Cancer Council Victoria

Research We Fund

Project:

Discovering new genetic causes of familial breast cancer

Research team:

Dr Na Li

Institution: Peter MacCallum Cancer Centre

Cancer type: Breast

Years funded: 2020

What is the project?

Many breast cancers are caused by faulty inherited genes. For more than half of families the gene causing breast cancer remains unknown, preventing reliable risk reduction advice. No substantial discoveries have been made in identifying new breast cancer genes for decades, so to advance gene discovery we have established one of the largest sequencing datasets in the world, and have now identified over 40 genes that seem likely to cause family breast cancer. In this unique study I will use several innovative approaches to provide additional proof of their involvement in breast cancer.

What is the need?

Familial breast cancer is commonly associated with a young diagnosis age which represents a major personal and public health burden. The risk of breast cancer is not evenly distributed in the general population, with up to 10& of patients thought to have a much higher risk due to their family histories. This is thought to be due to an inherited genetic defect, the identification of which is crucial for clinicians, patients and their families to make informed decisions on risk reduction and treatment. However, the gap in understanding the genetic causes means most high-risk families have no genetic defect identified. Therefore, discovering the missing causes is critical to develop effective prevention and risk management strategies.

What are you trying to achieve?

This project aims to identify new genetic causes of breast cancer from the BEACCON study, one of the largest sequencing datasets in the world. I hope these results may lead to the discovery of genetic defects to make tailored risk management and customised cancer treatment available for high-risk families.

Project timeline

	Early-2020	Mid-2020	Late-2020
Complete data analysis and prepare manuscript describing design and findings of BEACCON study			
Complete genetic data analysis, tumour sequencing and integration of data for priority candidate gene NTHL1			
Apply series of validation approaches to validate the next ten highest priority candidate genes			

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