Out From the *(BRCA)* Shadows:

A supplementary chapter exploring the reality of living with a hereditary form of metastatic breast cancer

Picture features Brigitte, mother-of-two, living with *gBRCA* metastatic breast cancer

AstraZeneca

The diagnosis

"You have metastatic breast cancer" is one of the most difficult sentences for anyone to hear.

"You have a hereditary form of metastatic breast cancer" is potentially even more shattering, as it means a gene mutation driving someone's incurable disease could also affect members of their family.

As Sarah Powell, the CEO of Inherited Cancers Australia points out, hereditary cancer "is cancer that cuts across generations and forces multiple generations into the fight of their lives. When cancer runs in the family, the stakes and the emotional toll are even higher".[1]

Testing times

Between 5 and 10 per cent[2] of all breast cancers are estimated to be hereditary. However, approximately half of those carrying a genetic mutation do not present with a family history of cancer.[3] This means that breast cancer screening relying primarily on family history may preclude early diagnosis of some high-risk women, potentially compromising treatment outcomes.[4]

Genetic testing is a critical tool in many people's diagnosis and treatment journey. The more information a person has about their genetic history, the more options or specific treatment pathways they will have available to them.

Genetic testing also complements proactive self-surveillance of breast cancer and may significantly improve patient outcomes through the early identification of patients at high risk via screening, risk-reducing surgery or individualised systemic anti-cancer treatment.[4]

The emotional challenges

A hereditary cancer predisposition puts a person at a higher lifetime risk of developing cancer.

Women with a breast cancer gene (*BRCA1*) mutation, for example, have a 72 per cent lifetime risk[2] of developing breast cancer. If they are diagnosed, this tends to happen at a younger age[3] than those facing non-hereditary breast cancers.

A metastatic breast cancer diagnosis has the potential to disrupt family dynamics, carrying profound emotional implications not only for the patient but also for their wider family – their children, parents, cousins, aunts, uncles, and grandparents and/or grandchildren.

For a person diagnosed with germline *BRCA* metastatic breast cancer, they are faced not only with their own significant illness and unending treatment but also potentially feelings of guilt, however unfounded, that they are responsible for any mutations they may have passed on to their children and/or grandchildren.

They may also feel resentment towards older generations who might not have understood the significance of disclosing a family history of breast, ovarian and/or prostate cancer and/or taken steps to seek genetic screening, which could have allowed younger generations to do the same and seek risk-reducing options.

The cascade effect

The discovery of a *BRCA* mutation sparks what is known as predictive or 'cascade testing', the process of informing family members that a genetic condition has been found within the family, followed by family members getting tested for the condition. For every one person diagnosed with hereditary cancer, an average of 3.3 family members will undertake genetic testing to understand their own risk.

For those facing the risk of inherited cancer, psychological distress, anxiety, and fear of developing cancer are common.[5]

These people must navigate through life with the knowledge that they have a significantly higher chance of developing cancer, but with no dedicated specialist (unlike those with a cancer diagnosis).

And, as more and more Australians undergo genetic testing each year, this community is expanding.

Brigitte's story

"My diagnosis with breast cancer came when I was eight months' pregnant with my second child. I found a lump, and was told it was mastitis. When the doctor said it was breast cancer, I was completely floored. Never in my wildest nightmares would I have imagined I'd be diagnosed with breast cancer at age 31.

"After my diagnosis, things happened very quickly. I had my daughter via C-section five days later, and a few days after that I had scans to see if the cancer had spread to anywhere else in my body. The day after I got home from the maternity ward, I was called back to the hospital to get the results of those scans.

"I was told that the cancer had already spread. It was in both breasts, my lymph nodes and my bones. I never asked my oncologist what my prognosis was because I didn't want to know. I knew it was bad.

"At the time I didn't know a lot about breast cancer, but I knew if it had already spread it would be incurable. I spoke to three oncologists, who agreed it was essential I get tested for genetic mutations. I didn't have a family history of cancer, so I was actually very surprised to find out that I carry a genetic mutation in my BRCA1 gene.

"Having cancer, you often feel like a burden on the people around you. The weight of knowing my mum, sisters and daughters might also carry the mutation felt like an added burden. It's not that I thought I was in any way responsible for the mutation, but there is a kind of responsibility that comes with being the bearer of bad news.

"To their credit, my family were great. My parents and sisters were tested immediately and we learned the mutation comes from my mum's side. My children will have the choice to be tested when they turn 18 and when the appropriate time comes, I will encourage them to do so. That knowledge is worth so much because it means that you can be proactive in screening and taking preventative measures.

"As a metastatic breast cancer patient, I often avoid telling other patients my diagnosis. Everyone knows metastasis is a possibility, but for a lot of people it's such a terrifying thought it can be difficult to even say the word. And because it carries such fear and uncertainty, those of us living with metastatic cancer can end up feeling pushed into the shadows.

"Cancer in a breast doesn't kill someone, but it's a different story when that cancer has spread to other parts of the body. Because of that difference, the resources and supports that are available to breast cancer patients are often not appropriate for metastatic patients. The needs are just so different, so there really is a support gap that needs to be filled."

Finding the funding to fight risk

Once a genetic predisposition is established, the challenge is to break the cycle.

Knowledge is power – but often that power comes with a prohibitive price tag.

An Inherited Cancers Australia survey[1] in 2024 revealed that of the

of respondents who'd had **risk-reducing surgery**,

had spent more than **$5,000** out of pocket;

spent more than **$15,000**, and some reported costs in excess of **$50,000**.

