Our team
Our research program is aimed at improving current diagnostics and opening up new avenues for intervention and treatment of fragile X-associated disorders.

Cognitive neuroscience, psychiatry and neuroimaging
(Monash University, UNSW, La Trobe University)

Molecular biology, genetics & epigenetics (MCRI, VCGS)

Genetics & Genetic Counselling (GOLD Service)

Genetics Education & Health Research (MCRI, VCGS)

+ Erin, Chriselle, and Belinda

Statistics (University of Melbourne)
Individuals who carry an *FMR1* premutation

- Genetic
- Epigenetic
- Brain structure and function
- Cognitive function
- Physical health
- Mental health
Study sites

**Sydney:**
25 males, ages 26-80

**Melbourne:**
35 females, ages 18-55
Summary of findings: males

Higher rates of:
- Depression
- Anxiety
- Hearing loss

With increasing age:
- Volume loss in brain regions important for balance and motor control

Some associated with CGG repeat length
Summary of findings: females

Higher rates of
• Depression
• Anxiety
• Attention difficulties

Difficulties with
• Executive function
• Working memory

Associated with abnormal methylation of a specific region of the $FMR1$ gene (FREE2)

(Kraan et al., 2013, Am J M Gen)    (Cornish et al., 2015, Neurology)
Implications of findings

• Complex relationships between genes, environment, lifestyle, and behaviour

• Possible markers to help understand risk, and provide support as appropriate

• Guide future research
What else are we doing?

Monash University:
• Questionnaire about general health and wellbeing in females
  o Status: 184 questionnaires completed

• Study of hormones in females
  o Status: Analysis and write up in progress
Where are we headed?

Subject to further funding:

- Longitudinal follow-up

- Development of information resources, e.g.:
  - For families
    - Fact sheets
    - How to get help
  - For health care providers (GPs, Neurologists, Geriatricians)
    - Information sheets
    - Journal articles
    - Practice guidelines
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