Australian Government
Cancer Australia

National Framework for Genomics in Cancer Control

January 2025

Statement of Acknowledgement

Cancer Australia acknowledges Aboriginal and Torres Strait Islander people as the Traditional Custodians of Country throughout Australia. We pay our respects to Elders, past and present.

We celebrate the ongoing connections of Aboriginal and Torres Strait Islander peoples to Country, culture, community, family and tradition and recognise these as integral to health, healing and wellbeing.

Cancer Australia acknowledges great diversity among Aboriginal and Torres Strait Islander peoples, and the contribution of the many voices, knowledge systems and experiences that guide all efforts to create a culturally safe and responsive cancer system that is equitable to all.

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ISBN Online: 978-1-74127-014-3

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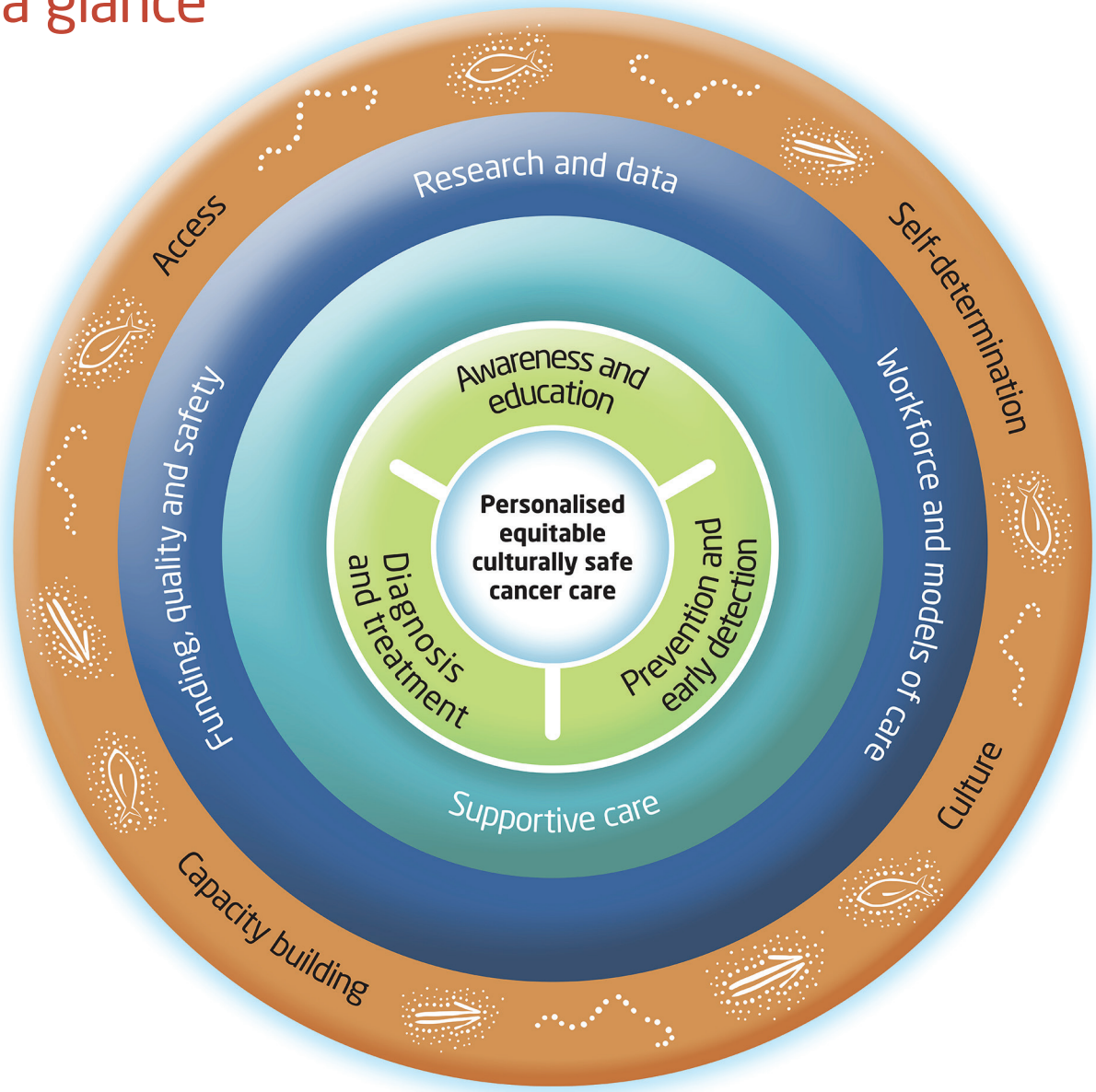

National Framework for Genomics in Cancer Control

Personalised, equitable, culturally safe cancer care

Strategic Objectives

Prevention and early detection

Genomic testing is integrated into healthcare to reduce cancer incidence and enable early detection

Goal

Evidence-based genomic testing is accessible to individuals and their families to support personalised prevention, risk-reduction and earlier detection of cancer.

Actions

1.1 As the evidence is established, expand access to affordable genomic testing to enable personalised cancer prevention and risk-reducing strategies.

1.2 Give priority to the establishment of evidence for genomic testing in Aboriginal and Torres Strait Islander people and other priority population groups and increase the scope of genomic reference datasets to more accurately reflect the diversity of the Australian population.

1.3 Promote translational research into personalised risk prediction and genomic risk-based cancer screening.

1.4 Embed evidence-based culturally safe genomic testing into national cancer screening programs, to enable risk-stratified screening.

Diagnosis, treatment and clinical trials

Genomic testing is used to guide diagnostic and treatment decisions to deliver better outcomes for people affected by cancer

Goal

Genomics technology is incorporated into routine cancer care to inform accurate diagnosis, personalised treatment and access to clinical trials.

Actions

2.1 Enhance access to genomic testing for people affected by cancer where there is evidence of benefit, supported by information and recommendations for management.

2.2 Enable timely access to genomics-informed cancer treatments through Health Technology Assessment (HTA) processes and clinical trials.

2.3 Promote translational research into cost-effective evidence-based genomic tools to support cancer detection, genomics-guided treatments and monitoring for progressive disease or recurrence, and transition from research to routine cancer care.

2.4 Build the evidence for genomics-guided treatments, including through expanded access to appropriate clinical trials, ensuring that the specialist cancer and primary care sectors, including Aboriginal Community Controlled Health Services, are engaged to support equitable patient participation.

Supportive care

The experience of people affected by cancer is improved by genomics-informed cancer care

Goal

Genomics is used to optimise treatment response and minimise treatment toxicity, and people affected by cancer receive psychosocial care when needed.

Actions

3.1 Provide tailored clinical and psychosocial support to people affected by cancer, according to their genomic profile and individual preferences.

3.2 Provide Aboriginal and Torres Strait Islander people and other priority populations with holistic navigation support and wrap-around personalised genomic cancer care.

3.3 Build understanding of how genomics influences treatment response and patient experience, integrating pharmacogenomics into routine care to enhance treatment effectiveness and minimise treatment toxicity.

3.4 Minimise financial toxicity for patients and families associated with access to best-practice genomics-informed cancer care.

Awareness and education

Health professionals, consumers and the community have appropriate genomics health literacy to support informed use of genomic technologies and decision-making

Goal

All health professionals are competent in using genomics in cancer care, and the community is empowered to understand the role of genomic testing and genomics-informed care.

Actions

4.1 Embed cancer genomics education into health professional curricula and ongoing professional development to build competence in using genomics in cancer care.

4.2 Develop tools for the specialist cancer workforce and primary care, including Aboriginal Community Controlled Health Services, to support shared decision-making with patients about the use of genomics in cancer care.

4.3 Develop awareness campaigns about cancer genomics, including in community spaces, GP waiting rooms and community pharmacies.

4.4 Co-design resources with Aboriginal and Torres Strait Islander communities, other priority populations and consumer groups, to raise awareness of, and address concerns about, genomic testing and genomics-informed care.

Foundational Objectives

Research and data

Evidence-based and data-driven cancer genomics is incorporated into the health system

Goal

Cancer genomic research and data are representative of population diversity, underpinned by Indigenous Data Sovereignty principles, and used to inform best practice cancer care.

Actions

5.1 Promote equitable access and culturally safe participation in cancer clinical trials incorporating genomics.

5.2 Implement nationally consistent processes to collect, store, share and link cancer genomic data safely and securely.

5.3 Apply Indigenous Data Sovereignty principles to the governance of cancer genomic data across the continuum.

Workforce and models of care

An agile workforce is enabled to adapt to genomics evidence, treatment and technologies

Goal

Development of workforce capability in genomics and cultural safety are prioritised to support sustainable and equitable delivery of genomic cancer care.

Actions

6.1 Upskill all cancer health professionals to offer mainstreamed culturally safe cancer genomic testing, information and support, to meet current and future demand.

6.2 Increase patient and family access to specialist genetic counselling services as part of multidisciplinary cancer care.

6.3 Co-design education for all staff in the specialist cancer sector and primary health sector (mainstream and Aboriginal Community Controlled Health Services) on the role of genomics in cancer control.

6.4 Drive system level changes relating to cultural safety, with education for health services which acknowledges the complex history of genetics and genomics for Aboriginal and Torres Strait Islander people.

6.5 Investigate the potential for oncology outreach services, genetics outreach services and clinical trials to be delivered through local health services to enable more cancer care to happen on Country, or closer to home.

Funding, quality and safety

Genomics is integrated safely, consistently and cost-effectively across the cancer care continuum

Goal

Ethical, legal, financial and regulatory considerations are addressed as part of the design and expansion of best-practice genomics-informed cancer care.

Actions

7.1 Evolve Health Technology Assessment (HTA) methodologies and processes to streamline access to reimbursed genomic testing and treatment.

7.2 Apply relevant national and international policies, laws, and guidance to the design and delivery of genomic cancer care to ensure equity of experience and outcomes.

7.3 Ensure innovative models of care for integrating genomics across the cancer care continuum maintain the standard of high-value culturally safe care.

7.4 Strengthen consideration of ethical issues in relation to use of genomic testing and genomics-informed cancer treatment for all people affected by cancer.

7.5 Develop protocols to ensure Aboriginal and Torres Strait Islander sovereignty over data and samples collected for cancer genomics, particularly when retained in a biobank or data registry.

Aboriginal and Torres Strait Islander themes — Self-Determination — Culture — Capacity Building — Access

Figure 2. Framework on a Page

The Australian Cancer Plan

Cancer outcomes in Australia are among the best in the world, but this is not the story for all Australians. There are significant disparities in cancer outcomes among specific groups in our society.

The Australian Cancer Plan (the Plan) is designed to improve cancer outcomes for all Australians, and particularly for those groups whose health outcomes are poorest. Achieving equity in cancer outcomes will be a fundamental measure of success for the Plan and will align Australia with global calls to improve cancer outcomes for all people.

The National Framework for Genomics in Cancer Control (the Framework) has been developed under the Plan's Strategic Objective to deliver Maximised Cancer Prevention & Early Detection however the Framework applies across the entire cancer care continuum and to all the Plan's Strategic Objectives. The Framework shares the vision of world-class cancer outcomes and experiences for all Australians, as well as the Plan's guiding principles.

- **Person-centred:** the Plan is designed with, and for, all people affected by cancer. This includes people at risk of cancer, people diagnosed with cancer, and their families and carers.
- **Equity-focused:** the need for equity in cancer outcomes and experience is at the centre of the Plan. If the Plan does not 'shift the dial' for people whose outcomes are poorest, it will not be successful.
- **Future-focused:** the Plan addresses both current and future cancer and health trends and challenges, so Australia can take advantage of emerging opportunities to improve cancer outcomes.
- **Strengths-based:** the Plan adopts a strengths-based approach which identifies gaps and issues in the system, and builds on the strengths, opportunities, and the diversity of Australia's population groups and our cancer care system.
- **Evidence- and data-driven:** the Plan is evidence-informed, and promotes better, ongoing use of data to drive, understand, and evaluate the performance of Australia's cancer care system.
- **All cancers:** the Plan addresses issues relevant to all cancer types, with a focus on addressing disparity of experience and outcome.
- **Encompassing the cancer control continuum:** the Plan addresses the whole continuum of cancer care – spanning prevention and early detection, diagnosis, treatment, survivorship care, end-of-life care, and supportive care.
- **Collaborative:** the implementation of the Plan, as with its development, will encourage and involve system-wide, cross-sector, inter-jurisdictional, and national collaboration.

Aim

The use of genomics in cancer care is transforming our approach to cancer prevention, screening, diagnosis and treatment. Genomic medicine[i] and personalised medicine[ii] in cancer care are rapidly evolving with huge potential to transform outcomes for Australians with cancer. The Framework will guide policy to enable equitable access to genomic medicine for all Australians affected by cancer.

Equity

The Framework is underpinned by Cancer Australia's Framework for Health Equity in Cancer Outcomes, which focuses on:

- engaging and empowering priority population groups
- tailoring and co-designing health programs, services, systems, and strategies to accommodate needs
- integrating and enhancing the experience of consumers including their carers and families across the care continuum.

Introduction to genomics across the cancer continuum

Cancer is the leading cause of death and largest contributor to disease burden in Australia.[1] Use of genomics in prevention, early detection, diagnostic methods and personalised treatments all have potential to improve survival rates and reduce the impact of cancer for all Australians.

The term 'genomics' refers to both the study of single genes (genetics) and the study of an individual's entire genetic makeup or genome and how it interacts with environmental or non-genetic factors. In this framework, the terms genomics and genomic testing are used throughout to include genetics and genetic testing, to acknowledge the cross-over of issues between genomics and genetics.

Some Australians are more at risk of developing cancer than others. Genomic testing, which is a type of test that identifies a change in a person's chromosomes or genes, is an emerging opportunity to better identify an individual's risk of certain cancers.[2] For example, the finding of a BRCA gene variant is associated with an increased risk of developing breast, ovarian and other cancers. Identifying individuals at increased risk of developing cancer can support personalised prevention, risk-reducing interventions and earlier detection. This has significant potential to reduce the development of cancer and reduce the risk of later stage cancers, particularly among high-risk individuals, and improve overall survival rates.

