AUSTRALIAN FIRST CHROMOSOME 15 BIOBANK ANNOUNCED!

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For the first time in Australia a purpose-built Biobank will be established to store tissue samples and clinical data for research into rare genetic diseases caused by changes to genes on Chromosome 15, including Prader-Willi Syndrome (PWS) and Angelman Syndrome (AS).

Prader-Willi Research Foundation Australia (PWRFA) founder and CEO Kathlene Jones announced the project today, in partnership with Associate Professor David Godler and Professor David Amor from the Murdoch Children's Research Institute (MCRI) and the Royal Children's Hospital (RCH), and Meagan Cross, Chairperson of Foundation for Angelman's Syndrome Therapeutics (FAST).

"For the first time Australia will have a purpose-built biobank of biological samples from people with genetic diseases caused by changes to genes on Chromosome 15," Ms Jones said today.

Biological samples, such as blood and saliva, will be collected from people with PWS or AS, and linked to detailed clinical and medical history data.

This resource will help us to better understand the links between the genetic changes that cause Prader-Willi syndrome and Angelman syndrome and the challenges faced by these people. A particular focus is on the mental health problems experienced by people with Prader-Willi syndrome and Angelman syndrome and how these might be treated more effectively.

"This is the gold standard for biomedical sample collection. It meets a glaring need in Australia, laying the foundation for new lines of research to potentially help thousands of families around the world," Ms Jones said.

The Biobank initiative has been made possible by over $150,000 in funding. Importantly a $65,500 competitive grant awarded by Laurence G and Jean E Brown Charitable Trust through Perpetual's IMPACT program to PWRFA and MCRI underpins the project and is being supplemented with $25,000 from donors to the PWRFA, $25,000 in kind contribution from MCRI and $45,000 from FAST.

PWRFA Director and Scientific Advisory Council Chair, Professor Joe Proietto said today, "The establishment of a biobank linked to clinical history can be a powerful tool to link phenotype with genotype. This linking is the first essential step to developing effective treatments."

The Chromosome 15 biobank will be constructed over the next 2 years and will be housed at MCRI.

Data generated by the biobank will be analysed using advanced artificial intelligence software to identify biological pathways that are disrupted in these disorders, with the aim of developing new treatments. Data and samples from the biobank will also be made available to other researchers and industry, maximising the impact of this resource.

ADDITIONAL BACKGROUND

The National Chromosome 15 Biobank model:

Researchers can access samples from the biobank through a best practice governance process that ensures samples are used ethically and maximise research outcomes for the communities it serves. Researchers email the Biobank Coordinator to submit an application, which is then reviewed by a panel of experts. Suitable projects then send submissions to the relevant Human Research Ethics

Committees for governance and/or ethics approval. Researchers who are granted access to biological materials and medical information from the Chromosome 15 Biobank need to provide 12 monthly updates and a final report upon project completion.

Prader-Willi Syndrome

Prader Willi Syndrome (PWS) is a randomly occurring genetic condition where some Chromosome 15 genes are inactive or missing, causing symptoms that affect every aspect of life. People with PWS have complex medical needs, global developmental delays, challenging behaviours, mental illness and, the hallmark feature of the condition, a feeling of relentless hunger. PWS affects 1 in every 15,000 babies. The strength, resilience and courage people with Prader-Willi Syndrome show to do things others take for granted is remarkable.

Although the most common genetic cause of obesity, PWS research is little known and grossly underfunded.

Prader Willi Research Foundation of Australia

Prader Willi Research Foundation of Australia is a registered health charity and Australia's only dedicated research foundation for Prader Willi Syndrome. The Foundation exists to fund and facilitate research to find life-changing treatments for PWS.

PWRFA is governed by a highly respected Board and subjects project proposals to a rigorous evaluation process via its Scientific Advisory Council (SAC). The SAC is chaired by Professor Joseph Proietto AM, Emeritus Professor at the University of Melbourne in the Department of Medicine, Austin Health and Head of the Austin's Weight Control Clinic and made up of a group of eminent clinicians and scientists from Australia's leading hospitals and research institutes. More information can be found at the PWRFA website:http://praderwilli.org.au/.

Angelman Syndrome

Angelman syndrome (AS) is a rare neurogenetic disorder that affects approximately one in 15,000 individuals. Children and adults with AS typically have impaired motor function and balance, and debilitating seizures. Some individuals never walk. The majority do not speak. Disrupted sleep cycles also can be a serious challenge to the individual and caregivers. Individuals with AS require continuous care and are unable to live independently. They have a normal life expectancy. People with Angelman syndrome have some distinct behavioural traits, including a happy demeanour, characterised by frequent laughing, smiling and excitability.

Foundation for Angelman Syndrome Therapeutics Australia

The Foundation for Angelman Syndrome Therapeutics Australia is dedicated to assisting individuals living with Angelman syndrome to realise their full potential and quality of life through funding research, education, and advocacy. Our focus is facilitating a greater understanding of the syndrome to foster further research and clinical trial capacity through a Global Angelman Syndrome registry. Our goal is treatments that will improve the symptoms of Angelman syndrome and ultimately research that will provide a cure.

About Murdoch Children's Research Institute (MCRI)

MCRI is the largest child health research institute in Australia and one of the top three worldwide for research quality and impact. The institute has made over 100 genetic discoveries, led the development of national and international genomic alliances, and is home to Australia's leading paediatric clinical trials centre, Melbourne Children's Trials Centre (MCTC).