

# Australia's Genetic Testing Boom: Time for Collaborative Action

A Position Paper by **Inherited Cancers Australia (ICA)** & the **Human Genetics Society of Australasia (HGSA)** – To seek a coordinated, national response to the escalating demand and unmet needs resulting from increased germline genetic testing for inherited cancer risk in Australia.



## Key Messages

- 1** The benefits of genetic testing cannot be realised without sustained support after risk identification.
- 2** This is no longer a niche clinical issue; it is a whole of system challenge requiring coordinated national action.
- 3** Community and clinical services are an important part of the solution but need investment in people, pathways, and partnerships.
- 4** We must act now to save lives and ensure equitable access to risk-reduction strategies.



*"I was only 20 when I found out I had a BRCA mutation. It shook my entire world. Suddenly I had to make major decisions about my health, with cancer always in the back of my mind. I felt lost. There was so much information, but I was too overwhelmed to make sense of it."*

—Meg, BRCA2

Australia is witnessing an unprecedented surge in genetic testing, propelled by advances in precision medicine and innovative therapies that target cancers with specific genetic markers. Genetic testing provides the first step toward targeted treatments for cancer patients but also provides an opportunity for prevention of cancer in people at high risk.

Through increased testing of people with a cancer diagnosis, via tumour or germline testing, more people with an inherited cancer risk are also being identified, including close family members.

With genetic testing now occurring across a range of funding mechanisms, it is difficult to accurately estimate overall volumes. The expansion of criteria for MBS item 73295 in January 2025, supporting diagnostic genetic testing for breast cancer patients, has further accelerated demand, with testing increasing more than twelve-fold in its first six months. This growth demonstrates that testing is needed and valuable, and is expected to continue to grow as awareness and access expand. Although this provides an excellent opportunity to improve treatment outcomes and identify individuals with an inherited cancer risk, the healthcare system is not adequately resourced to meet the associated needs.

Multiple downstream services are required to support those who receive a positive test result, including clinical, psychosocial, and logistical care.

The shortage of funded specialist genetic health services poses a critical bottleneck, risking the timely delivery of care and undermining the potential benefits of testing.

With so many people seeking testing, increased support is being sought from already stretched clinical genetics services and unfunded community organisations. Waitlists continue to grow, patients struggle to access necessary surveillance and risk-reducing procedures, and they often lack adequate emotional and decision-making support.

These barriers need to be overcome to ensure people have access to lifesaving information which allows for proactive decision making, harnessing the preventative health benefits.

**Genetic testing, supported by informed risk management, saves lives. Without immediate, systemic investment and innovation, the benefits of Australia's advances in genetic testing and the opportunity to prevent cancer in a high-risk priority population are being lost.**

# The Case for Action

## Evidence of Unmet Need

**Findings from Inherited Cancers Australia's report "Breaking the Cycle" highlight:**<sup>1</sup>

- Excessive wait times for risk-reducing surgery, in some cases leading to cancer diagnoses while waiting.
- Inconsistent access to cancer surveillance procedures (MRI, colonoscopy).
- Financial hardship due to reliance on private healthcare to fund surveillance and risk management options – 26% of people spent over \$15,000 on managing cancer and cancer risk.
- Psychosocial and decision-making burdens borne largely by patients and families, with support coming from unfunded charities.

## The Challenge

**Rising demand:** Uptake of MBS-funded germline genetic testing, combined with growing awareness, have driven rapid and sustained growth in testing volumes.

**System bottlenecks:** Clinical, psychological and logistical services are not resourced to keep pace with follow-up, risk management, and ongoing care, leading to gaps in long-term support for patients and their families.

**Mainstreaming:** While genetic testing is increasingly offered through mainstream clinical services, specialist genetic support remains essential to ensure patients and at-risk family members can access and interpret results appropriately.

**Workforce funding:** Funded positions for specialist genetic health services, including genetic counsellors, clinical geneticists and hereditary cancer specialists are limited. With few pathways for career progression and professional support, the capacity to meet rising service demand is constrained.

**Unfunded support:** Patient advocacy organisations like ICA provide crucial post-testing services, including education and peer support, but operate without government funding or formalised integration into the health system.

**Fragmented systems:** Disconnected models of care across jurisdictions, combined with the absence of coordinated patient tracking, compromise continuity of care and threaten long-term outcomes.