After diagnosis of cancer, genomic testing can help determine mutations or variants in genes that drive various cancer behaviours; from how aggressive the cancer may be to whether it is likely to spread and how it will respond to treatment.[3] Genetic and genomic testing technologies are being continually improved and becoming more precise, accessible and cost-effective.

'Omic' technologies include genomics (genes), transcriptomics (mRNA), proteomics (proteins) and metabolomics (metabolites), and these have a broad array of potential applications in oncology.[4] This Framework focuses on genomics, but it is a fast-moving field and the principles of equitable access in this Framework also apply to these related technologies. Other biomarker-driven technologies and treatments will impact cancer care by improving

i 'Genomic medicine' is a medical discipline that involves using a person's genomic information as part of their clinical care.

ii 'Personalised medicine' (also known as precision medicine) uses the knowledge of genetics, including the specific links between genes and some diseases, and between genes and the effectiveness of some medicines or treatments, to predict disease development and to influence decisions about lifestyle choices or to tailor treatment to an individual.

characterisation of tumours, identifying therapeutic targets and informing pre-clinical research models and clinical trials.

By studying genes and their functions, and how they interact with each other and with the environment, genomics may lead to improved understanding of how cancer forms and new ways to prevent, diagnose, and treat cancer more effectively.

Cancer genomics

Cancer is a genetic disease; it occurs when changes in DNA inside cells cause these cells to start growing and dividing uncontrollably. These changes can be inherited from a parent (germline variants) or develop at some point in a person's life (somatic variants).

Inherited cancer predisposition syndromes, such as Lynch Syndrome which increases a person's risk of colorectal, endometrial and other cancers,[5] are caused by a single inherited variant. International research has shown that 10% of adults with cancer have a disease-causing (pathogenic) germline variant,[6,7,8,9,10,11,12,13,14,15] with higher rates found in people with rare and advanced cancers.[7,10,15] As our knowledge improves and the number of genes screened increases, this proportion is likely to continue to increase. For conditions such as Li-Fraumeni syndrome, which increases a person's risk of sarcoma, breast, brain and other cancers,[5] surveillance based on *TP53* variant status enables early detection of cancer which is associated with increased survival.[16,17,18,19]

The combined effect of several small genetic changes which are called single nucleotide polymorphisms (SNPs), can also increase a person's risk of developing cancer. Although the effect of each individual change is small, SNPs can be combined into a single measure of cancer risk, known as a polygenic risk score (PRS, or PGS). PRS is an emerging technology, currently performed in limited National Association of Testing Authorities (NATA) - accredited laboratories in Australia,[20] and testing is still largely experimental.[21]

Knowledge of germline variants can also be used to guide treatment decisions, resulting in better outcomes than standard therapy.[22] For instance, people with BRCA1- or BRCA2-positive breast, ovarian and pancreatic cancers are more likely to respond to medications called poly-ADP ribose polymerase (PARP) inhibitors, such as olaparib.[22,23,24]

Variants that originate within the tumour (somatic variants) drive the development, growth, and invasion of cancer. By testing the tumour for these variants, relevant variant-matched treatments can be identified.[25] This is known as precision oncology.

Cancer Genomics in Adults

The majority of adult cancers (between 75 and 95%) have one or more somatic variants in a cancer-associated gene. Some of these alterations can be targeted with approved or experimental therapies and are known as "actionable variants" [26] (identified in between 27 and 88% of patients).[6,13,14,15,27,28,29,30,31,32,33,34,35,36,37,38,39,40] The identification of actionable variants may inform use of targeted treatments, eligibility for enrolment into genomic clinical trials and may lead to reduced side-effects of treatment and improved survival outcomes. As our knowledge improves, more cancers are found to have actionable variants.

Circulating tumour DNA (ctDNA) are short fragments of DNA shed by tumours that are present in bodily fluids, including blood, urine and cerebrospinal fluid. ctDNA can be detected using a liquid biopsy (generally a blood test) that identifies whether cancer is present, what type of cancer is present, and at what stage.[41] Applications might include detecting and profiling cancers of an unknown primary (CUP) or tumours inaccessible for biopsy, monitoring treatment effectiveness, detecting residual disease after treatment, and screening for cancer in the general population.

An emerging field of interest is cancer vaccines, which can be utilised to prevent or treat cancer. Prophylactic, or preventive, cancer vaccines, such as the human papillomavirus (HPV) vaccine, reduce the risk of particular types of cancers by preventing infections that could lead to cancer.[42] Therapeutic cancer vaccines are used to treat existing cancers by training the immune system to recognise and attack cancer cells.[43] There are many types of therapeutic cancer vaccines in development. These vaccines use a range of technologies including peptides, DNA, RNA, cell-based or viral technology. The technology that has shown the most promise is the mRNA vaccine. This type of vaccine has been widely used to prevent or reduce the impact of COVID-19. One of the key aspects is that these vaccines can be developed rapidly and have shown excellent safety in the clinical trials setting.[44] The first trials looking at mRNA vaccines are underway in cancers including melanoma,[45] and pancreatic cancer.[46]

Cancer Genomics in Children, Adolescents and Young Adults

Approximately 18% of children, adolescents and young adults with cancer are found to carry germline variants associated with hereditary cancer syndromes. Germline variants are more common in solid tumours than blood cancers, and rare cancers are associated with the highest incidence of germline variants.

Many pathogenic variants in early childhood cancers arise years prior to diagnosis and are either inherited from either parent or arise in early embryonic development.[47, 48, 49] In children, adolescents and young adults with newly diagnosed cancers, between 50 and 95% have been found to carry germline or somatic variants, between 16 and 57% of these are variants which inform prognosis and between 25 and 65% may lead to specific genomics-informed treatments.[50, 51, 52, 53, 54]

Equity of outcomes

The rapid advancement of cancer genomics, while beneficial to many populations, has the potential to increase and embed existing inequities. Evidence highlights that access to genomic testing in the cancer context is not currently equitable,[55] and that populations with less access to genomics are the same groups as those that are already impacted by social and cultural determinants of health.[56]

The Australian Cancer Plan (The Plan) considers an intersectional and health equity approach for all priority population groups. This is more than recognising the multiple backgrounds, experiences, and ways people identify. It acknowledges that membership of multiple priority population groups compounds complexity and disparity in accessing equitable and appropriate care. Ten priority population groups are represented in the Plan: *Aboriginal and Torres Strait Islander people, People living in rural and remote areas, People from Culturally and Linguistically Diverse (CALD) backgrounds, Children, People living in low socioeconomic groups, People living with disability, People living with a mental illness, Adolescents and young adults, Older Australians, Lesbian, Gay, Bisexual, Transgender, Intersex, Queer and Asexual (LGBTIQA+) people*.

Populations who face different outcomes and experiences in relation to genomics in cancer care have been identified as part of the development of the Framework:

Aboriginal and Torres Strait Islander people

Every day around five Aboriginal and Torres Strait Islander people are diagnosed with cancer.[57] Aboriginal and Torres Strait Islander Australians have a slightly higher rate of cancer diagnosis and are approximately 40% more likely to die from cancer than non-Aboriginal and Torres Strait Islander Australians.[58] This likely reflects cancer health care that has not been designed to respond to the needs of Aboriginal and Torres Strait Islander peoples affected by cancer.[59] Historical unethical practices in relation to theft and misuse of genetic material, and existing systemic racism and structural discrimination has led to a lack of Aboriginal and Torres Strait Islander community trust in genomic research, which has the potential to further exacerbate inequity.

The strengths of Aboriginal and Torres Strait Islander cultures have continued to evolve despite the ongoing impacts of colonisation, systemic discrimination and intergenerational trauma, including through the Stolen Generations.[60] Research is ongoing to understand the role of epigenetic mechanisms in how intergenerational trauma can affect health outcomes. The enablers include improving cultural safety in cancer services and provision of care closer to home. The barriers include experiences of racism in cancer care and costs of care.[61]

Culturally safe genomics-guided personalised cancer care offers an opportunity to reduce cancer health disparities experienced by Aboriginal and Torres Strait Islander peoples. For this to happen in Aboriginal and Torres Strait Islander communities, services must be adaptive to fit specific community needs, with recognition of the context of historic and contemporary barriers to accessing medical treatments and clinical trial participation.

People living in rural and remote areas

People living with cancer in rural and remote areas experience poorer cancer outcomes compared to those living in metropolitan areas. Barriers to accessing cancer care include distance as well as health workforce shortages in remote and very remote areas.[62,63] Access to new technologies, including genomics, may not be made immediately available in rural and remote Australia, resulting in out-of-pocket costs as people travel to access care. Due to this distance, alternate models of care, including telehealth may be preferred by people living in rural and remote areas.[64]

Children, adolescents and families

Children and adolescents with cancer, and their families, face unique experiences when it comes to the use of genomics in cancer control. The ZERO Childhood Cancer program has demonstrated how access to precision medicine for children with high-risk cancers may be feasible and beneficial for these children, and in 2023, the program was expanded to be made accessible for all children with cancer. The finding of a cancer-causing genetic alteration may have unique considerations for family members, including reproductive carrier screening and early detection of cancers through surveillance.[16]

People from culturally and linguistically diverse (CALD) backgrounds

People from diverse backgrounds experience greater barriers to accessing culturally responsive care and information due to communication barriers, limited support to enable health literacy, and cultural differences.[65] Genomic resources produced in English may not address the needs of people from CALD backgrounds including addressing language barriers, cultural factors, and perceptions of genomics.[66] In turn, this may impact communication channels between patients and health professionals and impact patient understanding when making decisions about genomics in cancer care.

People with rare cancers

Genomics may benefit patients with rare and less common cancers, who might otherwise miss out on tumour-specific clinical trials and treatment pathways. Genomic testing of a person's tumour can inform personalised treatment and clinical trial options. Additionally, the increasing knowledge base for genetic conditions potentially linked to rare and less common cancers may allow for risk-stratified screening programs.

Future state

The Australian Cancer Plan has a ten-year time horizon for achievement of world-class cancer outcomes for all Australians. In line with the Plan, the ideal future system will be one where the following elements of genomics in cancer control are being delivered.

1. Equitable access to genomic testing

Evidence-based genomic testing is available to all Australian cancer patients who would benefit from it. The benefits of genomic testing may include the following:

- confirming, refining or challenging a diagnosis
- determining whether a particular treatment is indicated
- determining the risk of cancer or the expected course of the disease (prognostic value)
- identifying familial cancer risk
- providing information for the person (the value of knowing).

In a future system, mainstreaming[iii] of testing will facilitate equitable access to genomics-informed cancer care and delivery of optimal care as close to home and as safely as possible. The genomic testing (single gene, gene panel or comprehensive genomic profiling) will be undertaken in NATA-accredited laboratories, supported by a skilled multidisciplinary clinical and bioinformatics workforce, and digital and laboratory infrastructure.

2. Equitable access to genomics-guided treatment

Access to genomics-guided medicines is provided to all those who are found to have an actionable variant for whom a matched therapy is available. This is achieved by genomic testing, and the availability of a funded treatment that is biomarker-driven ('tumour agnostic') instead of diagnosis-driven (based on tumour histology and primary cancer location).

Sustainable funding models are in place to support equitable cost-effective access to genomic testing and genomic medicines. These will build on current funding mechanisms, which include Medicare Benefits Schedule (MBS) and Pharmaceutical Benefits Scheme (PBS) items, activity-based funding under the National Health Reform Agreement (NHRA), other state and territory arrangements, and private-payer arrangements.

All efforts to develop new cancer genomic interventions for Aboriginal and Torres Strait Islander peoples are linked to a robust public health systems approach with input and leadership from Aboriginal and Torres Strait Islander people to avoid widening the equity gap.[59]

iii 'Mainstreaming' refers to genomics testing being ordered by clinicians not primarily trained in genetics. In the broader sense it refers competency in genomics across all levels of the health system including oncology health professionals, primary care, nursing and allied health.

Development of the Framework

The development of this Framework was informed by an evidence review of national and international literature and stakeholder engagement, with oversight from a multidisciplinary Expert Advisory Group and Indigenous Governance Group.

The evidence review aimed to:

1. Evaluate the current and potential use of genomics in cancer control.
2. Summarise the ethical, legal, and social implications of cancer genomics.
3. Review models of care for providing genomic testing for prevention and treatment of cancer.
4. Explore the economic costs of cancer genomics to health systems and consumers.

Consultation

Consultation with priority population stakeholders was undertaken in August 2024 via virtual focus groups for children, young people and families, rural and remote, culturally and linguistically diverse, and rare cancer communities. In-person workshops with Aboriginal and Torres Strait Islander community members, health care providers, cancer patients, and community-controlled Aboriginal health service representatives were undertaken across Australia between June to October 2024.

The key themes emerging from consultation with Aboriginal and Torres Strait Islander people and the draft objectives, goals and actions were tested with Aboriginal and Torres Strait Islander stakeholders at a workshop in October 2024, to ensure that perspectives and needs of Aboriginal and Torres Strait Islander people and communities are embedded in the Framework. Key themes are reflected in the 'Framework at-a-glance' diagram. These themes of Self-determination, Culture, Capacity Building and Access are highlighted in the outer circle of the 'Framework at-a-glance' and are embedded across the objectives, goals and actions of the Framework.

- Self-determination
  - Uphold the importance of informed decision-making about genomic testing and personalised cancer care. Ensure Aboriginal and Torres Strait Islander people have self-determination over their biological samples (biospecimens) and genomic data in health care and research.
- Culture
  - Acknowledge the importance of Aboriginal and Torres Strait Islander culture and identity, as it intersects with genomics in cancer care. The intersection of culture and genomics is complex, relating to family engagement in cancer care, family and genomic testing, and family history and genomics.
- Capacity Building
  - Provide education and raise awareness for the Aboriginal and Torres Strait Islander health workforce and communities on genomics in cancer control.
- Access
  - Strengthen enablers and reduce barriers for Aboriginal and Torres Strait Islander people accessing cancer genomic services.

Implementation

Providing equitable personalised cancer care is a shared responsibility that extends beyond the boundaries of any one organisation or the specific domain of the health and social sectors. Achieving the ambitions of the Framework requires sector-wide collaboration and partnerships between governments, the private health sector, consumers, non-government organisations, Aboriginal and Torres Strait Islander stakeholders and leaders, researchers, health professionals, health services, professional colleges and regulators.

