed
5.5 Ensure evaluation of information materials and supports provided

Thematic Area 6: Data Requirements and IT infrastructure
6.1 Explore the role of virtual patient assessment within hereditary cancer services
6.2 Identify the IT infrastructure required to enable the implementation of this model of care, including a genetic counselling network and referral to a national specialist genetics MDT.
6.3 Examine potential data capture options with the National Cancer Information System
6.4 Engage with the National Genetics and Genomics Office in relation to development of registries for cancer predisposition
6.5 Agree minimum datasets for common presentations and cancer predispositions
6.6 Agree key performance indicators for the delivery of specialist cancer genetics and Regional Cancer Predisposition Services

8. Benchmarks of success

Strategic Priorities		Details
	Partnerships/ MDT working	A networked approach to all public services treating patients undergoing cancer genetic testing across the state Development of a formal network, encompassing the Specialised Cancer Genetics Service and Regional Cancer Predisposition Services Timely access to genetics assessment +/- testing and result Each cancer centre should have a clear pathway of how they refer patients with an identified cancer predisposition/ pathogenic gene variant(s) to suitably qualified healthcare professionals Development of National MDMs Creation of new services specific to the needs of patients within the Specialist Genetics Service and the Cancer Predisposition Service Co-design of services by those with lived experience of the Specialist Genetics Service and the Regional Cancer Predisposition Service A systematic and collaborative approach to the private and charity sector supporting patients receiving care in the Specialist Genetics Service and the Regional Cancer Predisposition Service Representation by patients from the Specialist Genetics Service / the Regional Cancer Predisposition Service with lived experience contributing to relevant working groups within the NCCP Representation by parent/family members in key working groups

	Education and Information	The provision of accessible, jargon-free and inclusive/ equitable (across age, language and ethnicity) educational resources Development of additional education and training resources to improve genetic literacy among healthcare professionals Provide healthcare professionals responsible for implementing mainstreamed pathways with specific, accredited training
	Workforce	An integrated network of healthcare professionals with special education and training in both the Specialist Cancer Genetics Service and Regional Cancer Predisposition Service All Regional Cancer Predisposition Services should have an identified, appropriately skilled workforce, to include consultants, ANPs, CNSs and administrative support, with dedicated access to genetic counsellors regionally. Create, promote and nurture a network of skilled and highly motivated professionals across the spectrum of health and social care professions that are educated and skilled in identifying and addressing the specific and unique needs of service users within the Specialist Cancer Genetics Service and the Regional Cancer Predisposition Services. Partner with the voluntary sector to create specific support for those receiving care from both the Specialist Genetics Service and Regional Cancer Predisposition Services
	Research	Strengthen the research community within the Specialist Cancer Genetics Service and the Regional Cancer Predisposition Service in Ireland Increase participation in multi-site international research to further develop evidence-based cancer genetics practice/ services
	Data	An ability to collect standardised data nationally to support planning, service monitoring and quality improvement Creation of Key Performance Indicators and benchmarking against international standards/services

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Appendix B: Membership of Advisory Group

Risteárd Ó Laoide	**Director, NCCP and Chair of Advisory Group**
Alan Smith	Specialist in Public Health Medicine, National Screening Service
Andrew Green	Professor of Genetic Medicine, University College Dublin and Consultant Clinical Geneticist CHI at Crumlin and Temple Street
Andrew Kelly	Cancer Policy Unit, Department of Health
Barbara McGrogan	Medical Scientist, NCCP
Cathal O'Brien	Chief Scientist, Cancer Molecular Laboratory, St. James's Hospital
Ciaran Murphy	Cancer Policy Unit, Department of Health
Claire Giffney	Principal Genetic Counsellor, CHI at Crumlin
Claire Meaney	Senior Pharmacist, NCCP
David Gallagher	Consultant Medical Oncologist and Cancer Geneticist, St. James's Hospital
Deirdre Browne	Project Manager, NCCP
Deirdre Murray	Director, National Cancer Registry Ireland
Eoin Dornan	Cancer Policy Unit, Department of Health
Eve O'Toole	Head of Evidence and Quality Hub, NCCP
Fiona Kelly	CNM 3 Nursing Projects Manager, NCCP
Heather Burns	Specialist in Public Health Medicine, NCCP
Helen Simon	Cancer Policy Unit, Department of Health
John Gleeson	Programme Manager, NCCP
Karen Cadoo	Consultant Medical Oncologist and Cancer Geneticist, St. James's Hospital
Karl Silver	Department of Health Coordinator for Cancer Patient Advisory Group
Lisa Bradley	Consultant Clinical Geneticist and Lead for Cancer Genetics, CHI at Crumlin
Lisa Cadden	Breast ANP, St. Vincent's University Hospital
Mary Forry	Colorectal ANP, Beaumont Hospital
Mechelle Loughrey	Colorectal ANP, St. Vincent's University Hospital
Michael Kerin	Consultant Breast Surgeon, University Hospital Galway and Saolta Cancer MCAN Director
Niamh Killgallen	Senior Researcher, NCCP
Nicolette Warner	Patient Representative, Marie Keating Foundation BRCA Peer Support Group
Nina Orfali	Consultant Haematologist, St. James's Hospital
Noelle Cullinan	Consultant Paediatric Oncologist, CHI at Crumlin
Owen Smith	National Clinical Lead for CAYA, NCCP
Pat Fahey	Patient Representative, Lynch Syndrome Ireland
Patricia Heckmann	Systemic Therapy Programme Lead, NCCP
Rachel McKeon	Patient Representative, Marie Keating Foundation BRCA Peer Support Group and OvaCare
Richard Bambury	Consultant Medical Oncologist, Cork University Hospital
Roberta Horgan	Patient representative, Lynch Syndrome Ireland
Rory Kennelly	Consultant Colorectal Surgeon, St. Vincent's University Hospital
Rosie O'Shea	Principal Genetic Counsellor, St. James's Hospital
Sarah Rose Flynn	Cancer Policy Unit, Department of Health
Sinead Whyte	Senior Genetic Counsellor, CHI at Crumlin
Terry Hanan	National Clinical Lead Cancer Nursing, NCCP
Triona McCarthy	Consultant in Public Health Medicine, NCCP
Trudi McDevitt	Senior Clinical Scientist, CHI at Crumlin
Una Kennedy	GP Advisor, NCCP

