The New Zealand Familial Breast Cancer Study

Kia ora,
It has been a very busy year for the research team, with several major projects reaching important milestones. We greatly appreciate your continued support, and we are pleased to share some exciting updates.

The Confluence Project – Identifying common genetic changes that influence breast cancer risk

A landmark global study has uncovered hundreds of new genetic clues to breast cancer risk. These discoveries are helping researchers build more accurate tools to predict risk and improve prevention for future generations.

The Confluence Project is the largest international study ever conducted. It has brought together data from more than 290 studies across 61 countries, including many participants in the NZFBCS. The project has analysed genetic information from over 426,000 women with breast cancer and more than 1.7 million without the disease.

The first major results have been released, linking 238 new regions of the genome with breast cancer risk, including several genes not previously associated with the disease. Because this study included people from a wide range of ethnic backgrounds, as well as many individuals carrying *BRCA1* and *BRCA2* variants, researchers are now developing improved risk prediction tools that are expected to provide more accurate risk estimates across diverse populations.

While research is still ongoing, these discoveries are already providing important new insights into the genetic factors that contribute to risk. In the future, this knowledge is expected to help clinicians deliver more personalised risk assessments, improve prevention strategies, and guide the use of genetic testing and screening. NZFBCS participants continue to play a valuable role in these international efforts to improve outcomes for future generations. We look forward to sharing more results from this exciting study as the analyses are completed.

Developing preventative therapies for breast and ovarian cancer

We are exploring new ways to reduce the impact of high-risk genetic variants and improve prevention choices for future generations.

Our research is investigating new ways of preventing breast and ovarian cancer in people who carry high-risk genetic variants. In earlier laboratory studies, we demonstrated that specialised molecules called antisense oligonucleotides could reduce the harmful effects of specific disease-causing variants in a gene linked to increased breast cancer risk.

We are now building on this work to make the approach relevant to a wider range of people carrying high-risk variants. While this research is at an early stage, it is an important step towards developing preventative treatments that could one day complement existing risk-reduction options, such as preventive surgery.

RNA diagnostics research helping improve genetic testing

Not all genetic changes increase cancer risk, but knowing which ones do is crucial. Our research is helping give individuals and families clearer answers about their inherited cancer risk.

One of our major research programmes focuses on improving how genetic variants are interpreted in diagnostic laboratories. Through our Health Research Council-funded RNA diagnostics programme, we are developing methods that allow researchers and clinicians to better understand whether particular genetic variants interfere with the normal function of important cancer susceptibility genes.

Recent work led by members of our team, including Dr George Wiggins and Emily Shaw (Honours student), has contributed to the development of analytical approaches that can be implemented by diagnostic laboratories throughout New Zealand. These methods have the potential to improve the accuracy of genetic testing and provide more certainty for individuals and families undergoing genetic assessment for inherited cancer risk.

Expanding our understanding of breast cancer in young women

Results from our study investigating breast cancer in women under 45 has revealed important differences in diagnosis, treatment, and survival.

We are nearing completion of a four-year collaborative project between the University of Otago and the University of Auckland, funded by Breast Cancer Foundation NZ, which examined breast cancer in young New Zealand women.

Using national cancer datasets, our research has shown that women diagnosed before the age of 45 are more likely to present with advanced-stage disease than women diagnosed later in life. Importantly, this difference was particularly evident among wāhine Māori, Pacific, and Asian women. We also identified differences in outcomes between breast cancer subtypes and found that survival can vary according to the treatments received. The findings from this work will be presented at the 2026 New Zealand Society of Oncology Conference in Wellington in October.

Funding success: New pathways for breast and ovarian cancer testing in New Zealand

Dr Magdalena Ratajska

Dr Vanessa Lau

We are delighted to announce that our research team, co-led by Dr Magdalena Ratajska and Dr Vanessa Lau, has been awarded Health Research Council of New Zealand funding to re-evaluate genetic testing strategies for breast and ovarian cancer in New Zealand.

Current testing guidelines are largely based on overseas data and may not perform equally well across New Zealand's diverse population. This project will investigate inherited genetic variants, tumour characteristics, and biological markers to identify who is most likely to benefit from genetic testing and targeted therapies.

As part of this project, we will sequence breast cancer susceptibility genes in around 1,000 New Zealand patients to determine whether current referral guidelines successfully identify all individuals carrying clinically important variants, regardless of ethnicity.

The findings will help ensure that future genetic testing services are more accurate, equitable, and cost-effective, while supporting improved prevention, earlier diagnosis, and more personalised treatment for New Zealand families.. You can read more about our project by clicking here or by scanning the QR code.

Professor Logan Walker

Health Research Council Liley Medal awarded to Professor Logan Walker

In October 2025, Professor Logan Walker was awarded the Health Research Council of New Zealand's prestigious Liley Medal at the Research Honours Aotearoa awards held at Te Pae Convention Centre in Christchurch.

The award recognised Logan's contributions to understanding and interpreting genetic variants associated with inherited disease. His research has played an important role in developing internationally adopted guidelines that help diagnostic laboratories determine whether specific genetic variants are harmful and should be used to inform clinical care.

These advances are helping improve the accuracy and consistency of genetic testing both in New Zealand and internationally. This award reflects the collective efforts of many researchers, clinicians, collaborators, and importantly, study participants whose contributions make this research possible.

Finally, we would like to extend our sincere thanks to every NZFBCS participant. Your participation continues to make a real difference to our understanding of inherited breast and ovarian cancer, both in New Zealand and around the world.

Ngā mihi nui,

Dr Vanessa Lau - *Study Coordinator*
Professor Logan Walker - *Principal Investigator*