The implementation plan will be developed in collaboration with Government and key stakeholders. It will outline ongoing and upcoming activities to achieve the actions within the Framework and identify stakeholders who will lead the implementation activities.

Alignment with overarching Commonwealth policy

In developing the Framework, consideration has also been given to existing Commonwealth policy and programs to avoid duplication, strengthen alignment, and leverage the broader genomics agenda to optimise outcomes.

National Health Genomics Policy Framework 2018 – 2021

The National Health Genomics Policy Framework 2018 – 2021 is being reviewed and updated by the Health Technology and Genomics Collaboration (HTGC) as part of its 2025 workplan, led by the Australian Government Department of Health and Aged Care in collaboration with state and territory health departments.

The aim of this overarching framework is to provide a consistent national and strategic view for integrating genomics into the Australian health system. It aims to address the lack of coordination of activities across Australia and is designed to drive national effort on agreed priorities, and address policy issues and challenges.

On 15 November 2024, the Australian Government announced the establishment of Genomics Australia from 1 July 2025 to provide national leadership and coordination to drive the integration of genomics into the Australian health system. The new national body will support the transition from a research focus to clinical service delivery, where efficiencies and better health outcomes for all Australians can be achieved.

The National Aboriginal Community Controlled Health Organisation (NACCHO) Aboriginal and Torres Strait Islander Cancer Plan

The NACCHO Aboriginal and Torres Strait Islander Cancer Plan was developed for the Aboriginal Community Controlled Health (ACCH) sector to change cancer experiences and create a new foundation for partnership in Australia's national approach to cancer control. To develop this Plan, NACCHO undertook extensive consultations with the ACCH sector and other stakeholders across the country. Consultations highlighted the need to focus on structural reform including sustainable funding, increasing accessibility of services and ensuring mainstream cancer centres are culturally safe and responsive. NACCHO also learned from the lived experience of Aboriginal and Torres Strait Islander individuals, their families and Community leaders.

National Agreement on Closing the Gap

The Australian Cancer Plan and its priority initiatives (including this Framework) support and uphold the priority reforms of the National Agreement on Closing the Gap[67] to enable Aboriginal and Torres Strait Islander people and governments to work together, and to overcome the inequality experienced by Aboriginal and Torres

Strait Islander people so that Aboriginal and Torres Strait Islander people can achieve life outcomes equal to all Australians.

Framework structure

This Framework is comprised of four strategic objectives which span the cancer care continuum:

- Prevention and early detection
- Diagnosis, treatment and clinical trials
- Supportive care
- Awareness and education

The strategic objectives are supported by three foundational objectives which have a genomic-specific lens on wider health system structures and functions:

- Research and data
- Workforce and models of care
- Funding, quality and safety

Strategic Objective 1: Prevention and early detection

Ambition Statement

Genomic testing is integrated into healthcare to reduce cancer incidence and enable early detection.

Goal

Evidence-based genomic testing is accessible to individuals and their families to support personalised prevention, risk reduction and earlier detection of cancer.

Actions

1.1 As the evidence is established, expand access to affordable genomic testing to enable personalised cancer prevention and risk-reducing strategies.

1.2 Give priority to the establishment of evidence for genomic testing in Aboriginal and Torres Strait Islander people and other priority population groups and increase the scope of genomic reference datasets to more accurately reflect the diversity of the Australian population.

1.3 Promote translational research into personalised risk prediction and genomic risk-based cancer screening.

1.4 Embed evidence-based culturally safe genomic testing into national cancer screening programs, to enable risk-stratified screening.

The use of genomic testing can enhance efforts to prevent cancer, and detect it earlier, resulting in improved cancer outcomes. Where the evidence is established, access to genomic testing should be expanded to optimise personalised cancer prevention and risk reduction. This needs to be guided by co-design, with a focus on embedding health promotion and strengthening health literacy to ensure existing inequities are not worsened, and that diverse populations have equitable access to testing and risk-reducing strategies. In Aboriginal and Torres Strait Islander communities, the focus of genomic testing for risk of cancer needs to be framed as part of collective community prevention, rather than simply for the individual.

Genetic testing before a cancer diagnosis enables a person to undertake risk-reducing behaviours, reducing their risk of developing cancer, and enabling diagnosis at an earlier stage through surveillance.[68, 69,70,71] Beyond the individual, cascade testing (i.e. predictive testing in unaffected blood relatives) can enhance the benefits of genetic testing by empowering family members to also undertake risk-reducing interventions and surveillance.

The Human Genetics Society of Australasia recommends that genetic testing in children should only be for conditions that manifest in childhood and where there are options to treat, monitor or prevent the condition.[72] For cancer, this might mean cascade testing, where siblings of children with cancer with identified germline variants are tested to identify their risk of developing cancer.

In the future, genetic markers may be incorporated into personalised risk assessments in risk-stratified population-based cancer screening programs, to better target screening interventions, maximising the benefits and minimising harms of screening.

Explanatory text for actions

1.1 As the evidence is established, expand access to affordable genomic testing to enable personalised cancer prevention and risk reducing strategies.

Germline genomic testing is highly likely to be cost-effective for the prevention and early detection of breast and ovarian cancer, colorectal and endometrial cancer.[73] In families with hereditary cancer predisposition, cascade testing allows for risk-reducing interventions such as risk-reducing medication (chemoprevention), prophylactic surgery, and customised screening for those who carry a cancer predisposition genetic variant, thereby facilitating early detection, and resulting in better outcomes.[68, 69, 70, 74]

In Australia, genetic testing for hereditary cancer syndromes in adults is currently based on agreed criteria that includes personal and family history.[75] Guidance for health professionals ordering genetic testing, including specific heritable pathogenic variants, is available via the digital EviQ resource.[76] However, it has been noted that more than half of adults with cancer with identified germline variants may not have met the clinical criteria for genetic testing prior to their diagnosis.[7,77] Optimal selection criteria (based on evidence of clinical- and cost-effectiveness) for genetic testing for people unaffected by cancer, could improve outcomes by enabling people to undertake preventive actions against cancer risk and surveillance for earlier detection of cancer.[78]

The Australian Government's Population Based Screening Framework sets out the principles of screening programs and informs decision makers about the key issues to consider when deciding whether to introduce a screening program.[79] The Population Based Screening Framework is currently being reviewed to better guide the incorporation of genomics into population screening programs.

Moving forward, as our understanding of the interaction between an individual's germline polygenic variants and certain somatic variants which drive the development and growth of a cancer increases, screening for these polygenic variants will also become increasingly important in personalised cancer prevention and risk-reducing strategies.

1.2 Give priority to the establishment of evidence for genomic testing in Aboriginal and Torres Strait Islander people and other priority population groups and increase the scope of genomic reference datasets to more accurately reflect the diversity of the Australian population.

The diversity of the human genome is not currently reflected in genomic databases which are used to benchmark genomic testing. For minority populations this can potentially reduce the applicability of conclusions of genomic testing and exacerbate existing inequities.[80]

There are multiple reasons underpinning Eurocentric bias within genomic datasets, including European cohorts being in many cases easier and more affordable to access. In Australia, the ongoing effects of colonisation combined with historically poor research practices has created widespread distrust of genomics and genomic researchers, and contributed to a reduction in Aboriginal and Torres Strait Islander participation in research.[81, 82] In this context, Aboriginal and Torres Strait Islander people have historically been excluded from genomic research, leading to downstream exclusion from clinical benefits.[83] A pharmacogenomic study with Tiwi Islander participants identified variants important for informing treatment recommendations, including for cancer therapeutics such as thiopurines and tamoxifen.[84] Given the diversity of Aboriginal and Torres Strait Islander peoples, these findings highlight the importance of broader engagement to produce data relevant for all Aboriginal and Torres Strait Islander Australians.

The establishment of diverse genomic datasets needs to address the systemic barriers that Aboriginal and Torres Strait Islander people encounter when participating in clinical trials and precision medicine research; the potential for data misuse; and provide tangible benefits for Aboriginal and Torres Strait Islander people. Consideration

needs to be given to Indigenous ways of knowing, being and doing, ensuring genomic research reflects values, priorities, and lived experiences of Aboriginal and Torres Strait Islander people rather than being solely defined by non-Indigenous researchers.

Lack of representation in genomic reference datasets is not limited to First Nations Australians. The majority of genomics studies including genome-wide association studies (GWAS) have been conducted in individuals of European descent (86.3%), followed by East Asian (5.9%), African (1.1%), South Asian (0.8%), and Hispanic/Latino (0.08%) populations.[85] The extent to which mistrust plays a role in the underrepresentation of CALD Australians with non-European ancestry in research is less clear than for First Nations Australians.[86] More work such as the OurDNA project,[87] is needed to ensure diverse representation including people affected by cancer in CALD communities. OurDNA, funded under the Government's Medical Research Future Fund (MRFF), is partnering with communities from the Pacific, South-East Asia, the Middle East, and East Africa collect and then securely and anonymously share de-identified genetic data from more than 20,000 diverse participants.

1.3 Promote translational research into personalised risk prediction and genomic risk-based cancer screening.

There is growing interest in offering genetic testing for high-risk germline variants, circulating tumour cells and tumour DNA to asymptomatic people in the general population.

The DNA Screen pilot [88] study offered genetic testing to 10,000 young adults in Australian aged 18 to 40 to demonstrate a proof of concept for population screening for DNA variants that increase the risk of developing certain types of cancer and heart disease, and to provide participants with genetic counselling and support. The two cancer types, hereditary breast and ovarian cancer and Lynch syndrome, were selected as they are relatively common and evidence-based risk-management options are available.[89] The findings of this study suggest that offering combined genomic screening for high-risk conditions to young adults would be cost-effective in the Australian public healthcare system. However, the researchers note that other issues, including psychosocial impacts, ethical and societal issues, and implementation challenges also need consideration prior to population-wide delivery.

Multi-cancer early detection (MCED) tests measure circulating tumour cells and tumour DNA that may identify the presence of multiple cancer types from different organ sites.[90] Tests such as the GRAIL-Galleri-MCED test are proposed as a screening blood test that captures circulating tumour DNA and in combination with machine learning, aim to identify the presence of multiple different cancers and their location. Further research is required to demonstrate the tests' sensitivity, accuracy, impact on cancer-specific mortality, and cost-effectiveness.[91]

Translational research into personalised risk prediction, ensuring it addresses the distinct genomic landscapes of adult and paediatric cancers, would allow the development of the most effective tests, surveillance, and treatment, for every patient.

Translational research is required to demonstrate clinical and cost-effectiveness, accessibility and acceptability of these genomic tests to all Australian communities, including Aboriginal and Torres Strait Islander Australian communities.

1.4 Embed evidence-based culturally safe genomic testing into national cancer screening programs, to enable risk-stratified screening.

In Australia, polygenic risk scores (PRS) are already being used in clinical trials to refine breast cancer risk in people carry a variant in a high-risk gene such as BRCA 1 or 2 and to explain risk in people with breast cancer who do not carry a variant in a high-risk gene. PRS are being increasingly utilised in research for risk refinement in other diverse cancer types. In the near future, it is likely PRS may be used as a tool in conjunction with traditional risk factors for population-based risk stratification and wider genomics-informed cancer care.

Personalised genomic risk information based on a PRS or a known genetic variant which increases risk of cancer

might be used to stratify participants within population cancer screening programs. This may be used to identify individuals who might start screening at an earlier age or be triaged to a different screening technology or a different screening interval. However, several key challenges for population-wide implementation of PRS testing remain.[92]

Australian research includes the Australian Cancer Risk Study which aims to develop cancer risk prediction tools,[93] and an interventional study on the impact of personal genomic risk information on melanoma prevention behaviours, which improved some skin cancer prevention and early detection behaviours in trial participants.[94]

International research has included assessment of a cell-free DNA (cfDNA) blood-based test to improve bowel cancer screening adherence, to detect bowel cancer earlier, and to reduce bowel cancer–related mortality.[95] Other research has considered use of molecular biomarkers in lung cancer screening to improve the selection of individuals undergoing screening, and characterisation of nodules found during low dose computed tomography (CT)-based lung cancer screening.[96]Cultural safety must be embedded at systemic, organisational, and clinical levels through co-design and clear accountability measures. Tailored solutions are essential for populations without a known family history, such as the Stolen Generation survivors and their descendants, to ensure equitable access.

Stakeholder Input

Stakeholders emphasised the role for the Framework to prioritise equity of access to genetic testing and risk-reducing interventions for priority population groups.

Stakeholders highlighted the importance of genomics services that are locally available and supported to facilitate access to culturally safe care.

Stakeholders supported broadening indications for genomic profiling across priority population groups, reducing wait times and out-of-pocket costs to promote early detection.

Stakeholders called for appropriate training of the healthcare workforce, in particular GPs, nurses, and cancer care health professionals, to support communication around genomic testing.

Stakeholder Quotes

> *"Prevention programs. Because if you look at the breast screening, the Beautiful Shawl Project, which is very, very successful and brought a lot of partners together, it's opened up the door for a lot of women to have their screening safely"*- Stakeholder Workshop Participant

> *"For all children who are at increased risk, identify them in a timely manner, then have a trained workforce who can undertake it [genomic testing and treatment] and monitor them for a long time"* - Stakeholder Workshop Participant

> *"The way to improve equity is to really ensure access exists, that the knowledge and the expertise and the professional growth and development of those that can perform the tests have the skills and the expertise to do this"* - Stakeholder Workshop Participant

Aboriginal and Torres Strait Islander people

Stakeholders highlighted that access is fundamental for genomics in cancer control. Challenges such as accessing genomic testing services across geographic locations and different health care providers were identified.

People living in rural and remote areas

Stakeholders identified that there was limited access to genomic testing for people living in rural and remote areas. Stakeholders identified that greater accessibility to genomic testing services is needed to reduce waiting times and out-of-pocket costs for these services for people living in rural and remote areas.

Children, adolescents and families

Stakeholders identified the need for greater access to genomic testing for children, adolescents and young people who are at higher risk of developing cancer due to a genetic predisposition and other factors. This includes establishing indications for genomic testing for adolescent and young adults to acknowledge the unique differences between adolescent and young adult cohorts, and children and adults.

