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Minister for Medical Research
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MEDIA RELEASE

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UNLOCKING TREATMENTS FOR COMPLEX DISEASES

Six NSW genomics research teams working on unlocking the causes of some of the most debilitating genetic conditions will share in $1.54 million in funding as part of the NSW Government's 2015/16 Genomics Collaborative Grants Program.

NSW Premier Mike Baird and Minister for Medical Research Pru Goward announced the grant recipients during a tour of the Kinghorn Centre for Clinical Genomics at the Garvan Institute of Medical Research today.

"Genomics has the potential to fundamentally change the way medical treatments are delivered and these grants will help NSW researchers continue to lead the nation in this field of research," Mr Baird said.

"These cutting-edge researchers are helping answer the questions why only some people develop certain diseases, and the reasons may lie in their genetic makeup."

Ms Goward said the 2015/16 grants are supporting research into genetic conditions including hereditary heart disease, genetic bone disorders, immune deficiencies and childhood epilepsy.

"These grants will help our researchers continue to advance how patients can benefit from this ground breaking science and the role genomics research can play in identifying individualised treatments based on a patient's genetic makeup," Ms Goward said.

The NSW Government is investing $24 million over four years in the Sydney Genomics Collaborative. As a result of this support, researchers have already explored better treatments for cancer, mitochondrial disease, inherited heart disease in babies and schizophrenia.

2015/16 NSW GENOMICS COLLABORATIVE GRANT PROJECTS

1. **Associate Professor Robyn Jamieson , University of Sydney, $340,000**
   Blinding genetic retinal dystrophies lead to progressive irreversible and untreatable visual impairment and blindness. Inherited retinal disease is currently the most common cause of blindness in the working-age population.

This project will include genetic diagnosis for patients where none has been previously available and aim to facilitate avenues to treatment.

2. **Dr Tony Roscioli, Garvan Institute of Medical Research, $200,000**
   Primary Immunodeficiency diseases (PIDs) involve a predisposition to infection or autoimmune disease. While individually rare, more than 250 different genes are known to cause these diseases, with more than 3,000 more predicted to do so. This project will use Whole Genome Sequencing to test 100 patients and their relatives who have a PID where a gene has not been found.

3. **Dr Tony Roscioli, Garvan Institute of Medical Research, $200,000**
   Mendelian disorders affect 1% of the population and many cause devastating illness that can lead to early childhood death or a severe lifetime disability. Whole genome sequencing provides a platform to identify new genes that cause disease and genetic diagnosis gives patients and families more accurate information about management or chances of recurrence. A group of 60 NSW families who've received extensive clinical investigation represent a significant resource for new disease gene identification. No causative gene has been identified in 70% of these families.

4. **Dr Tony Roscioli, Garvan Institute of Medical Research, $180,000**
   Severe epilepsy is a devastating, life-threatening condition, usually caused by a new or inherited genetic change. Children are born healthy but start to have epileptic seizures, often before 18 months of age, resulting in delayed development. Underlying causes are rarely identified and not knowing the genetic cause means doctors are unable to best target treatment, or provide information about the child's future or the risk of having future siblings affected. Whole genome sequencing of 15 children with early onset epilepsy who remain undiagnosed and 15 new children with later onset epilepsy and intellectual disability will take part in this study, helping doctors with diagnosis and improve the medical understanding of the cause of this condition, leading to improved treatments and better outcomes for children and their families.

5. **Professor Andreas Zankl, Garvan Institute for Medical Research, $300,000**
   Genetic Disorders of bone are rare and affect skeletal development and function. Patients can present at birth, in childhood, or later in life. Early life onset may present with bones that are malformed, fail to grow or fracture easily. Later life onset may present with unusual fractures that don't heal properly. Both result in lifelong disability or even premature death. The study will fully genetically characterize 100 patients with different bone dysplasias. Patients will be given a genetically confirmed diagnosis, allowing for accurate genetic counselling and diagnosis-specific management. The study will also

provide new insight into the role of a multitude of genes on skeletal development, with a better understanding of bone and cartilage biology.

6. Professor Christopher Semsarian, Centenary Institute, $320,000
Genetic inherited heart diseases include cardiomyopathies which primarily affect the heart muscle, like hypertrophic cardiomyopathy (HCM) and dilated cardiomyopathy (DCM). These two inherited cardiomyopathies are common and can lead to heart failure, need for transplantation, stroke, and sudden death. While there has been significant progress in defining some of the genetic underpinnings of HCM and DCM, many questions remain unanswered. In most, the underlying disease causing gene mutations have not yet been identified in 30-70% of cases. This is a world-first comprehensive genetic and clinical analysis of patients, and their families that aims to improve the care of families with inherited cardiomyopathies.