2014 NSW Genomics Collaborative Grants recipients

Professor Graham Mann from the Westmead Millennium Institute for Medical Research / the University of Sydney will lead chief investigators from Melanoma Institute Australia and Macquarie University.
Grant: $811,150
For: Research into new therapies for metastatic melanoma. The team aims to ensure that emergent, expensive therapies will deliver maximum effects to patients likely to benefit from them, while supporting more efficient delivery of care. The data obtained will also guide development and delivery of appropriate tumour and patient germ line genetic testing.

Professor Carolyn Sue from the Kolling Institute of Medical Research will lead chief investigators from the Children's Hospital at Westmead and Charles Perkins Centre, University of Sydney.
Grant: $740,000
For: Research aimed at transforming the diagnostic paradigm for mitochondrial disease, using whole genome sequencing to establish a more simplified and accurate genomic approach.
Mitochondrial disease (MD) is the most common type of inherited metabolic disease in Australia (1:500 live births), affecting both children and adults of all ages. Due to its clinical variability and complexity, MD is often difficult to diagnose. Early diagnosis is critical to early intervention and the introduction of preventative lifestyle changes that may limit its severity.
Professor Sue expects to discover knowledge about genetic causes of MD, build an experienced workforce with expertise in bioinformatics analysis of genetic disorders and improve how patients with MD are cared for.

Professor Sally Dunwoodie from Victor Chang Cardiac Research Institute will lead chief investigators from The Children's Hospital at Westmead and Sydney Children's Hospital.
Grant: $370,000
For: Research into the genetic causes of congenital heart disease (CHD) in NSW families, who will benefit from a genetic diagnosis and will be able to receive personalised advice and treatment.
Heart defects due to congenital heart disease (CHD) are the most common form of birth defect, occurring in 1% of live born babies. In 80% of families it is not known why they occur. Such uncertainty makes it very difficult for parents to cope with CHD in their family as they want answers to the following questions: Why did it occur? Why us? Will it happen again in our family? What is the best course of treatment? What is the prognosis?
The team is identifying the genetic causes of CHD by comparing the genetic code of affected babies with unaffected family members. A genetic diagnosis means that the cause of the defect has been found and the

family can be informed about the likelihood of CHD occurring again. It might also allow treatment and clinical care to be optimised for each family. Capitalising on the world-class cardiovascular expertise within the Victor Chang Cardiac Research Institute, the team will use cutting edge DNA sequencing and data analysis technologies to decode the genetic causes of heart defects in babies in Australia.

Associate Professor Murray Cairns from the University of Newcastle will lead chief investigators from the Schizophrenia Research Institute, the University of New South Wales, St Vincent's Hospital.
Grant: $800,000
For: Research to identify genomic system motifs and associated markers that can inform the development of new interventions that cater more specifically to the individual's genomic architecture and its associated neurocognitive symptoms in relation to schizophrenia.
Schizophrenia is a severe life-long psychotic disorder with enormous burden for sufferers and the health system. While the genetic, neurobehavioural and neuroanatomical complexity of this syndrome has made it one of the most significant challenges to health research, recent developments in high-throughput genomics, computing and phenotyping has provided an unprecedented opportunity to comprehensively explore the genetic and epigenetic aetiology. With the support of NSW Health, this team's neurobehavioural genetics group has pioneered the use of this technology to make an integrated map of functionally significant genetic lesions in schizophrenia, using the largest cohort in Australia.
While a significant feature of the investigation has been the substantial contribution of non-coding segments of the genome, the sequencing effort to date has focussed predominately on the protein-coding fraction by an exome enrichment protocol (Exome-Seq).
With the Garvan Institute's acquisition of the Illumina HiSeqX10 and continued support from NSW Health, there is an exciting opportunity to broaden this investigation to whole genome sequencing (WGS) to comprehensively explore variants in structural and regulatory features associated with both coding and non-coding DNA.