People from culturally and linguistically diverse (CALD) backgrounds

Stakeholders identified that improved communication about genomic testing with individuals from diverse backgrounds is required. This included ensuring accessible and culturally appropriate communication about the role of genomics in identifying cancer risk, and addressing fears and myths.

People with rare cancers

Stakeholders identified the need to reduce waiting times and out-of-pocket costs for genetic testing for people with rare cancers. This includes broadening indications for testing for both germline and somatic variants, and screening for secondary cancers.

Links to the Australian Cancer Plan

- Maximising cancer prevention and early detection - Australian Cancer Plan
- 1.2.1: Deliver cancer prevention and health promotion activities, including healthy lifestyles, immunisation, and population screening participation, co-designed and tailored to a range of settings. - Australian Cancer Plan
- 1.2.4: Undertake ongoing assessment of the evidence for risk-based, cost-effective population cancer screening. - Australian Cancer Plan
- 1.5.3: Implement new, and improve existing, evidenced-based, risk-stratified cancer screening programs. - Australian Cancer Plan
- 1.5.5: Increase access to and uptake of health assessments through Medicare for cancer prevention and early detection for Aboriginal and Torres Strait Islander people. - Australian Cancer Plan
- Aboriginal and Torres Strait Islander people - Australian Cancer Plan

Other relevant links

- Population-based screening framework - Australian Government Department of Health and Aged Care

Strategic Objective 2: Diagnosis, treatment and clinical trials

Ambition Statement

Genomic testing is used to guide diagnostic and treatment decisions to deliver better outcomes for people affected by cancer.

Goal

Genomics technology is incorporated into routine cancer care to inform accurate diagnosis, personalised treatment and access to clinical trials.

Actions

2.1 Enhance access to genomic testing for people affected by cancer where there is evidence of benefit, supported by information and recommendations for management.

2.2 Enable timely access to genomics-informed cancer treatments through Health Technology Assessment (HTA) processes and clinical trials.

2.3 Promote translational research into cost-effective evidence-based genomic tools to support cancer detection, genomics-guided treatments and monitoring for progressive disease or recurrence, and transition from research to routine cancer care.

2.4 Build the evidence for genomics-guided treatments, including through expanded access to appropriate clinical trials, ensuring that the specialist cancer and primary care sectors, including Aboriginal Community Controlled Health Services, are engaged to support equitable patient participation.

Genomic testing can be used to guide diagnostic and treatment decisions, resulting in better outcomes than standard diagnostic tools and therapies. Routine integration of genomic testing based on evidence of benefit is critical to give patients access to the best treatments for their cancer, maximising treatment effectiveness and reducing treatment toxicity. Genomics is a rapidly evolving field, and as evidence of benefit and cost-effectiveness emerges, clinical applications will evolve.

Capacity to deliver genomics-informed cancer care is highly dependent on the support of a high-quality, timely genomics pathology service and bioinformaticians to support the information flows, safe storage and interpretation of results.

Genomics clinical trials accelerate knowledge gathering and provide options for cancer patients when other therapies have failed or are yet to be approved. Currently, access to matched therapies in children and adults in Australia is largely facilitated through clinical trials.[39,40,97] For children in Australia, integration of research with clinical care has enabled widespread uptake of new technologies and data-informed care. Efforts now need to focus on bringing these therapies from the research setting into routine clinical care, while retaining the benefit of this integration.

Nationally coordinated and consistent investment into genomic cancer research and clinical trials is key to ensuring Australia's future research agenda is focused on patient priorities. There is scope to increase the

translation of genomic research into clinical outcomes and improve the rate of research commercialisation, raising the potential for sustainable funding for cancer research. There are also opportunities to expand genomic research on critical topics and to priority population groups, and to bring the national research agenda in line with areas that deliver the most value. Optimally, implementation would be supported by collection of genomic data 'in practice' to progress research. This aligns with the HTA Review report recommendation on improving use of real-world data (RWD) and real-world evidence (RWE).

Explanatory text for actions

2.1 Enhance access to genomic testing for people affected by cancer where there is evidence of benefit, supported by information and recommendations for management.

The benefits of genomic testing may include the following:

- confirming, refining or challenging a diagnosis
- determining whether a particular treatment is indicated
- determining the risk of cancer or the expected course of the disease (prognostic value)
- identifying familial cancer risk
- providing information for the person (the value of knowing)

As evidence of effectiveness becomes available, the use of gene expression profiling (GEP) testing and tumour genomic profiling may maximise treatment effectiveness and reduce treatment toxicity, leading to improved cancer outcomes.

GEP tests are an emerging technology which measures the pattern of specific genes expressed in cancer cells. GEP tests can be used at diagnosis to add to clinical and pathology information to assess risk of disease recurrence and to guide management.

Genomic profiling of the tumour allows identification of potential actionable somatic variants in the tumour that may enable a specific matched treatment or enrolment to a clinical trial. Research to date on comprehensive genomic profiling and matched therapies has been largely focused on advanced cancers in patients with poor prognoses. Patients with advanced cancer where a targetable variant is identified report reduced distress.[98]

Genomic profiling in adults with cancer has revealed that between 31 and 48% patients have at least one matched therapy available to them; however only a third of these patients receive it, generally due to deterioration of their condition.[6,13,14,15,27,28,30,32,34,35,36,37,39,40,97,99,100,101,102,103,104] Despite lower profiling success rates in cancers of unknown primary,[30] these patients are more likely to have actionable variants (91%), and an available matched therapy (41%).[30] Patients and health professionals view tumour profiling positively,[105,106,107] although patient willingness to pay decreases when out of pocket costs are higher.[108]

While the types of genetic variants in childhood cancers differ from variants in adult cancers,[109] the benefit of matched therapy remains. Between 25 and 65% of children have therapeutically relevant somatic variants in their cancers. In addition, 16-18% of children, adolescents and young adults with cancer carry germline variants, of whom only 40% had a significant family history of cancer.[110]

The Australian Rare Cancers Portal connects patients and treating medical practitioners with sub-specialists on genomic profiling and precision medicine, regardless of their location. It can assist a person diagnosed with a rare cancer and their treatment team with management guidelines, interpretation of genetic reports, streamlining access to national rare cancer specialists including by telehealth or multidisciplinary groups and identification of relevant clinical trials or other research opportunities.

2.2 Enable timely access to genomics-informed cancer treatments through Health Technology Assessment (HTA) processes and clinical trials.

The HTA Review found that a coordinated and standardised approach to collecting real-world data (RWD) and real-world evidence (RWE) is required to understand how health technologies are being used and are performing in the real world.

This is particularly important for genomics-guided treatments where RWD represents a promising alternative to the traditional evidence-based medicine approaches that are based on the use of randomised clinical trials.[111]

Matched therapies have the potential to improve survival for those who receive them, compared to unmatched therapies.[33] Research on the use of genomic-matched therapies compared to traditional treatments found that, of children found to carry a targetable variant, 13-67% received the matched therapy, usually via a clinical trial or compassionate access.[54, 112, 113, 54,114,115,116] Models like ZERO and PrOSPeCT offer examples of centralised clinical and research expertise, linked to patient services across state boundaries, providing national access. However, there are significant risks that lack of access to genomics technologies and novel (high-cost) genomics-informed treatments will contribute to further disparity in cancer outcomes, including for priority population groups.

One barrier to timely adoption in clinical practice includes the challenges of demonstrating the efficacy of genomics-informed strategies. Optimally, outcome studies should be planned as part of a pipeline from discovery to implementation. Similar to phased drug studies, outcome studies can prospectively validate a discovery, demonstrate the efficacy of genome-informed strategies, or assess the effectiveness of implementation.[111]

The HTA Review recommends (Recommendation 38) that the Australian Government:

- support the development of further guidance on methods for assessing tumour agnostic therapies, informed by approaches that have been used by the Pharmaceutical Benefits Advisory Committee (PBAC); models proposed in academic literature; models adopted in other jurisdictions and consultation with patients, clinicians and industry.
- support the development of guidance on the assessment and appraisal of genomic technologies and gene therapies for HTA decisions in Australia.

In an optimal, equitable system, access to evidence-based, cost-effective genomics-guided medicines would be provided to all those who are found to have an actionable somatic or germline variant for whom a matched therapy is available. This would be achieved by equitable access to genomic testing, and availability of funded treatment that is biomarker driven ('tumour agnostic') instead of diagnosis driven (based on tumour histology and primary location).

2.3 Promote translational research of cost-effective evidence-based genomic tools to support cancer detection, genomics-informed treatments and monitoring for progressive disease or recurrence, and transition from research to routine cancer care.

Circulating DNA (ctDNA) through liquid biopsies has shown promise in clinical trials as a potential prognostic biomarker, a substitute for biopsy (particularly useful in cancers of unknown primary), and monitoring response to treatment and recurrent disease. Monitoring for residual disease after treatment already has an increasingly established role in both predicting treatment outcome and early relapse detection in many haematological malignancies.[117, 118, 119]

Less research has been conducted on ctDNA in cancers in children, adolescents and young adults, but preliminary evidence suggests high levels of correlation with tissue sample variants, the ability to detect ctDNA in 70% of recently diagnosed and 80% of advanced cancer patients, and the genomic profile in serial samples is consistent with treatment responses.[73]

Translation of research findings into practice can also be cost-saving for the system. Economic modelling of next-generation sequencing (NGS) compared to polymerase chain reaction (PCR) biomarker testing strategies among patients with metastatic non-small cell lung cancer demonstrates that NGS is associated with greater cost savings compared to PCR. This is driven by more rapid results, shorter time to appropriate therapy initiation, and minimised use of inappropriate therapies while awaiting and after test results.[120]

The ZERO Childhood Cancer Program commenced in 2017 as a precision medicine program to guide potential treatment options, initially for children with poor outcome cancers and expanded in 2023 to all children diagnosed with cancer in Australia. Program data shows that 70% of children who received ZERO-recommended personalised treatment showed a complete or partial response, or disease stabilisation. The PrOSPect Study, led by Omico, provides genomic profiling to adults with advanced or incurable cancers and identifies potential matches for patients to clinical trials with new targeted therapies.

Research is needed to demonstrate the best use of genomic tools in practice across the cancer care continuum, with a focus on priority populations to ensure equity of access. Research also plays a pivotal role to establish the efficacy and safety of new treatments. There are limits to the current level of research translation into clinical practice.[121] Transition from a research study to national best practice standard of care requires consideration of sustainability and a co-ordinated approach to integration of genomic technologies into the Australian health system.

2.4. Build the evidence for genomics-guided treatments, including through expanded access to appropriate clinical trials, ensuring that the specialist cancer and primary care sectors, including Aboriginal Community Controlled Health Services, are engaged to support equitable patient participation.

There is a mismatch between the study populations participating in oncology clinical trials and the composition of the target cancer population.[122] Increasing access to genomic clinical trials can be achieved via: broadening eligibility requirements for trials, simplification of trial procedures, community outreach through patient navigators, decentralisation of clinical trial procedures, telehealth, and funding to offset costs of travel. Regulatory requirements can mandate that trial sponsors enrol diverse study populations and ensure that ethics and governance review prioritise equity and inclusivity.

Facilitation of access is the joint responsibility of the specialist cancer and the primary care sectors who have direct contact with priority populations who are not well served by the current genomic clinical trial ecosystem. Building of culturally safe service pathways, research sites and clinical trials staff is critical for success. Other levers include making funding contingent on actions that promote targeted enrolment and data sharing with Aboriginal and Torres Strait Islander communities, and engagement in partnerships to design clinical trials to enable participation in a trial closer to home. Promotion of Aboriginal and Torres Strait Islander-led genomics and cancer research, and capacity building for Aboriginal and Torres Strait Islander researchers will contribute to delivery of this action.

For example, Cancer Australia is delivering the Partnerships for Cancer Research grant program [123] to strengthen cancer research to improve the health outcomes of Aboriginal and Torres Strait Islander people and to build capacity of the Aboriginal and Torres Strait Islander cancer research sector.

Stakeholder Input

Stakeholders emphasised the role of the Framework to support equitable access to, and participation in, genomics-related clinical trials for priority population groups.

Stakeholders supported research and clinical trial risk minimisation activities such as accessible and culturally appropriate informed consent and data privacy arrangements.

Stakeholders called for funding to address costly treatment and out-of-pocket expenses, workforce training and outreach services.

Stakeholder Quotes

> *"Most mob don't get to treatment, that option [of a clinical trial] is not there because it's a late-stage diagnosis"* - Stakeholder Workshop Participant.

> *"Our sample sizes in cancer are getting smaller because we're getting better at refining all our subtypes. But we're just not seeing the increase in the services and the activities we need in our environment to action more trials"* - Stakeholder Workshop Participant

Aboriginal and Torres Strait Islander people

Stakeholders highlighted the importance of informed choice and informed consent, as well as self-determination over data and research. The ongoing effects of systemic racism on Aboriginal and Torres Strait Islander peoples and communities, and the historical harms associated with health research and genomics were also identified.

People living in rural and remote areas

A lack of clinical trials and genomics-trained health workforce was identified by people living in rural and remote areas. Stakeholders identified the burden of additional costs involved in accessing genomics-informed cancer treatment, including treatment not covered on the PBS, travel and accommodation away from home, and time off work.

Children, adolescents and families

Stakeholders identified the need for rapid processes to access genomic clinical trials for children, adolescents and young people. This includes establishing genomics-specific funding pathways and supporting the retention of existing clinical trials staff. Stakeholders highlighted the unique differences between child, adolescent and young adult patients and stressed that genomic testing indications should reflect these differences.

People from culturally and linguistically diverse (CALD) backgrounds

Improved communication about clinical trial involvement and informed consent processes was identified by individuals from diverse backgrounds. This included ensuring access to bi-lingual interpreters, simple language to ensure understanding and minimise any potential risks, including financial.

