LUMINESCE Alliance

Innovation for Children's Health

Innovation in Paediatric Precision Medicine - Seed Funding Projects

Webinar

Thursday 3 June 2021, 1:00pm AEDT

Luminesce Alliance has awarded seed funding to 5 innovative paediatric precision medicine projects.
Register and join us to hear from the Recipients as they discuss their projects, what they hope to discover and what will be the future.

Prof Phillip Hogg
Deputy Director, Centenary Institute Head, ACRF Centenary Cancer Research Centre, University of Sydney

A self-amplifying theranostic for treatment of neuroblastoma
This study aims to evaluate the therapeutic efficacy of 177Lu-CDI in unprimed or cyclophosphamide-primed murine neuroblastoma tumours. It is hypothesise that 177Lu-CDI will bind to dying/dead neuroblastoma tumour cells and deliver therapeutic radiation to viable, potentially more resistant tumour cells adjacent to the dying /dead cells, and in combination with sensitising chemotherapy will generate a self-amplifying cascade of neuroblastoma tumour cell kill.

Professor Hogg is a NHMRC Senior Principal Research Fellow and holds the Chair in Translational Cancer Research at the University of Sydney. He graduated with a PhD in biochemistry from the University of Queensland. His post-doctoral training was in the USA and Sweden followed by visiting fellowships at Children's Hospital, Harvard University and the Dunn School of Pathology, University of Oxford

Prof Wendy Gold
Group Head, Molecular Neurobiology
Research Lab Senior Research Fellow, Kids Research, CHW

Translating disease severity biomarkers into the clinic for Rett syndrome
Rett syndrome is a rare severe neurodevelopmental disorder caused by variants in the *Methyl-CpG-binding protein 2* (*MECP2*) gene. It is the second most common form of intellectual impairment in females. One of the aims of the study is to determine whether serum levels of FGF21 and GDF15 are prognostic biomarkers of disease stage and severity in girls with Rett syndrome.

Professor Gold has been studying the pathogenic mechanisms of Rett syndrome for the past 9 years. As group leader of the Molecular Neurobiology Lab at Kids Research, Westmead Children's Hospital and Adjunct Research Scientist at Children's Medical Research Institute, she conducts a program of interdisciplinary research spanning fields of neurology, neuropathology, fundamental neuroscience, clinical chemistry and pharmacology. Her translational research engages clinicians, chemists, fundamental scientists and bioinformaticians.

Prof Robyn Jamieson
Head, Genetics Medicine, CHW
Eye Genetics Research Unit
Children's Medical Research Institute

Precision medicine addressing a novel disease pathway to preserve sight in the retinal dystrophies
Inherited Retinal Diseases (IRDs) affect approximately 1:1000 people or leads to an inexorable degeneration to blindness. There is marked genetic heterogeneity hampering individual gene therapeutic efforts. This project aims to develop a novel therapeutic approach towards a disease pathway we have recently identified, that will be applicable to the broad group of IRDs, thus able to benefit a large proportion of patients.

Professor Jamieson is Professor of Genomic Medicine, University of Sydney, Australia. She leads the Eye Genetics Research Unit at Children's Medical Research Institute, Sydney Children's Hospitals Network and Save Sight Institute, University of Sydney. She heads the Western Sydney Genetics Program and Eye Genetics Clinic, Sydney Children's Hospitals Network. Her research is focussed on a comprehensive strategy for development and implementation of new therapies for blinding genetic eye diseases, especially of the retina. Her laboratory undertakes functional genomic, stem cell and gene editing approaches to determine underlying disease mechanisms and test new genetic therapies in the retinal dystrophies.

Dr Samatha Ginn,
Senior Researcher,
Gene Therapy Research Unit
Children's Medical Research Institute

Curing genetic metabolic liver disease by precise genomic and epigenomic editing
While individually rare, genetic metabolic liver diseases are collectively common, difficult to treat and carry high morbidity and mortality. This study hypothesise that precise genetic and epigenetic editing at the human Ornithine transcarbamylase deficiency (OTC) locus can be used to restore physiological OTC expression in male and female patient-derived primary human hepatocytes in vivo at clinically relevant efficiencies. One of its aims is to optimize the efficient delivery of genetic and epigenetic editing reagents to patient-derived OTC-deficient primary human hepatocytes in vivo using elite AAV capsid technology (AAV-LB12) in combination with lipid nanoparticle (LNP) technology.

Dr Ginn is a senior researcher in the Gene Therapy Research Unit of the Children's Medical Research Institute and a Senior Lecturer at the University of Sydney. She received her PhD degree in Molecular Biology and Bacterial Genetics from the University of Sydney and since then, has gained extensive experience in the field gene therapy, viral-based gene delivery technology and genome editing. She was a key team member involved in treating an infant with X linked severe combined immunodeficiency, the first infant in Australia to be treated with gene therapy. She is the current Chair of the Westmead Research Hub EMCR Committee, holds leadership positions in the Australasian Gene and Cell Therapy Society and Asia-Pacific Consortium of Gene and Cell Therapy and is an Editorial Board member for the Journal of Gene Medicine.

Adam Bournazos
Senior Research Assistant
Kids Neuroscience Centre
LA Centre for RNA Diagnostics

A pipeline of accredited RNA Diagnostics to extend diagnostic yield of rare disorders by 25% in 5 years
Variants of Uncertain Significance (VUS) leaves families and clinicians with no actionable answer and health systems with no diagnostic return on their investment into genetic sequencing. This health implementation project will establish a centre for RNA Diagnostics that will provide an accredited RNA diagnostic service with 95% diagnostic return (ie 95% variant re-classification). It will aim to resolve pathogenicity of splicing variant VUS for 60 families with rare monogenic disorders or germline cancer.

Adam completed a BSc Hons I at the UNSW then joined the Kids Neuroscience Centre as a Research Assistant supporting basic research projects into Dysferlin and its role in limb-girdle muscular dystrophy. Now a third year USYD PhD candidate analysing splicing variants with the aim of translating RNA diagnostics into clinical practice using mRNA from clinically accessible tissues.

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