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The Luminesce Alliance Newsletter

Issue 4 - December 2020

Innovation in Paediatric Precision Medicine

Seed Funding Outcomes

The Luminesce Alliance Innovation in Paediatric Precision Medicine Seed Funding Round has been a tremendous success. Fifty nine applications were received with a total funding request of just under $11 million. This was more than anticipated. With a total funding pool of just $1 million it was not possible to fund all projects and Luminesce Alliance was only able to fund those who were ranked the highest by the review panel. The successful seed funding applications were:

A self-amplifying theranostic for treatment of neuroblastoma

This study aims to evaluate the therapeutic efficacy of ^{177}Lu-CDI in unprimed or cyclophosphamide-primed murine neuroblastoma tumours. It is hypothesise that ^{177}Lu-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.

Prof Phillip Hogg
University of Sydney

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.

Prof Ian Alexander
Children's Medical Research Institute

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.

Prof Wendy Gold
University of Sydney

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.

Prof Robyn Jamieson
Children's Medical Research Institute

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.

Prof Sandra Cooper
University of Sydney

Our Partners

The reviewers have commented on the diversity received and the calibre of the applications. It was very encouraging to see the breath of paediatric precision medicine research emerging from the Luminesce Alliance partners and wish the unsuccessful applications the best with future funding opportunities.

2019-2020 Luminesce Alliance Paediatric Precision Medicine Annual Report

Luminesce Alliance had the opportunity to present the 2019-2020 Annual Report to the Dr Antonio Penna, Executive Director of the NSW Office of Health and Medical Research (OHMR) and his team on the 27th October 2020. He congratulated Luminesce Alliance on the work that has been achieved and how congealed the partners have been in working together to deliver a paediatric precision medicine program in NSW.

Paediatric Precision Medicine Video Updates

View our most recent video resources aimed at helping us keep up to date with the fantastic clinical and research responses that are occurring across our precision medicine program, all of which is benefiting the children we treat. Have a browse at the resources posted on our website News page, with more to come in 2021.

All the best for 2021

Luminesce Alliance wishes everyone the merriest for the festive season and all the best for the New Year. May everyone have a peaceful and relaxing time catching up with families and friends after a very eventful year. Luminesce Alliance will be closed between 25th December 2000 and the 4th January 2021.

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LUMINESCE Alliance
Innovation for Children's Health