People with rare cancers

Stakeholders identified the need to improve access to clinical trials for people with rare cancers. This includes addressing sample size requirements for clinical trials. Stakeholders suggested supporting existing clinical trial infrastructure, funding, testing and referral pathways whilst building workforce sub-specialities in clinical trials

Links to the Australian Cancer Plan

- **World class health systems for optimal care - Australian Cancer Plan**
- 3.2.2: Develop a national framework for networked, distributed comprehensive cancer care, to facilitate provision of services as close as safely possible to where patients live. This will include the role of Comprehensive Cancer Centres to enhance patient outcomes, strengthen transparency and accountability, and drive continuous improvements for all patients across the network regardless of where the care is

provided. -

- 3.2.3: Implement innovative, evidence-based and cost-effective models of care for people living with and beyond cancer. - Australian Cancer Plan
- 3.5.2: Establish an Australian Comprehensive Cancer Network (ACCN) to ensure connectivity and sharing of expertise between Comprehensive Cancer Centres, other cancer services, regional hospitals, community and primary care. The establishment of an ACCN will increase equity of access across services for all patients, deliver cancer care close to home, and monitor evidence-based system performance. - Australian Cancer Plan
- **Strong and dynamic foundations - Australian Cancer Plan**
- 4.2.2: Ensure targeted and innovative research investment into areas of unmet and emerging need; and improve clinical trial design and equitable access. - Australian Cancer Plan

Other relevant links

- National Pancreatic Cancer Roadmap – Cancer Australia

Strategic Objective 3: Supportive care

Ambition Statement

The experience of people affected by cancer is improved by genomics-informed cancer care.

Goal

Genomics is used to optimise treatment response and minimise treatment toxicity, and people affected by cancer receive psychosocial care when needed.

Actions

3.1 Provide tailored clinical and psychosocial support to people affected by cancer, according to their genomic profile and individual preferences.

3.2 Provide Aboriginal and Torres Strait Islander people and other priority populations with holistic navigation support and wrap-around culturally-informed personalised genomic cancer care.

3.3 Build understanding of how genomics influences treatment response and patient experience, integrating pharmacogenomics into routine care to enhance treatment effectiveness and minimise treatment toxicity.

3.4 Minimise financial toxicity for patients and families associated with access to best-practice genomics-informed cancer care.

Supportive care includes the management of physical and psychological symptoms and side effects across the continuum of the cancer journey from diagnosis through treatment to post-treatment care.[124]

Genomics can be useful in determining likely response to treatment and risk of side-effects, including in palliative care and at end-of-life. Pharmacogenomics is the study of how genomic variation is associated with the clinical effectiveness and safety of drugs.[125] Pharmacogenomics can influence the treatment of the cancer as well as management of the effects of the cancer and comorbidities. A genomic perspective on the safety of anti-cancer treatments may assist in placing the person with cancer, rather than the disease, at the centre of their care.[126] When access to the genetic profile of the patient and/or their tumour is possible, a better evidence-based decision on the type and intensity of treatment can be achieved for an individual patient. This has particular value when multiple regimens that are similarly effective are available, and both efficacy and safety can be maximised through testing for genetic biomarkers.[126]

Approximately 60% of patients with advanced cancer have been reported to be taking medications with pharmacogenomic interactions.[125] Pharmacogenomic testing using specific markers is currently used as standard of care for B-cell acute lymphoblastic leukemia in children. However, broader testing, including other markers such as "sensitivity" to certain supportive care treatments, or emerging genomic markers of risk of certain toxicities, has not yet been implemented in the clinical practice. There are ongoing research initiatives to implement this in paediatric oncology.[110]

Explanatory text for actions

3.1 Provide tailored clinical and psychosocial support to people affected by cancer, according to their genomic profile and individual preferences.

A genomic test result with a germline variant indicative of a hereditary cancer syndrome, has implications for the individual and their family, including an increased risk of cancer and opportunity for risk-reducing interventions. Patients who are found to be at increased risk of cancer due to hereditary cancer syndromes report clinically relevant distress, anxiety and depression compared to the general population. These patients need tailored support to cope with decisions regarding risk-reducing surgeries, communication with their families, and childbearing.[127]

Following a cancer diagnosis, an informative genomic test which demonstrates a somatic or germline variant could represent the opportunity for a targeted therapy, or entry to a clinical trial. Honouring individual preferences is an essential principle of supportive care which aims to maintain quality of life, and to ensure that people with cancer can achieve maximum benefit from their anticancer treatment. It is important to recognise that the experience of children affected by cancer can be significantly improved by genomics-informed cancer care. Both clinical and psychological support tailored to their unique circumstances is essential to ensure the best possible outcomes.[128]

In addition to coping with the worry and stress brought about by their diagnosis, people with cancer and their families often face the stresses of physically demanding cancer treatments, and potential disability, fatigue, and pain associated with treatment.[129] People affected by cancer need to be supported with information access when the person is ready for it, in their own time and space. Tailored support also makes excellent cancer care possible by easing the emotional burden for people with cancer and caregivers, and improving communication between people with cancer and caregivers about disease and prognosis.[124]

Genetic counselling can support people affected by cancer to provide informed consent for testing and to understand and adapt to the medical, psychological, familial and reproductive implications of their diagnosis.[130]

3.2 Provide Aboriginal and Torres Strait Islander people and other priority populations with holistic navigation support and wrap-around culturally-informed personalised genomic cancer care.

Personalised care across the cancer continuum can be complex to navigate, including decisions about genomic testing to enable personalised prevention, targeted cancer therapies, treatment dosages to optimise treatment responses and participation in genomic clinical trials. At each step, there may be a need to resolve access barriers, including strengthening health literacy, addressing financial costs, and providing culturally safe services and infrastructure within local Communities.

Stakeholders highlighted the need for more Aboriginal and Torres Strait Islander staff in cancer care, and their critical role in implementing safe and effective cancer care to Aboriginal and Torres Strait Islander communities, noting the support required for this workforce. The emerging role of Aboriginal and Torres Strait Islander-led care coordinators in assisting patients to access cancer care was highlighted as an important element of the workforce. Navigators need genomics literacy and support to upskill, including training pathways for Aboriginal and Torres Strait Islander Liaison Officers and for Aboriginal Health Workers and Practitioners, and for Aboriginal and Torres Strait Islander nurses to have genomics qualifications recognised in the clinical skill sets.

Aboriginal and Torres Strait Islander Communities have experienced positive outcomes when dedicated positions are funded to give people a single point of contact to support them to understand their care pathway, answer questions and regularly check in. In line with the NACCHO Aboriginal and Torres Strait Islander Cancer Plan, navigation support can build on other elements of a culturally safe environment, including self-determination

over data capture and research participation, translation and resources in appropriate language, a support person such as Aboriginal Liaison Officer, and a full description of the treatment or intervention to enable decision-making based on other obligations and priorities, and to ensure informed choice and informed consent.

A broad, open focus for supportive care will consider the impact of cancer on the patient's family members and the broader Community. Wrap-around care means that the individual's needs are met promptly, compassionately and without judgment. An individual's needs may vary across the life course, and this should be considered when planning wrap-around services tailored towards the individual and their family. Needs might be material, such as transport, cost and time; psychological such as mental health services; or social such as a need for legal services.[131]

Underserved segments of the population, such as those with a culturally and linguistically diverse background, those living in rural and remote areas, and those in lower socioeconomic or medically underserved communities, can benefit from patient navigation that provides culturally appropriate and relevant education and assistance.[132] Priority populations would benefit from patient navigation designed to support their understanding of genomics in cancer care and facilitate the accessibility of the health system for provision of genomics-informed care.

3.3 Build understanding of how genomics influences treatment response and patient experience, integrating pharmacogenomics into routine care to enhance treatment effectiveness and minimise treatment toxicity.

Many patients with cancer will experience drug toxicity and suboptimal response rates during their disease course. Both the patient's germline genetic variations and tumour-specific somatic variants may affect drug response in terms of both efficacy and toxicity. Ongoing research and information sharing is needed in parallel with development of novel treatments to ensure that genomics can be used to optimise treatment response, improving quality of life and impacting survival.

Pharmacogenomics has been proposed to individualise cancer therapy, for example by optimising the dose of treatment or avoiding treatment side effects. In current clinical practice, pharmacogenomics is used for example, in opioid treatment;[133] in considerations for radiation therapy in patients found to carry a p53 variant;[134] and modifying chemotherapy regimens in people with a genetic variation in the DYPD gene.[135]

Pharmacogenomics is evolving as a potential tool to improve patient care in pain management. Pain management medications are associated with a significant variability in the analgesic response based on variations in genes that are involved in the drug metabolism.[136] Pharmacogenomic Decision Support Systems in Palliative Care have been trialled to better support opioid drug selection and dose adjustments in chronically ill patients with advanced illness.[137]

Although certain germline pharmacogenes, such as TPMT, UGT1A1, and DPYD, have been recognised for decades, potential dosing implications for new anticancer medications, including targeted therapies, as well as the discovery of additional genetic variants has the potential to impact treatment responses.[138] In some cases, knowledge of these interactions and their implications for patient care has translated to the clinical sphere such as label guidance, drug prescribing guidelines and recommendations by international societies and governing bodies.

Pharmacogenomics also has an increasing role in predicting target drug levels in various supportive care medications, such as anti-fungal agents, allowing individualised dosing regimens to be developed.[139]

Ongoing research and information sharing is needed in parallel with development of novel treatments to ensure that genomics can be used to optimise treatment response.

Optimal use of pharmacogenomics involves a unique multidisciplinary collaboration between oncologists, laboratory scientists, bioinformatics, pharmacists and clinical pharmacologists. A co-ordinated approach to

integration of pharmacogenomics into routine care must be taken to ensure testing availability and equity of access to best practice care.

3.4 Minimise financial toxicity associated with access to best-practice genomics-informed cancer care.

Financial toxicity is the combined impact of out-of-pocket costs, direct and indirect costs (including travel and accommodation to receive treatment) and the changing financial circumstances of an individual and their household due to cancer, from diagnosis to treatment, supportive and palliative care and throughout survivorship.[140] It is important that there is 'visibility' of what these costs are and that patients are directed to appropriate resources.

The psychological and practical burden of financial toxicity on people affected by cancer is large and financial considerations can also influence decision-making around testing and treatment and lead to increasing inequities and poorer cancer outcomes.[141]

Out-of-pocket expenses relate to the requirement for upfront payment for services outside of in-patient care and high-cost tests and treatment. Typically, new treatments are expensive and therefore may not be accessible to all individuals prior to registration and government reimbursement, due to their financial cost. HTA processes aim to ensure that government-funded therapies and services are clinically effective, safe and cost-effective for particular patients, while also giving regard to ensuring the efficient use of health care resources and the financial impact that paying for the care might put on a society.

There are significant risks that lack of access to publicly subsidised genomics technologies and novel (high-cost) genomics-informed treatments will contribute to not only the financial toxicity faced by patients, but also to further disparity in cancer outcomes, including for priority population groups.

State funded Patient-Assistance Travel Schemes (PATS) provide patients (and eligible carers) with financial assistance towards the costs involved in travelling to, and staying near, approved medical specialist services that are not locally available.[142] However, each jurisdictional scheme is different, and costs are rarely fully covered.[5] In addition, most jurisdictional schemes are reimbursement models which means patients must have access to funds to pay for travel and accommodation upfront, which can create a significant financial barrier.[143]

Stakeholder Input

Stakeholders emphasised the role for the Framework to deliver psychosocial support for priority population groups.

Stakeholders supported workforce training to improve knowledge of supportive care services, data collection to better understand supportive care needs, and importantly the role of hope.

Stakeholder Quotes

> *"The big thing about personalised cancer care that we see in our remote communities is that family is so important as being part of it all"* - Stakeholder Workshop Participant

> *"Make sure that you know the support and survivorship needs. It really needs to be integrated or properly funded to allow people to manage their cancer"* - Stakeholder Workshop Participant

> *"Bringing faith leaders into those conversations [end of life care planning] can be very important for some communities, some families and for some individuals and there will need to be some awareness and education for them"* - Stakeholder Workshop Participant

"It's already expensive for our rural and remote area patients to access. Add screening and treatments and then you're adding the cost if they're not subsidised for these panels that are only going to increase in the future" - Stakeholder Workshop Participant

Aboriginal and Torres Strait Islander people

Stakeholders placed importance on Aboriginal and Torres Strait Islander culture, how culture intersects with cancer care and the cultural considerations needed for genomics in cancer control. For Aboriginal and Torres Strait Islander stakeholders the cancer care journey is not individualistic, and rather family should be part of the journey. Stakeholders also highlighted the desire of Aboriginal and Torres Strait Islander peoples and communities to receive care on Country, or closer to home.

People living in rural and remote areas

Psychosocial impacts of genetic testing and treatment were identified by people living in rural and remote areas. Stakeholders identified the importance of understanding both country-wide and state-wide variation in supportive care needs of people affected by cancer and that rural and remote is not a homogenous group.

Children, adolescents and families

Stakeholders identified the need to manage expectations of symptom burden and quality of life in families with children, adolescents and young people diagnosed with cancer. This includes establishing specialised support services for those at increased risk while promoting the role of hope in supportive care.

People from culturally and linguistically diverse (CALD) backgrounds

Stakeholders emphasised the role of the family in supporting people with cancer and how health professionals could be more accommodating of this role. Engaging with community and faith leaders was also identified by individuals from diverse backgrounds to build trust and facilitate palliative care discussions.

People with rare cancers

Stakeholders identified the psychosocial support needs for people with rare cancers, and their families and carers. This includes supporting health professionals, such as cancer specialists, nurses and GPs, to provide clear and simple information on future risk for the individual and, where hereditary, their family.

Links to the Australian Cancer Plan

- World class health systems for optimal care - Australian Cancer Plan
- 3.2.4: Develop and refine integrated care models to maximise access to high-quality, timely and evidence-based palliative and end-of-life care, including voluntary assisted dying. - Australian Cancer Plan

Strategic Objective 4: Awareness and education

Ambition Statement

Health professionals, consumers and the community have appropriate genomics health literacy to support informed use of genomic technologies and decision-making.

Goal

All health professionals are competent in using genomics in cancer care, and the community is empowered to understand the role of genomic testing and genomics-informed care.

Actions

4.1 Embed cancer genomics education into health professional curricula and ongoing professional development to build competence in using genomics in cancer care.

4.2 Develop tools for the specialist cancer workforce, and primary care including Aboriginal Community Controlled Health Services, to support shared decision-making with patients about the use of genomics in cancer care.

4.3 Develop awareness campaigns about cancer genomics, including in community spaces, GP waiting rooms and community pharmacies.