Appendix C: *BRCA* Health Needs Assessment Recommendations

Table C.1: Recommendations arising from the NCCP BRCA health needs assessment

	Theme	Recommendations
1	Information needs	1.1 Accessible, inclusive (across age, language and ethnicity) and jargon-free informational resources should be developed for and available as part of routine care to BRCA carriers. Consultation with BRCA carrier stakeholders should be part of the development of any such resources. 1.2 Development of BRCA information resources specifically for healthcare professionals working in primary care and cancer centres in Ireland is required. This should be informed by consultation with clinical subject matter experts.
2	Data on *BRCA* in Ireland	2.1 A national database with an agreed minimum dataset should be established capturing data pertaining to BRCA carriers under follow-up in this country. Ideally this would include data from public and private clinical services. This could be developed as part of a national database for inherited cancer predisposition. Such a database should be based on the use of a unique patient identifier. It should support national planning and co-ordination of services for the BRCA population and facilitate local follow-up of patients. There should be adequate resources to maintain it. 2.1 Local databases, standardised with respect to an agreed minimum dataset i.e. a single data dictionary, should be established in cancer centres which capture data pertaining to *BRCA* carriers under follow-up.
3	Specialist Genetics input	3.1 BRCA carriers should have access to suitably qualified healthcare professionals after their consultation at diagnosis with clinical genetics, to ask any questions that arise after they have had a chance to process their diagnosis. Some of these queries could be addressed by designated genetics-trained healthcare professional(s) (e.g Advanced Nurse Practitioners (ANPs) once appropriate training provided) in cancer centres, while other queries would fall within the remit of Genetic Counsellors. 3.2 BRCA carriers should be provided with information regarding, and signposted to, family planning resources including accessing pre-implantation genetic testing (PGD) and in-vitro fertilisation (IVF). 3.3 BRCA carriers should be considered eligible for publicly available onco-fertility services.

4	Structured Care Pathway and Co-ordination of Care	4.1 Improved coordination of care is required, including between clinical genetics services and cancer centres. This could be delivered by a nurse-led service, with the CNS/ANP also acting as the dedicated point of contact for BRCA carriers. 4.2 Development of a nurse-led service for BRCA carriers will require specialist training and support. There is benefit to the autonomous clinical role of an ANP in this context. 4.3 The model of care (including clinical governance) for BRCA carriers needs to be improved and standardised across all cancer centres, to ensure consistency in and access to optimal standards of care in all geographic areas. Further dedicated consultation on the desired model of care is needed with input from all relevant stakeholders. 4.4 All elements of the patient care pathway (structures, processes and outcomes) should have defined quality standards and a subset of specific key performance indicators to facilitate performance measurement. 4.5 Patient-reported experience measures should be an embedded quality indicator.
5	Risk-reducing surgery	5.1 Risk-reducing breast and ovarian surgery for BRCA carriers requires a dedicated pathway with protected resources and should be delivered in a timely fashion. The appropriate configuration of services for risk-reducing surgery, that would result in low wait times and the best clinical outcomes for women, should now be examined. 5.2 Adequate post-operative support following risk-reducing surgery (e.g. physiotherapy, psychological support) should be available to patients. 5.3 All women should have timely access to specialist plastic surgery expertise regarding breast reconstruction options, and reconstructive surgery itself, if desired.
6	Surveillance	6.1 There should be protected magnetic resonance imaging (MRI) and mammography slots for breast surveillance of asymptomatic BRCA carriers, to ensure surveillance imaging occurs at recommended intervals. 6.2 Ovarian surveillance by transvaginal ultrasound and/or Cancer Antigen (CA)-125 measurement is not recommended for BRCA carriers. 6.3 Female BRCA carriers who request ovarian surveillance should be made aware of the lack of evidence to demonstrate a survival benefit. 6.4 Greater awareness of the risks and benefits of prostate cancer surveillance for male BRCA carriers – particularly for BRCA2 – is needed among patients as well as healthcare professionals. It is not yet known whether surveillance using PSA reduces mortality in men with a cancer-predisposing BRCA variant. 6.5 An evidence review of international pancreatic cancer surveillance guidelines for *BRCA* carriers is needed.

7	Psycho-logical support	7.1 Psychological support including professional support should be offered (but not mandatory) to all BRCA carriers following a diagnosis of a cancer-predisposing BRCA variant. This should be publicly available if needed throughout the patient journey, including but not only at times such as undergoing risk-reducing surgery and in the event of a cancer diagnosis. 7.2 Education initiatives should include the education and training needs of the psychology profession specific to inherited cancer predisposition. 7.3 BRCA carriers should be signposted following diagnosis to psychological supports (both professional and peer-based), including those external to cancer centres.
8	Women's health	8.1 Women should have access to clear information regarding the symptoms and longer-term health risks associated with early menopause, prior to risk-reducing surgery. 8.2 Women should have access to expert advice regarding their options for menopause management (hormonal and non-hormonal), which could be provided by a suitably trained doctor, ANP or other adequately trained healthcare professional. 8.3 Women should be referred to specialist menopause clinics if adequate support or expertise is not available in their hospital.

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