



FX Update: Findings from Australian Projects

Never Stand Still

Medicine

Department of Developmental Disability Neuropsychiatry

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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)



Statistics (University of Melbourne)



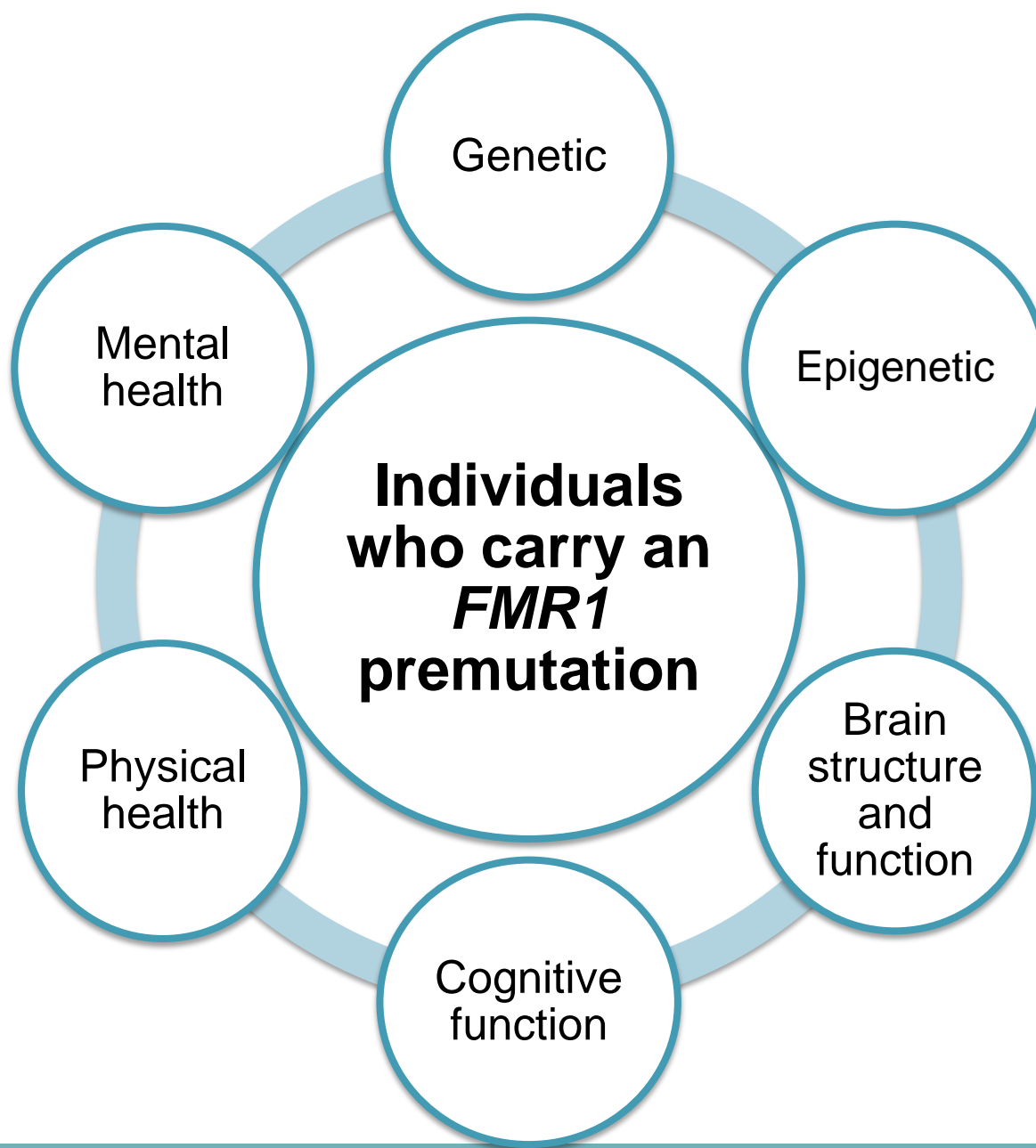
Genetics & Genetic Counselling (GOLD Service)