4.4 Co-design resources with Aboriginal and Torres Strait Islander communities, other priority populations and consumer groups, to raise awareness of, and address concerns about, genomic testing and genomics-informed care.

Awareness and education is built on a foundation of building and maintaining public trust in genomic medicine, with a focus on improving support for individuals and families to make informed choices about cancer genomic testing.[144] Educational activities for patients need to highlight not only the benefits but also the risks associated with testing such as discovery of incidental findings which may have implications for the individual and their family. Education on the subsequent choices and avenues for treatment and supportive care that may or may not be available is also important. It is also critical that education and awareness approaches are co-designed and tailored to the needs and preferences of different population groups to ensure that materials are linguistically and culturally appropriate.[121]

The amount of information available on genetics and genomics is growing rapidly, and it is important that people seeking cancer-related genomics information are able to locate and identify reliable health information. Health professionals must always be a trusted source of cancer genomics information. A commitment to increasing capacity and capability in cancer genomics recognises that many types of health professionals will need varying degrees of cancer genomic literacy as the implementation of personalised medicine proceeds. The role of genomics in cancer control will continue to evolve as research and technology progresses. This will necessitate new service delivery models, and assessment of the roles and skills that are required now and in the future by each of the different professional groups.

Explanatory text for actions

4.1 Embed cancer genomics education into health professional curricula and ongoing professional development to build competence in using genomics in cancer care.

Quality, evidence-based education on a large scale is required to build a healthcare workforce that can competently practice genomics-informed cancer medicine.[145] To make decisions regarding cancer genomic testing and to address patient-initiated questions, health professionals need to first and foremost accurately understand the role of genomics in cancer control and the basic premises of tests and treatments.

All cancer specialists need an understanding of clinically-applicable genomics, including being sufficiently proficient in interpreting genomic results to communicate and make decisions about testing and treatment. They should leverage support from genomic specialists and genetic counsellors and accessible guidelines and resources, to select tests and interpret genetic results.[146] Increased capability in primary care is required to communicate risk, manage the assessment and prevention strategies for people identified with high-risk genetic profiles, including follow-up testing, surveillance and advice.

Undergraduate and postgraduate curricula for all health professionals and ongoing professional development, need to include genomic and precision medicine to support the delivery of trauma-aware healing-informed personalised cancer care. This must include an understanding of the particular sensitivities that genomics poses for Aboriginal and Torres Strait Islander people, due to historical distrust, poor research practices and ethical concerns. Education and training materials also need to reflect current workforce development needs, including specific needs of Aboriginal and Torres Strait Islander Health Workers and Practitioners.

4.2 Develop tools for the specialist cancer workforce, and primary care including Aboriginal Community Controlled Health Services, to support shared decision-making with patients about the use of genomics in cancer care.

Shared decision-making involves a collaborative conversation between patients and health professionals to co-create a care plan that makes sense intellectually, practically and emotionally. Tailored resources to aid decision-making, and enable informed consent to genomic testing, should be available in all care settings, including primary care.

Genomic testing to identify whether a patient has a germline variant associated with a hereditary cancer syndrome represents a prime example of the importance of shared decision-making in cancer care.[147] Genetic test results can impact personal circumstances, including ongoing surveillance, prophylactic procedures and fertility interventions, which can be stressful and should only be undertaken with informed consent. Shared decision-making should be supported by tools that assist in conveying the relevant information. For primary care and ACCHS, tools should build on the variety of testing and screening tools available for genetic cancer risk assessment in general practice, to provide culturally safe resources for communicating genomic information and the implications for the patient's and their family's care.

4.3 Develop awareness campaigns about cancer genomics, including in community spaces, GP waiting rooms and community pharmacies.

Awareness campaigns can broaden public knowledge about genetic factors and cancer risk. They can also reinforce the importance of early or different approaches to screening for people at high risk of cancer due to hereditary factors.[148]

Positive factors associated with greater awareness of genetic tests include higher education, higher motivation for cancer information, greater belief in genetics influencing cancer risk, and higher medical literacy.[149]

Use of GP waiting rooms to disseminate health information is a well-established practice in Australia. Extending awareness campaigns into more community focused spaces has potential to overcome some of the barriers to genomic health literacy by bringing cancer genomic information to the individual, rather than depending on the individual to seek it out.

4.4 Co-design resources with Aboriginal and Torres Strait Islander communities, other priority populations and consumer groups, to raise awareness of, and address concerns about, genomic testing and genomics-informed care.

Co-design is an enabling factor for patient-centred care. One example of this in practice is co-design of Cancer Australia's Our Mob and Cancer website, developed for Aboriginal and Torres Strait Islander people by Aboriginal and Torres Strait Islander people. The site includes information about cancer from trusted sources; it is impartial, non-commercial, evidence-based and respects culture and traditions. This approach should be extended to develop genomics-focused resources that meet the needs of Aboriginal and Torres Strait Islander Australians and other priority population groups. This includes supporting people living with a disability to be actively involved in providing informed consent in genomic care decisions.

The development of *Key Principles and Best Practices for Co-Design in Health with First Nations Australians* was co-designed by Cancer Australia and led to a set of six key principles and twenty-seven associated best practices for co-design in health with First Nations Australians. The principles were: First Nations leadership; Culturally grounded approach; Respect; Benefit to community; Inclusive partnerships; and Transparency and evaluation.[150]

Aboriginal and Torres Strait Islander Ways of Working are reflected in co-designed guidance from the RACGP-NACCHO Partnership Project. The key domains of Ways of Working framework are: community related; the context in which we work; relational values; and work values (informing project outputs).[151]

Aboriginal Health workers, particularly in the primary health sector, require education in the role of genomics in cancer care, as well as health equity. An understanding of commonly held community miscomprehensions, concerns and hesitancy around genomics provides an opportunity for promotion of the role of genomics in improving personalised prevention, risk reduction and treatment options, and ultimately reducing disparity in cancer experience and outcomes.

Strategies for community empowerment, including though whole-of-community activities, and community-led place-based health promotion, could raise awareness in the community about genomics in cancer control. Workshops and face-to face approaches support effective communication of potential benefits and are an avenue for those with lived experience to share their stories with community. Aboriginal and Torres Strait Islander led media could lead public engagement, utilising radio network, podcasts, audio book, picture book, with an emphasis on hearing from people's experiences.

Stakeholder Input

Stakeholders emphasised the role for the Framework to support improving genomics literacy across priority population groups.

Stakeholders supported tailored communication delivered using simple language across multiple channels such as print, radio, television, web pages, videos and podcasts.

Stakeholders called for education and training of the healthcare workforce to facilitate awareness and decision-making around genetic testing and treatment for people affected by cancer.

Stakeholder Quotes

"And I think...having information for the community before people get sick [is important] because I'm assuming that the shock can wipe out a lot of rational thought about what you should be doing"- Stakeholder Workshop Participant

"People will take action to take care of their families, as early detection and prevention. But the communication and the approach need to be tailored" - Stakeholder Workshop Participant

"I want to mention around the multi-channel approaches for communication, we do quite a lot of stories and interviews in language, podcasts, videos" - Stakeholder Workshop Participant

Aboriginal and Torres Strait Islander people

Stakeholders identified the importance of building capacity of community members and health care workers to engage with genomics in cancer control. The need for health workers, particularly in the primary health sector, to be educated in the use of genomics in cancer control, as well as health equity was identified. Stakeholders also described the importance of engaging with community and effectively communicating.

People living in rural and remote areas

The importance of involving community members, local Elders and healthcare champions in awareness campaigns was identified by people living in rural and remote areas. Stakeholders suggested adopting simple language and local terminology in genomics communication that specifically targets families and advocacy groups.

Children, adolescents and families

Stakeholders identified the need to inform families of children, adolescents and young people about genomics-informed care soon as possible after diagnosis. This includes improving access to genomic information materials, especially in digital and easy to read formats.

People from culturally and linguistically diverse (CALD) backgrounds

Improving communication with both the GP and cancer team was identified by individuals from diverse backgrounds. This included the importance of bilingual educators, addressing family members and taking time to communicate complex information. Stakeholders from diverse backgrounds also supported the use of plain language and personal stories across print, visual and audio media while highlighting the need for tailored community information materials in language.

People with rare cancers

Stakeholders identified the important role of decision aids in supporting people with rare cancers, and their families and carers, to make decisions about genomics. Improving genomic literacy education for health professionals, such as oncologists, GPs and nurses, was suggested as a way to facilitate clear communication.

Foundational Objective 1: Research and data

Ambition Statement

Evidence-based and data-driven cancer genomics is incorporated into the health system.

Goal

Cancer genomic research and data are representative of population diversity, underpinned by Indigenous Data Sovereignty principles, and used to inform best practice cancer care.

Actions

5.1 Promote equitable access and culturally safe participation in cancer clinical trials incorporating genomics.

5.2 Implement nationally consistent processes to collect, store, share and link cancer genomic data safely and securely.

5.3 Apply Indigenous Data Sovereignty principles to the governance of cancer genomic data across the continuum.

The Australian Cancer Plan has a two-year goal of *nationally agreed frameworks for collection and reporting of comprehensive cancer data, and implementation of new technologies into routine cancer care*, with a focus on research priorities that drive innovation and fast-track opening of cancer clinical trials in Australia. The National Framework for Genomics in Cancer Control is complementary to the actions of the Australian Cancer Plan under this goal, including the development of a National Data Framework to improve accessibility, consistency and comprehensiveness of integrated data assets. Ownership of genomic data is a complex issue legally and practically and is particularly salient for First Nations people, given the history of extractive research practices in communities where knowledge is taken from its immediate context for the benefit of a highly specialised group of outsiders. Extractive research typically fails to uplift, represent, or reflect the experiences of the communities from which it was derived.[152] Some First Nations communities have experienced harms from genetic research that traces human origins or interprets results in ways that stigmatise groups which has contributed to fears around use of genomics to validate or discredit identity.[153]

The diversity of the human genome is not currently reflected in genomic databases and there is the potential for cancer genomics to worsen existing health disparities. One example is less reliable risk stratification for healthcare in underrepresented groups.[154] Genomic research to date has disproportionately focused on populations of European ancestry, and as a result, Aboriginal and Torres Strait Islander peoples are underrepresented in reference datasets used for implementation of genomic technologies.

The National Agreement on Closing the Gap[67] Priority Reform 4 aims to ensure that data and information is shared and made available to Aboriginal and Torres Strait Islander people, to enable communities to access and use locally-relevant data and information to set and monitor implementation efforts to close the gap. Building Indigenous Data Sovereignty (IDS) principles and Aboriginal and Torres Strait Islander governance into research is required to protect Aboriginal and Torres Strait Islander rights and collective interests.[59]

Structural barriers to genomic testing and research include systemic racial, socioeconomic and knowledge barriers to testing. Populations with less access to genomics are the same communities already experiencing significant negative effects of social determinants of health.[59]

Explanatory text for actions

5.1 Promote equitable access and culturally safe participation in cancer clinical trials incorporating genomics.

Targeted research in priority areas and improved identification of priority patient groups within research data is needed to support access to clinical trials for all Australians with cancer.

In Aboriginal and Torres Strait Islander communities there is an awareness that familial and genetic risk factors are likely to be important, but remain understudied in Aboriginal and Torres Strait Islander populations.[155] Inclusive approaches to clinical trial design can reflect practical considerations, such as larger clinical rooms to accommodate extended family and kin, and travel support and accommodation for more than one care person attending appointments with the patient.

Inclusion can also be reflected in: clinical trial protocols that recognise higher rates of co-morbidities of Aboriginal and Torres Strait Islander people and dictate inclusion criteria accordingly, use of telehealth in trials to support Aboriginal and Torres Strait Islander participation on Country, or as close to home as possible, and a strong understanding the philosophies of holistic health and wellbeing and the role of Aboriginal and Torres Strait Islander knowledge, values, beliefs, cultural needs and health history in decision-making processes about treatment and ongoing care.[156]

Aboriginal and Torres Strait Islander people, particularly those living in rural and remote communities, will require additional support and engagement to improve their access to clinical trials. Access to clinical trials is limited for rural and remote Australians, adding to the current health inequity between rural and metropolitan populations. The Australian Teletrial Model was developed by the Clinical Oncology Society of Australia to bring clinical trials "closer to home". In 2020, the Australian Teletrial Program was funded to expand and support the uptake of the model across six Australian states and territories.[157]

5.2 Implement nationally consistent processes to collect, store, share and link cancer genomic data safely and securely.

The Australian Cancer Plan emphasises that optimal cancer care and a high-performing system relies on access to, use of, and sharing of comprehensive health and cancer data across all settings. Effective use of these data is currently hindered. The National Cancer Data Framework sets the strategic direction for the collection, management, use and ongoing development of comprehensive and consistent cancer data. Integration of linked patient-level genomic and clinical data into the cancer data ecosystem and other population level data collections, can be used to inform clinical decisions across the cancer care continuum, to provide a comprehensive view of outcomes across patient groups and be a valuable knowledge base for future research. The alignment between both the National Cancer Data Framework and National Framework for Genomics in Cancer Control and other existing and planned national health data initiatives, is key to enabling nationally consistent processes and integration of cancer genomic data to inform person-centred, equitable genomic and data-driven cancer care.

National genomic data strategies will also guide the use of cancer genomic data into the future. The National Approach to Genomic Information Management (NAGIM) outlines principles for managing genomic and health data for clinical and research use. Cancer genomic data generated across different jurisdictions, healthcare systems and research organisations require a unified approach for collection, management and use. Australian Genomics has developed the NAGIM implementation plan for the consideration of governments. It focuses on delivery of a learning healthcare system, with genomic data, knowledge and outputs flowing interactively through clinical care and research. A genomics learning healthcare system sees cycles of new genomic knowledge moving from '*bedside to bench, and bench to bedside' for the mutual benefit of healthcare systems, research and patients*.[158]

5.3 Apply Indigenous Data Sovereignty principles to the governance of cancer genomic data across the continuum.

The CARE Principles reflect the crucial role of data in advancing Aboriginal and Torres Strait Islander innovation and self-determination. The Global Indigenous Data Alliance (GIDA)[159] formulated the CARE Principles to address a clear deficit in FAIR regarding Aboriginal and Torres Strait Islander peoples' rights and interests. 'CARE' is the acronym for: Collective Benefits, Authority to control, Responsibility, Ethics. The CARE Principles complement the existing FAIR Principles, which require data to be findable, accessible, interoperable and reusable. While the FAIR Principles are about making it easier to share and reuse data, the CARE Principles ensure that data is used ethically.[160] Implementation of the CARE Principles in practice would include the development of protocols within both research and health systems to enable return of Aboriginal and Torres Strait Islander biospecimens.

