THE AUSTRALIAN BIOBANK FOR CHROMOSOME 15 IMPRINTING DISORDERS

If you are the parent of a person with Prader-Willi syndrome or Angelman syndrome, or yourself have been tested and identified to have one of these conditions, we would like to invite you to participate in the Australian Biobank for Chromosome 15 Imprinting Disorders

What is the research project about?

Prader-Willi syndrome (PWS) and Angelman syndrome (AS) are rare neurodevelopmental disorders caused by genetic alterations that affect chromosome 15. PWS and AS are both associated with intellectual disability as well as other medical problems. It is known that chromosome 15 contains genes that are very important for brain development, but it is not known how changes in these genes cause the health problems that are seen in PWS and AS. We want to find out how genetic changes on chromosome 15 lead to particular medical and psychological problems in people with PWS and AS. In order to do this, we are creating the *Australian Biobank for Chromosome 15 Disorders* ('the Biobank'), which will contain information about a large number of people with PWS and AS. This Biobank will contain both biological samples, such as DNA samples from blood, saliva, and cheek swabs, along with clinical and psychological information about people with PWS and AS.

Information in the biobank will be made available, in a secure and confidential manner, to other researchers who are trying to learn more about PWS and AS. The information will be used to try and solve important research questions about PWS and AS. These research questions include how we can develop new treatments and interventions, how we can start effective treatment in a timelier manner, and how we can best match treatments to individual patients with PWS and AS. We hope that 50 people with PWS and 50 people with AS of any age, from anywhere in Australia, will participate in the biobank.

What does taking part in the Biobank involve?

To take part in the biobank involves one visit at the Murdoch Children's Research Institute (based at The Royal Children's Hospital in Melbourne). During this visit we will take a blood, and/or saliva and cheek swab samples and conduct an assessment of your child's (or your) behaviour, thinking and memory skills, which will involve for example being asked to solve puzzles and remember lists. We will also ask you to complete some questionnaires about your child's (or your) medical history and mental health. The assessment will take approximately 5 hours. We will reimburse you a reasonable amount for your visit travel costs to the site of the appointment in order to participate in this project. Alternatively, we can organize two research assistants to come to your home to do the assessments. If you opt for the assessment at home, the research coordinator may organise a separate time most convenient to you for the biological samples to be collected.

What are the possible benefits of this research?

The samples and results we collect from this project will help us to have a better understanding of the problems that are caused by missing genetic information from chromosome 15 and may help to develop better treatments and early intervention programs in the future. Depending on the outcomes of the assessments we do for your child (or you) we may be able to discuss with you any potential referral for specific intervention programs in the future.

If you would like more information about the project or if you need to speak to a member of the research team, please contact:

Dr Emma Baker; Phone: (03) 8341 6245; Email: emma.baker@mcri.edu.au