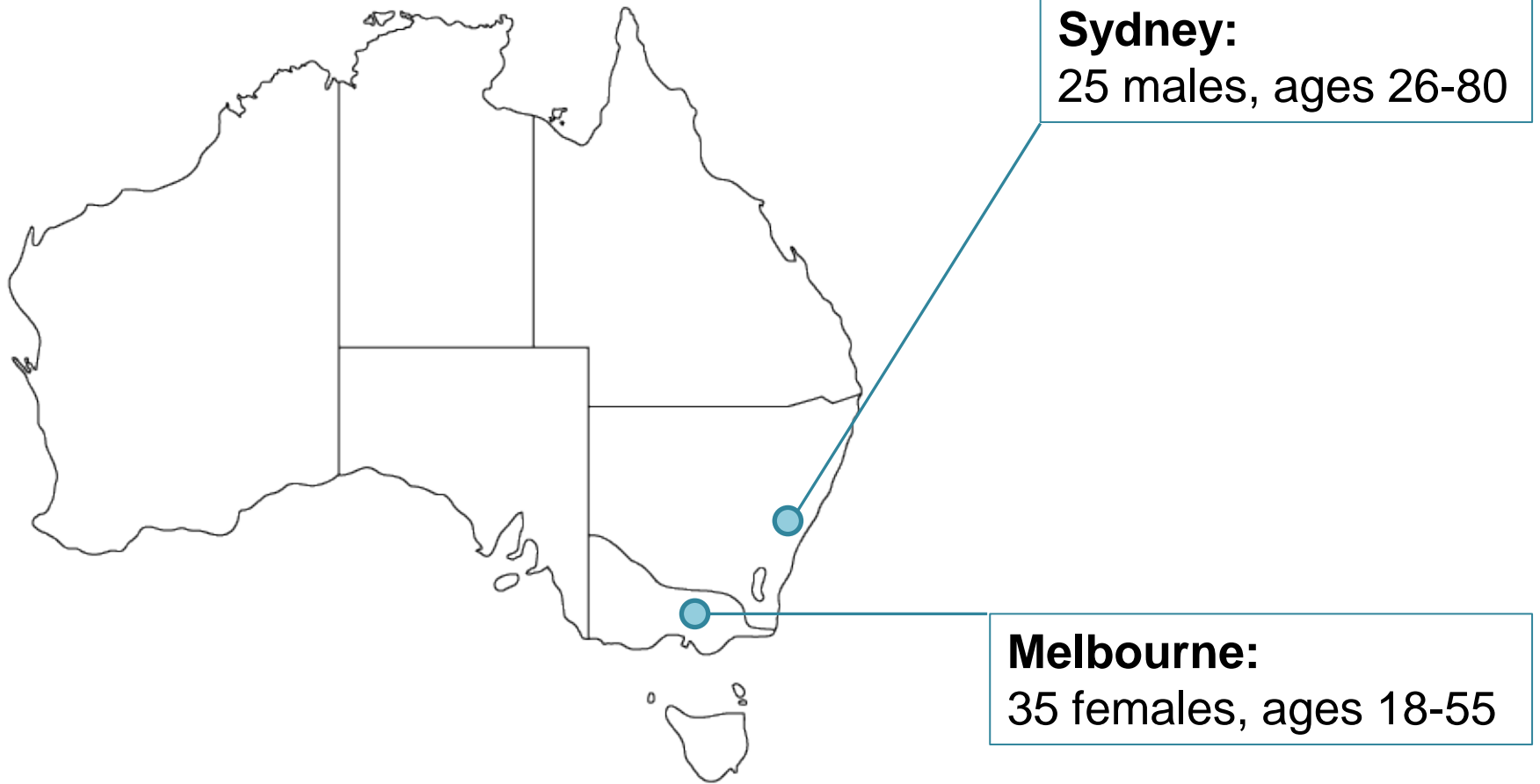
Genetics Education & Health Research (MCRI, VCGS)



+ Erin, Chriselle, and Belinda



Study sites



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
 - Status: 184 questionnaires completed
- Study of hormones in females
 - 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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