The Maiam nayri Wingara Indigenous Data Sovereignty Principles[161] assert that, in Australia, Aboriginal and Torres Strait Islander peoples have the right to:

- Exercise control of the data ecosystem including creation, development, stewardship, analysis, dissemination and infrastructure.
- Data that are contextual and disaggregated (available and accessible at individual, community and First Nations levels).
- Data that are relevant and empower sustainable self-determination and effective self-governance.
- Data structures that are accountable to Indigenous peoples and First Nations.
- Data that are protective and respects our individual and collective interests.

IDS principles apply to all people who identify as Indigenous. While it is not possible to apply these principles retrospectively, all new genomic datasets need to give due consideration to their implementation. This includes data governance structures where Indigenous representation is present and empowered to exercise true decision-making power.

Links to the Australian Cancer Plan.

- Australian Cancer Plan – Cancer Australia – Strategic Objective 4 – Strong and Dynamic Foundations

Foundational Objective 2: Workforce and models of care

Ambition Statement

An agile workforce is enabled to adapt to genomics evidence, treatment and technologies.

Goal

Development of workforce capability in genomics and cultural safety are prioritised to support sustainable and equitable delivery of genomic cancer care.

Actions

6.1 Upskill all cancer health professionals to offer mainstreamed culturally safe cancer genomic testing, information and support, to meet current and future demand.

6.2 Increase patient and family access to specialist genetic counselling services as part of multidisciplinary cancer care.

6.3 Co-design education for all staff in the specialist cancer sector and primary health sector (mainstream and Aboriginal Community Controlled Health Services) on the role of genomics in cancer control.

6.4 Drive system level changes relating to cultural safety, with education for health services which acknowledges the complex history of genetics and genomics for Aboriginal and Torres Strait Islander people.

6.5 Investigate the potential for oncology outreach services, genetics outreach services and clinical trials to be delivered through local health services to enable more cancer care to happen on Country, or closer to home.

As genomics increasingly becomes part of routine cancer care, all health professionals, not just genomics specialists, need to have a working understanding of its relevance and the potential to impact the diagnosis, treatment and management of cancer patients and their families. Historically, cancer genomics has not been part of broader oncology training,[162] and genetic testing has been offered through tertiary genetic services where results are discussed with a genetic counsellor or clinical geneticist.[163] The Human Genetic Society of Australasia (HGSA) has identified core capabilities in genetics and genomics for medical graduates including knowledge of the pathogenesis of cancer and common inherited cancer predisposition syndromes.[130]

Sustainable funding of the health system is interlinked with workforce supply and demand. Health economic analysis of the upskilled clinician model of genomics-informed care has shown cost-effectiveness, largely due to reduced number of clinical genetics appointments. A multi-disciplinary team (MDT)- based mainstreamed approach is a preferred model for integrating genetic counselling to ensure equitable access to genomics-informed cancer care. In some circumstances related to complexity of clinical need, specialist genetic expertise is required as a part of the multidisciplinary team.

Explanatory text for actions

6.1 Upskill all cancer health professionals to offer mainstreamed culturally safe cancer genomic testing, information and support to meet current and future demand.

Competency in genomics is required across all levels of the health system including primary care, specialist medical, nursing and allied health. Health professionals must receive the education and training they need to build genomic capability in cancer care. This 'mainstreaming' will permit patients to receive timely, culturally safe genomics-informed care and accommodate increased demand that is unlikely to be met through clinical genetics services. Upskilling in genomics should be built into health, medical and nursing education curricula, and within ongoing professional development requirements for health professionals, with specialist support from clinical geneticists and genetic counsellors. Effective referral pathways to genetic counsellors, when needed, will allow genetic counsellors to work at the top of their scope of practice, applying their skills and expertise to provide tailored clinical and psychosocial support, including for complex patients and families.

6.2 Increase patient and family access to specialist genetic counselling services as part of multidisciplinary cancer care.

Genetic testing has traditionally been offered through tertiary genetics services, where patients received pre-test counselling either in-person or via telehealth and results are discussed in a subsequent appointment by a genetic counsellor or clinical geneticist.[163] However, the increasing demands on genetics services are outweighing the capacity of the current workforce in Australia[164, 165, 166] and globally.[166]

The workforce shortage combined with the increased demand for genomic testing has driven the development of alternate models of care. One such model is 'mainstreaming', which involves integrating genomic testing into routine care in non-genetics specialties, either through embedding a genetic counsellor in the cancer care team or upskilling non-genetics health professionals.[167, 168, 169] Genetic counselling is generally recommended before any genetic testing for inherited cancer risk and to support patients who require access to genetic testing for treatment decisions.[170]

To date, mainstreaming of cancer genomic testing has predominately been facilitated by embedding genetic counsellors in oncology clinics or upskilling cancer specialists to offer testing. Both models have been shown to improve access to and utilisation of genomic testing and be feasible, cost-effective and to provide significant benefits to patients, including and reduced time from cancer diagnosis to test results, improved continuity of care, and high levels of decisional satisfaction.[73, 169]

Access to genomics-informed cancer care has also been shown to depend on awareness of genetic services in primary care, especially for priority populations.[171] Culturally appropriate genomic literacy resources and flexible service delivery models are especially critical to support Aboriginal and Torres Strait Islander communities and reduce access barriers.[172]

6.3 Co-design education for all staff in the specialist cancer and primary health sectors (mainstream and Aboriginal Community Controlled Health Services) on the role of genomics in cancer control.

The role of primary care professionals, especially GPs and community health nurses is important at all stages of cancer care from prevention to palliation. Primary care professionals provide continuity of care and a link to appropriate specialist care.[173] Many GPs are unclear about the difference between germline and somatic testing, and report a lack of confidence in this area, with little formal genetics training received.[174] Understanding the relevance of genomic testing to practice is regarded by GPs as a barrier to integration into care. Education that can be accessed by practitioners without cost or travel is essential. The Gen-Equip project, funded by the European Union is an example of effective education in genetics for primary healthcare in Europe to improve patient care.[175] Primary care providers need support via point-of-care tools (such as the Evi-Q General practitioner referral guidelines for cancer genetics assessment)[176] and co-designed culturally relevant resources for the primary care workforce in the Aboriginal community-controlled health sector.

Tailored education for health professionals in the specialist cancer sector is also important for the delivery of

optimal genomics-informed cancer care. Expansion of accessible, online training such as the 'Mainstreaming genetic testing for oncology teams: A focus on genetic testing in cancer patients' developed by The Human Genetics Society of Australia (HGSA) and the Clinical Oncology Society of Australia (COSA) Cancer Special Interest Group (SIG) is the first step in a wider approach to delivering mainstreaming.[177]

6.4 Drive system level changes relating to cultural safety, with education for health services which acknowledges the complex history of genomics and genetics for Aboriginal and Torres Strait Islander people.

Culturally safe cancer programs such as the Beautiful Shawl Project, can provide a collaborative, Community-led initiative providing safe and empowering breast screening experiences for Aboriginal and Torres Strait Islander women as an alternative to traditional screening. The project provides customised screening shawls to Aboriginal and Torres Strait Islander women that are culturally appropriate, familiar and beautiful to wear during their breast screen.

Cultural safety reflects work undertaken by health professionals to create supportive and accessible care environments for Aboriginal and Torres Strait Islander patients. It reflects both the physical environment, such as consideration of privacy in service layouts and rethinking use of cancer 'bells' often sourced from Aboriginal and Torres Strait Islander Mission buildings, and promoting understanding and empathy surrounding the different priorities and family and community obligations of Aboriginal and Torres Strait Islander people and how these considerations impact on cancer care and clinical trials.

Cultural considerations for cancer genomics span the importance of incorporating traditional healing, to the unique priorities and obligations of Aboriginal and Torres Strait Islander peoples, and the cultural importance of biospecimens. For Aboriginal and Torres Strait Islander people the cancer care journey is not individualistic, and rather family should be part of the journey.

Cancer genomics services and clinical trials can be designed for inclusivity of Aboriginal and Torres Strait Islander families. Examples include use of larger size of clinic rooms to accommodate multiple family members and travel support schemes which allow for more than one support person to accompany the patient.

6.5 Investigate the potential for oncology outreach services, genetics outreach services and clinical trials to be delivered through local health services to enable more cancer care to happen on Country, or closer to home.

The accessibility and costs of different types of cancer care such as genetic testing, standard cancer care and clinical trials can be addressed via different models of outreach care. These models of care should also address challenges for Aboriginal and Torres Strait Islander patients accessing quality, timely care across geographic locations and different health care providers. Care on Country refers to the desire of Aboriginal and Torres Strait Islander people and communities to receive care on Country, or closer to home.

New models of care are needed to enable equitable, consistent access to high-quality genetic health services and clinical trials. Innovative models such as telephone-based counselling, tele-chemotherapy models and enhancing the role of primary care and community health services in genetic counselling are required to address increasing demand and access to underserved communities, especially those in in rural and remote areas.

Links to the Australian Cancer Plan.

- Australian Cancer Plan – Cancer Australia – Action 5.2.2: Build on existing capability of the primary care workforce to collaboratively and sustainably support the needs of consumers.

Foundational Objective 3: Funding, quality, and safety

Ambition Statement

Genomics is integrated safely, consistently and cost-effectively across the cancer care continuum.

Goal

Ethical, legal, financial and regulatory considerations are addressed as part of the design and expansion of best-practice genomics-informed cancer care.

Actions

7.1 Evolve Health Technology Assessment (HTA) methodologies and processes to streamline access to reimbursed genomic testing and treatment.

7.2 Apply relevant national and international policies, laws, and guidance to the design and delivery of genomic cancer care to ensure equity of experience and outcomes.

7.3 Ensure innovative models of care for integrating genomics across the cancer care continuum maintain the standard of high-value culturally safe care.

7.4 Strengthen consideration of ethical issues in relation to use of genomic testing and genomics-informed cancer treatment for all people affected by cancer.

7.5 Develop protocols to ensure Aboriginal and Torres Strait Islander sovereignty over data and samples collected for cancer genomics, particularly when retained in a biobank or data registry.

There are many safety and quality implications for genomics-based cancer care. Integration of genomics in the health system relies on timely and appropriate referrals to multidisciplinary health professionals, use of germline and/or somatic testing where indicated, accurate interpretation of test results to inform interventions and/or treatments, and provision of clear culturally appropriate information for patients and families.[178]

Value-based healthcare is the equitable, sustainable and transparent use of the available resources to achieve better outcomes and experiences, for every person at all stages of their health journey.[179] Increasing value also means identifying and communicating unwanted variations in care, and measuring outcomes, patient experience and resource use.[180] High-value cancer care is complex with multimodal treatments such as surgery, chemotherapy, radiation therapy, targeted therapy and immunotherapy. The Institute of Medicine identifies six elements of value in cancer care: effectiveness, safety, patient-centeredness, efficiency, timeliness of therapy, and equity, all of which can be positively influenced by genomics.[181]

Explanatory text for actions

7.1 Evolve Health Technology Assessment (HTA) methodologies and processes to streamline access to reimbursed and up-to-date evidence-based genomic testing and treatment.

Genomic medicine and precision medicine are rapidly evolving with huge potential to transform outcomes for Australians with cancer. While cancer research is delivering innovative genomics-informed care, there is often a lag between evidence of benefit and reimbursement. There is increasing patient and clinician expectation of access to genomic technology as standard of care such as access to genomics-guided medicines for all those who are found to have an actionable variant where a matched therapy is available. This may be achieved via genomic profiling, and funded treatment that is biomarker driven ('tumour agnostic') instead of diagnosis driven (based on tumour histology). Current approaches to decision-making within HTA focus on tumour histology, rather than adopting a tumour agnostic approach. The HTA Review identifies strategic approaches to enabling contemporary, fit for purpose public funding for genomic testing and precision medicine.

7.2 Apply relevant national and international policies, laws, and guidance to the design and delivery of genomic cancer care to ensure equity of outcomes.

Incorporating genomics into cancer control in the Australian health care system takes place against a background of broad ethical, legal, social and cultural issues (ELSCI) which vary across the cancer continuum. ELSCI issues relevant to cancer genomic testing include equity of access, family considerations, legal considerations, data and cyber security, and consent processes.

Genomic information has unique properties such as being predictive of future health, potentially generating findings beyond the scope of an initial test, the possibility of inducing stigma or discrimination, and being relevant to other family members, that differentiate it from other kinds of testing and other non-cancer domains of health care.

Legal considerations include privacy and confidentiality, and genetic discrimination. In September 2024 the Australian Government announced the intention to enact legislation that will ban the use of adverse predictive genetic test results in life insurance underwriting.[182] Previously, the potential for genetic discrimination had discouraged people from undertaking genetic testing to inform them of their cancer risk, limiting their potential for early detection and risk-reducing interventions.[183]

Digital genomic data contain especially sensitive types of information, which underscores the need for secure electronic storage and transfer. Cyber security in healthcare is an ongoing ELSCI consideration for delivery of safe and quality genomics-informed cancer care.[184]

The breadth of these issues in relation to genomics reinforce the need for application of relevant national and international policies, laws, and guidance to the design and delivery of genomic cancer care to ensure equity of outcomes.

7.3 Ensure innovative models of care for integrating genomics across the cancer care continuum maintain the standard of high-value culturally safe care.

Implementation of new models of care must align with nationally agreed standards of safety and quality and ensure best practice patient management. Clinical care needs to be supported by high quality genomics pathology services, performed in NATA-accredited laboratories providing time- critical results to guide clinical decision-making and underpinned by secure and comprehensive data sharing, including with cancer registries.

Mainstreaming[iv] of testing facilitates equitable access for patients and delivery of optimal care as close to home as possible as part of routine care.