Patient groups in Australia, including Breast Cancer Network of Australia[6], are calling for a nationally consistent approach to prioritising access to prophylactic (preventative or risk-reducing) mastectomy, a surgical procedure to remove the breasts to cut the risk of breast cancer by at least 95 per cent.

This removes the need for, and cost of, treatments and also the fear and anxiety that comes with it, however risk-reducing surgeries are not consistently classified as semi-urgent elective surgeries in public health services.

Inherited Cancers Australia (ICA) says one of the biggest problems is the wait times for preventative mastectomies as performed by public hospitals – the model for which was set up in a time before Australia was performing this amount of genetic testing.

Those undergoing risk-reduction surgery also need expert and peer support in the lead up to and afterwards. This support is provided by ICA, the only patient group dedicated to empowering those with or at risk of hereditary cancer with advice and support, however its reach is limited due to a lack of government funding.

To help drive preventative treatment and monitoring of this high-risk population, ICA says funded genetic counselling is needed to guide any decisions on preventative treatment. Funding is also needed for ongoing peer support to address the significant psychological needs of people managing their lifetime risk of cancer.

According to the Human Genetics Society of Australasia (HGSA), the value of genetic testing is limited without the funded supportive services of genetic counsellors in community settings.[7]

The HGSA points out that at present, no MBS item number exists to allow for professional genetic counsellor risk assessments in community settings. Due to this and an overburdened public healthcare system, the wait to access genetic counsellors can be up to 36 months, the HGSA says.

It's also viable for Governments, as for every $1 invested in genetic counselling, there is $2.50 to be gained in health, social and economic benefits, according to modelling conducted by the HGSA.

The absence of a clear family history also contributes to limited uptake of genetic testing and counselling in Australia. Under-referral by doctors, limited awareness of the testing process and lack of confidence in supporting the patients through the testing process further compound the problem.

What MAGENTA found

More than 1,200 breast cancer patients from around the world, including Australia, shared their views on the barriers they experienced in accessing genetic testing and counselling. The AstraZeneca-led MAGENTA study[4] of 2024 revealed:

Critical gaps in access to, as well as awareness and perceived value of, genetic testing and counselling, with regional variance or difference between the tested and untested groups;

71 per cent of breast cancer patients have 'moderate to low' awareness of potentially needing genetic testing and counselling;

Most patients are not offered counselling, which may be associated with the low uptake of testing;

Action is urgently needed to drive policy-shaping and improve access to testing and counselling, including raising patient awareness and improving patient experience for better treatment outcomes.

Prioritising the psychosocial needs of people with hereditary metastatic breast cancer, as well as their families

Just as 2024's '*Out from the Shadows*' report called for greater **visibility**, **voice** and **support** for Australians living with metastatic breast cancer, this supplementary chapter affirms the pressing need for a special focus on the unique needs of those with a hereditary form of the disease.

All people with metastatic breast cancer need dedicated nurses, psychologists and counsellors but those facing a hereditary form need extra psychosocial support to ease the burden, in the form of:

EQUITY OF SUPPORTIVE CARE – Federal Government funding to allow an expansion of tailored, accurate information and psychosocial support, including professionally facilitated peer support, for people affected by germline *BRCA*-mutated (g*BRCA*m) metastatic breast cancer, as well as expert guidance for their families.

References:

1. Breaking the Cycle: Insights from the Inherited Cancers Australia community. Accessed June 2025. www.evohealth.com.au/wp-content/uploads/2025/06/Breaking_the_cycle.pdf.
2. Breast Cancer Network Australia, Genetic Mutations that Increase Breast Cancer Risk. Accessed June 2025. https://www.bcna.org.au/resources/screening-and-diagnosis/family-history-and-genetics/genetic-mutations-that-increase-risk/.
3. Sessa C, et al. Risk reduction and screening of cancer in hereditary breast-ovarian cancer syndromes: ESMO Clinical Practice Guideline. *Ann Oncol.* 2023;34(1):33-47.
4. Powell S, et al. MAGENTA: a Multinational patient survey assessing the Awareness, perceptions and unmet needs in GENetic Testing and counselling among patients with breAst cancer. *Front Oncol.* 2024;14:1380349.
5. Kastner AM, et al. Distress, anxiety, and depression in persons with hereditary cancer syndromes: Results from a nationwide cross-sectional study in Germany. *Cancer Med.* 2023;12(12):13701-13711.
6. BCNA, Prioritising access to risk-reducing surgery. Accessed June 2025. www.bcna.org.au/our-impact/prioritising-access-to-risk-reducing-surgery/.
7. Human Genetics Society of Australasia, Genetic experts call for more frontline staff to combat long waits for support. Accessed June 2025. www.hgsa.org.au/Web/Web/News/Articles/HGSA250404.aspx#:~:text=NATIONAL%3A%20Human%20Genetics%20Society%20of%20Australasia%20Ltd%20%28HGSA%29,%28MBS%29%20as%20part%20of%20the%202025%20Federal%20Election.

AstraZeneca Pty. Ltd. ABN 54 009 682 311. 66 Talavera Road, Macquarie Park, NSW 2113. www.astrazeneca.com.au. For Medical Information enquiries or to report an adverse event or product quality complaint: Telephone 1800 805 342 or via https://contactazmedical.astrazeneca.com. July 2025, AU-22734, INDE18687.