**Data limitations:** There is no single, reliable source capturing the total number of tests performed, nor a national database to track individuals with inherited cancer risk, limiting planning, monitoring, and evaluation of services for those at high risk.

## Why Now?

**The health system is at a tipping point:**

Demand for genetic testing and follow-up care is outpacing service capacity, placing individuals at risk.

**Policy momentum:**

National frameworks and strategies now explicitly recognise the importance of genomics, and the increased psychosocial support required.

**An opportunity for national leadership:**

Australia can lead in designing a forward-thinking, collaborative, and sustainable response.

**Precision medicine:**

This is becoming best practice in oncology care, and the gateway to precision medicine is genetic testing. Without action to address the support needs of patients, demand and patient safety concerns will continue to grow and become harder to manage.

## Policy Alignment

**Australian Cancer Plan:**

Focuses on Maximising Prevention and Early Detection, Enhanced Consumer Experience and Workforce to Transform the Delivery of Cancer Care.<sup>2</sup>

**National Framework for Genomics in Cancer Control:**

Calls for evidence-based, equitable access and ongoing support post-result.<sup>3</sup>

**Genomics Australia:**

Envisions 'An Australian health system which routinely includes health genomics in clinical practice to prevent disease and provide personalised treatments'.<sup>4</sup>



*"With Ashkenazi Jewish ancestry, I knew my inherited cancer risk was high (1 in 40). My sister was diagnosed with breast cancer and tested positive for BRCA2.*

*"When I tested positive, too, my world collapsed. I was terrified of losing her while also facing my own risk, trapped between grief, fear, and overwhelming decisions about my future."*

*—Nicole, BRCA2*

## Recommendations

### 1 Fund Lifelong Support after Identification of Cancer Risk

Commit funding that goes beyond genetic testing to cover the entire journey including genetic counselling, psychosocial support, risk navigation, enhanced surveillance, and long-term follow-up. Modernise models of care by embedding genetics into oncology, gynaecology, primary care, and public health, ensures services are accessible, interdisciplinary and prepared for future growth in genomic medicine.

*“Clinical genetic services in Australia are struggling to meet the need for ongoing care for the increasing number of people at a high risk of cancer identified through genetic testing, with follow up support for risk navigation falling on unfunded patient organisations.”*

–Paul James, Director, Parkville Familial Cancer Centre

### 2 Build and Sustain the Workforce

Invest in recruitment, retention, and career development of genetic counsellors, clinical geneticists, and hereditary cancer specialists. Expanding workforce capacity including sustained funding for genetic counselling, will allow services to meet increasing demand, reduce burnout, and maintain high-quality care. A well-resourced workforce, fully integrated into cancer care services, ensures patients receive comprehensive and coordinated care throughout their risk management pathway.

*“The benefit of learning about hereditary cancer risk can only be realised if people can access services.”*

–Julia Mansour, Chief Executive Officer, Human Genetics Society of Australasia

### 3 Strengthen Patient-Centered Support and Evidence base

Provide ongoing government funding to patient advocacy organisations that deliver education, peer connection, and long-term support from others with a lived experience. Develop a national registry/patient tracking system to enable longitudinal coordination of care, timely risk management, and equitable access across the life course.

*“It’s like diagnosing diabetes but funding no educator, no dietitian, no follow-up, just leaving people to figure it out themselves. We would never accept that for other serious health conditions.”*

–Sarah Powell, Chief Executive Officer, Inherited Cancers Australia

#### References:

1. <https://d20suj4g28m6xs.cloudfront.net/9bc637f562472ce9faa4f4be953552c5.pdf>
2. <https://www.australiancancerplan.gov.au/so/maximising-cancer-prevention-and-early-detection> – Action 1.5.4: Identifying individuals at increased risk of developing cancers or cancer recurrence can support preventive interventions and earlier detection. This has significant potential to reduce the prevalence of later stage cancers, particularly among high-risk individuals, and improve overall survival rates.
3. <https://www.canceraustralia.gov.au/sites/default/files/2025-02/national-framework-for-genomics-in-cancer-control-cancer-australia-january-2025.pdf> – 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. <https://pmc.ncbi.nlm.nih.gov/articles/PMC10315723/>
4. <https://www.health.gov.au/our-work/genomics-australia>