A culturally safe workforce considers power relations, cultural differences and the rights of the patient, and encourages workers to reflect on their own attitudes and beliefs. Cultural respect is achieved when individuals feel safe and cultural differences are respected.[185]

7.4 Strengthen consideration of ethical issues in relation to use of genomic testing and genomics-informed cancer treatment for all people affected by cancer.

Ethical issues in cancer genomics include: the appropriate use of genomic tests in high- and average-risk people; genomic testing of children; familial implications of genetic information; management of unactionable mutations, incidental findings and future discoveries for the individual; protection of genetic privacy; informed consent processes to enable the collection, storage and use of personal genomic data, including potential future use of data; and cultural and social contexts.[186]

Use of genomic information in cancer prevention and early detection has specific ethical considerations particularly related to the idea of personal responsibility for health and the role of the individual in minimising their risk through behaviours.[187]

Ethical considerations in relation to family communication relate to who is responsible for communicating genetic information to extended family, noting that family members are generally considered to have an ethical duty to communicate genomic information that will be relevant to other family members. There is ongoing debate over whether health professionals also have this duty and that doing so is legally defensible in at least some jurisdictions in Australia.[187]

7.5 Develop protocols to ensure Aboriginal and Torres Strait Islander sovereignty over data and samples collected for cancer genomics, particularly when retained in a biobank or data registry.

The Australian National University's National Centre for Indigenous Genomics (NCIG) is leading best practice in return of blood samples collected from First Nations communities for research purposes. Samples were originally collected in the late 1960's and many of the original study participants whose blood was sampled have since passed away. Repatriation of samples has been built upon a model of strong consent and self-determination, with NCIG undertaking consultation and development of equivalent partnerships.[188] The NCIG model could inform wider protocols to ensure prospective sovereignty over samples collected for cancer genomics, with clear distinctions for samples being returned after clinical use, and samples being returned after research use.

The Lowitja Institute's Indigenous Data Sovereignty (IDS) Readiness Assessment and Evaluation Toolkit[189] was developed to evaluate IDS principles and practices in action within research and academic organisations, and as a resource for Aboriginal and Torres Strait Islander communities and organisations to identify IDS in practice. The Toolkit assesses and evaluates how Aboriginal and Torres Strait Islander people and communities' priorities, values and practices are incorporated in the processes that control, develop, use, maintain and protect the data that pertains to Aboriginal and Torres Strait Islander people. The Toolkit provides a model for First Nation's sovereignty over data collection and storage for cancer genomics.

iv 'Mainstreaming' refers to genomics testing being ordered by clinicians not primarily trained in genetics. In the broader sense it refers competency in genomics across all levels of the health system including oncology health professionals, primary care, nursing and allied health.

Links to other policies and reports.

- National Health Genomics Policy Framework and Implementation Plan | Australian Government Department of Health and Aged Care
- The Health Technology Assessment Policy and Methods review

Acknowledgements

Cancer Australia wishes to acknowledge the many individuals and organisations who contributed to the development of this Framework. In particular, the contribution of the Expert Advisory Group, chaired by Professor Chris Karapetis; the Indigenous Governance Group; The University of Queensland (Frazer Institute), The Australian National University (National Centre for Aboriginal and Torres Strait Islander Genomics) and Dr Aideen McInerney-Leo, who conducted the evidence review to inform this Framework; and Adjunct Professor Jacinta Elston, Kelly Trudgen, Professor Alex Brown and Dr Justine Clark who led the Aboriginal and Torres Strait Islander engagement.

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Glossary

Term	Description
Aboriginal Community Controlled Health Services (ACCHS)	Non-government, not-for-profit primary health care services initiated and operated by the local Aboriginal community to deliver holistic, comprehensive and culturally responsive health care. ACCHS are incorporated, based in an Aboriginal community, and governed by a majority Aboriginal board which the local community elects.
Aboriginal and/or Torres Strait Islander Health Workers	Aboriginal and/or Torres Strait Islander Health Workers (ATSIHWs) are health professionals who provide flexible, holistic and culturally sensitive health services to Aboriginal and Torres Strait Islander communities to achieve better health outcomes and better access to health services for Aboriginal people.
Aboriginal and Torres Strait Islander Health Practitioner	An Aboriginal and Torres Strait Islander Health Practitioner is a primary health care professional providing high-quality, culturally-safe, clinical care services to Aboriginal and Torres Strait Islander people and communities. They are independent practitioners who work alongside and collaboratively with other clinicians including doctors, nurses, midwives, allied health and oral health practitioners in a range of settings.
Actionable mutation or variant	DNA change that, if detected in a patient's tumour, would be expected (or predicted) to affect a patient's response to treatment.
Antigen	Any substance that causes the body to make an immune response against that substance.
Cancer mutational signatures	Characteristic patterns of genetic variants found throughout the genomes of cancer cells, arising from disruptions in DNA repair processes.
Cancer of unknown primary	A metastatic cancer (cancer that has spread) where the starting point where the cells first started growing (the origin or primary site) cannot be determined.
Cancer Vaccine	A form of immunotherapy that can help educate the immune system about what cancer cells "look like" so that it can recognise and eliminate them.
Chromosomes	Located in the centre of the cell. Each person typically has 46 chromosomes, with one copy inherited from each parent. The role of chromosomes is to carry genes.
Copy Number Variants (CNVs)	Sections of the genome are repeated or deleted (>1,000 bases in size).
Circulating tumour DNA (ctDNA)	DNA shed by tumour cells into the bloodstream which can serve as a proxy for tumour genomic sequencing.

Term	Description
Cultural safety	Health consumers are safest when health professionals have considered power relations, cultural differences, and patients' rights. Culturally safe practice is the ongoing critical reflection of health practitioner knowledge, skills, attitudes, practicing behaviours, and power differentials in delivering safe, accessible, and responsive healthcare free of racism. Cultural safety is defined not by the clinician but by the health consumer's experience – the individual's experience of the care they are given and their ability to access services and to raise concerns. For Aboriginal and Torres Strait Islander people cultural safety is determined by Aboriginal and Torres Strait Islander individuals, families, and communities.
Depth of coverage	The number of times that a given nucleotide in the genome has been read using genomic sequencing.
DNA	Composed of nucleotides, DNA carries the genetic information essential for the growth and functioning of organisms.
Driver variants	Variants/mutations which drive the development, growth, and invasion of a cancer.
ELSCI	Ethical, legal, social and cultural issues
Epigenetics	Epigenetics describes the way cells switch on (express) or switch off genetic information. There are a number of epigenetic factors which play a role in the way genetic information is switched on. Epigenetics does not change the genetic information (DNA code) but can affect the way this information is read by the cell. The changes occur as a natural process of development and tissue differentiation, and can be altered in response to environmental exposures such as diet, exercise, drugs, and chemicals. These changes can affect a person's risk of disease and may be passed from parents to their children.
Exon	Exons are coding sections of an RNA transcript, or the DNA encoding it, that are translated into protein. Exons can be separated by intervening sections of DNA that do not code for proteins, known as introns.
Financial Toxicity	The distress or hardship arising from the financial burden of cancer treatment.
Gene	Section of DNA that codes for a protein. Can be further divided into subsections called exons (coding region) and introns (non-coding region). The gene is considered the basic unit of inheritance. Genes are passed from parents to offspring and contain the information needed to specify physical and biological traits.

Term	Description
Gene expression	Gene expression is the process when cells convert the instructions in DNA into a function. This mostly occurs from the transcription of RNA molecules that code for proteins or non-coding RNA molecules that serve other functions. Gene expression can be thought of as an "on/off switch" to control when and where RNA molecules and proteins are made and as a "volume control" to determine how much of those products are made. The process of gene expression is carefully regulated, changing substantially under different conditions and cell types.
Gene expression profile (GEP)	Information about all of the genes in a cell or tissue that are making messenger RNA. Messenger RNA molecules carry the genetic information that a cell needs to make proteins. A gene expression profile may be used to help diagnose a disease or condition, such as cancer. It may also be used to help plan treatment, find out how well treatment is working, make a prognosis, or predict whether cancer will come back or spread to other parts of the body.
Gene fusion	Genomic rearrangements lead to the fusion of two genes and subsequently abnormal protein production.
Genetic markers	A specific sequence of DNA at a known location on a chromosome. There are many genetic markers on each chromosome. Genetic markers and genes that are close to each other on a chromosome are usually inherited (passed from parent to child) together. This may help researchers find a gene near a marker that may cause a certain disease or trait within a family. Examples of genetic markers are single nucleotide polymorphisms (SNPs) and microsatellites.
Genomic profiling	A method of testing that uses a sample of tissue, blood, or other body fluid to learn about all the genes in a person or in a specific cell type, and the way those genes interact with each other and with the environment. Genomic profiling makes it possible to find out what changes are occurring in the gene (biomarkers) and inform personalised treatment that targets the specific cancer's features.
Genome	The set of genes within an organism.
Genomics	Genomics is the study of genes and other genetic information, their functions, how they interact with each other and with the environment, and how certain diseases, such as cancer, form. This may lead to new ways to prevent, diagnose, and treat cancer.
Genomic medicine	A medical discipline that involves using a person's genomic information as part of their clinical care.
Genetic testing	A type of test that identifies changes in chromosomes, genes, or proteins. These are usually one or a few genes at a time.

Term	Description
Genomic testing	A laboratory method that looks for genetic changes across all the DNA (including the entire set of genes) in a person or in a specific cell type or tissue. Genomic testing is done on samples such as blood or saliva to identify inherited genetic changes that may increase the risk of developing a specific disease or condition, such as cancer, or on a tumour sample to detect all the genetic changes in the cancer cells. This may help to understand how quickly the tumour is likely to grow and spread, and how well it may respond to certain types of treatment.
Genome-wide association studies (GWAS)	Identifies single nucleotide polymorphisms (SNPs) responsible for certain genetic traits across the genome by comparing the frequency in large groups of affected and unaffected individuals.
Genomic sequencing	Sequencing process that shows each letter of the genetic code for a specific section of DNA or RNA, mostly performed using next generation sequencing.
Genotyping	The process of determining the DNA sequence, referred to as the genotype, to detect the presence or absence of specific variants.
Germline variant or germline mutation	A genomic variation that is present from conception and is inherited or can arise for the first time in an individual (de novo variant).
Health professional	Includes medical, nursing and allied health professionals who provide cancer care services across the cancer control continuum. Health professionals work in the community (see also *Primary health care*) and in hospital settings.
Hereditary cancer syndrome	Caused by an inherited germline variant which increases an individual's risk of developing certain tumours, often at a younger age.
HTA	Health Technology Assessment
ICER	Incremental cost-effectiveness ratio (the difference in costs divided by the difference in outcomes).
Intron	Introns are noncoding sections of an RNA transcript, or the DNA encoding it, that are spliced out before the RNA molecule is translated into a protein.
Liquid biopsy	A genetic sample extracted from blood that can include ctDNA, circulating tumour cells, protein biomarkers and cell-free RNA.
Matched tumour	Both tumour and unaffected samples (e.g. blood or saliva) are collected and tested simultaneously to determine whether variants are germline or somatic.
MBS	Medicare Benefits Schedule

Term	Description
Microsatellite Instability	Microsatellites are short sets of repeated DNA that are not present in the corresponding germline DNA, which can contribute to genetic instability in cells.
MSAC	Medical Services Advisory Committee
Next generation sequencing (NGS)	Technique used for DNA and RNA sequencing and consequently the detection of genetic variants.
OOP costs	Out-of-pocket costs
Panel testing	Involves sequencing the exons of a specific group of genes, ranging from tens to hundreds of genes.
PARP Inhibitor	**p**oly-**A**DP **r**ibose **p**olymerase (PARP) is an enzyme that helps damaged cells repair themselves. PARP inhibitors are a type of targeted cancer drug.
Pathogenic variant or pathogenic mutation	An alteration of the DNA code that affects the quantity or quality of protein produced and can increase the risk of particular types of disease.
PBAC	Pharmaceutical Benefits Advisory Committee
PBS	Pharmaceutical Benefits Scheme
People affected by cancer	This includes people at risk of cancer, people diagnosed with cancer, and their families and carers.
Polygenic risk score (PRS)	A numerical assessment to summarise an individual's genetic susceptibility to a particular trait or disease (such as cancer), based on many genetic markers across the genome.
Personalised medicine	Personalised medicine (also known as precision medicine) uses the knowledge of genetics, and genomics including the specific links between genes and some diseases, and between genes and the effectiveness of some medicines or treatments, to predict disease development and to influence decisions about lifestyle choices or to tailor treatment to an individual.
Precision oncology	Selection of cancer treatments based on the DNA signature of an individual patient's tumour.

Term	Description
Primary health care	Primary health care is usually the first contact an individual with a health concern has with the health system. Primary health care covers health care that is not related to a hospital visit, including health promotion, prevention, early intervention, treatment of acute conditions, and chronic condition management. Primary health care services are delivered in settings such as general practice, community health centres, allied health practices, Aboriginal Community Controlled Health Services and via technologies such as telehealth and video consultations. Primary health care professionals include general practitioners, nurses, nurse practitioners, allied health professionals, midwives, pharmacists, dentists, and Aboriginal and/or Torres Strait Islander Health Workers and Health Practitioners.
QALY	Quality-adjusted life-year (measure of disease burden including both the quality and quantity of life lived).
RNA	A molecule that is translated into the amino acids that build proteins.
RNA sequencing	Technique that quantifies the RNA in a sample and can assist in identifying which genes are turned on, or expressed, in a cell.
Sequencing	A laboratory method that is used to determine the entire genetic makeup of a specific organism or cell type. This method can be used to find changes in areas of the genome.
Single nucleotide polymorphism (SNP)	A common genetic variation that occurs in the population. In combination with environmental/lifestyle factors, some SNPs increase the chance of developing certain conditions (e.g., cancer).
Somatic variant or somatic mutation	New genomic variations that arise in individual cells or groups of cells and are not inherited.
The tumour mutation burden (TMB)	The number (or rate) of somatic variants in the DNA of cancer cells.
Variant of uncertain significance (VUS)	A genetic variant whose role in disease is not yet understood or determined.
Whole exome sequencing (WES)	Identifies nucleotide variants in the exons and the areas of introns immediately prior to and following exons.
Whole genome sequencing (WGS)	Identifies nucleotide variants in the exons and introns (entire gene), as well as structural variations (areas of the genome which that have been rearranged).

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