Sam‘s story– *BRCA1* high-risk variant

Sam’s life (and that of her family) changed significantly when she signed up for Monash University’s DNA Screen study.

Sam is 39 years old, and lives in small town Cudgewa in Victoria near Albury-Wodonga. Growing up, she always felt there were unanswered questions in her family’s medical history. Her mother was diagnosed with breast cancer at just 40, but genetic testing was not offered to her at the time.

In her 20s, Sam raised concerns about her possible risk with GPs, who dismissed them and informed her that she did not qualify for testing because of an “insufficient family history”. However, just before she turned 30, a visiting Royal Flying Doctor’s women’s health doctor finally validated her concerns and recommended genetic testing.

Coincidentally, not long afterwards Sam saw a segment about DNA Screen on the ABC. At the time, her primary health concern was heart disease, not cancer, as years of reassurance from doctors had convinced her that inherited cancer risk was not an issue. Her DNA Screen results, however, told a different story.

Through the study, Sam found she had a high-risk variant in the *BRCA1* gene, giving her a lifetime risk of breast cancer of over 70%, and an ovarian cancer risk of over 40%. While this was confronting, she was not entirely surprised. Her maternal grandfather, a Polish immigrant who came to Australia after World War II, had prostate cancer and multiple melanomas. For Samantha, her genetic result explained generations of cancer vulnerability.

“I would have been more shocked if they’d said there was nothing there,” she said.

As she was approaching the age of her mother’s diagnosis, she moved quickly. She received genetic counselling and a risk-management plan, and is now on a waiting list for a preventative double mastectomy.

While waiting, she continues to receive regular high-risk breast surveillance, including an annual ultrasound and MRI. This gives her some reassurance that any early cancer would likely be detected early, when treatment is most effective.

Sam’s genetic result has had far-reaching impacts on her family. Her three children—aged 22, 20 and 16—have all faced the question of testing. Her two eldest children were tested and both received negative results, which was a profound relief. Her sister and her niece have both tested positive. Her sister is now also on a waiting list for a mastectomy, and her niece has begun early surveillance.

Being the first in her family to undergo testing has introduced some emotional complexity. While some relatives have been supportive and proactive, others have struggled to accept the implications. Some family members have questioned whether preventative surgery is “too dramatic.” Her response is simple: “Breast cancer and six months of chemotherapy is dramatic.”

Her mother, who remains well and attends regular check-ups, has chosen not to be tested.

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Sam’s health journey has also been logistically exhausting. Living rurally means a five-hour trip each way to Melbourne for appointments, some of which have been cancelled after she has already arrived in Melbourne. Alongside this, she continues to work as a nurse, providing care to around 2,000 people in nearby communities through her service.

Samantha believes strongly that programs like DNA Screen are critical. In her view, too many people with high genetic risk fall outside current testing criteria and are not getting access to this crucial health information.

“This is preventive medicine,” she says. “And preventive medicine outweighs the cost of treating disease once it happens.

“If I had cancer, what would that have cost Medicare…compared to preventive surgery?”

She has a clear message to the Australian government: “They need to fund this. They need to sit down at home and look at their family and say what would they do it if it was their daughter or their mother or their wife or their sister that had a significant increased risk of a genetic mutation that could give them cancer.”

She is clear that without DNA screening, her life might look very different today.

Open about her experience, Samantha has shared her story publicly, including on social media, and has been struck by how many people in her small community are affected. In her small town alone, she is aware of two other women with *BRCA1/2* variants mutations, and one who has already undergone a preventive mastectomy.

“People just aren’t talking about it enough,” she says.

Through online communities, including the Inherited Cancers Australia Support Group, BRCA1 & BRCA2 Support Group Australia, DIEP FLAP Australia and others, and groups like the Breast Cancer Network Australia, Samantha has found connection and support. She remains committed to sharing her story—believing that awareness, early detection and access to testing can change, and save, lives.

For more information about DNA Screen, head to https://dnascreen.monash.edu/.

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Photo credit: Posterboy Media

Location of this story: https://dnascreen.monash.edu/participant-stories.html#participant-interviews

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