AHRA
AUSTRALIAN HEALTH RESEARCH ALLIANCE

Zeroing in on Epilepsy

What is the problem?

Epileptic encephalopathies (EEs) are severe epilepsies that begin in infancy and childhood with devastating outcomes. Children have uncontrolled epilepsy, severe to profound developmental impairment, and other conditions such as autism spectrum disorder and high mortality risk. For most children there is a genetic cause and in 50% of cases the gene involved has been identified. But the problem is very complex and overall more than 100 genes are involved.

About this research translation project

This project will expand on an existing patient group of more than 700 to help understand which genes cause their disease and, of those, which ones might provide opportunities for new therapies. Stem cells derived from the patient group will be utilised to safely test new pharmaceutical treatments.

There are elements of how epilepsy works that are well understood. This project will also investigate the use of existing drugs that have been developed for other conditions to determine their value in treating epilepsy. Because these drugs are already in the market and are safe, they can be rapidly repurposed if useful. Importantly, the research team already has strong working relationships with the families of the children involved – a critical factor in successful trials of this type.

What will be the impact?

This project will have significant impact for epilepsy patients and families. It will enable a specific diagnosis for affected individuals in whom a genetic mutation is identified. This information will mean we can identify which patients will develop other diseases at the same time and the excruciating 'diagnostic odyssey' that families endure often for years will end.

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