

Exploring the Genetics of Preterm Birth in Western Australian Families

Research overview

Preterm Birth is a serious and growing problem with over 15 million babies born prematurely each year – this is more than one in ten of all babies born around the world, and approximately 27,000 Australian babies every year. Preterm birth complications are the leading cause of death among children under 5 years of age.

New technologies are being utilised to investigate preterm birth, and develop predictive and preventative measures. One of these technologies is Next Generation Sequencing, which enables the identification of each 3 billion base pairs of a person's genome in a time and cost effective way. The Preterm Birth Genome Project Team are utilising whole exome sequencing to investigate preterm birth in families who have been affected by preterm birth across multiple generations.

Research highlights

This project was awarded research funds by the Channel Seven Telethon Trust in order to carry out this valuable research. We have recruited Western Australian families who are affected by recurrent preterm birth; this includes grandparents, parents, aunts and uncles of the preterm babies, and the preterm babies themselves. The genomes of these participants have been sequenced – each individual letter of their 'coding' DNA is identified. This comprehensive investigation utilising data from numerous generations of the same family will enable the identification of genes which are related to preterm birth.

Once genes associated with preterm birth have been identified, we will calculate a personalised 'preterm birth risk score' incorporating information about a patients' genes, environment and other relevant factors. This will enable preterm birth prevention methods to be targeted towards women who are most at risk of preterm birth.

Research achievements

The exomes or 'coding' DNA of nearly fifty participants in six families have been sequenced utilising two complimentary methods. This is a significant achievement and has generated substantial amounts of data on the genomes of each family member. The genome of each person yields more than 13GB of data. In order to analyse this data, sophisticated statistical methods are required. These methods are continually evolving to reap the most benefit from sequencing and computing advances. Wei Ang, a member of the PGP Team, is making



a significant contribution to the development of new methods for this type of data. Wei has elected to undertake his PhD focusing on the development of analytic methods for Nest Generation Sequencing data.

<u>THE TEAM</u>

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