ILLUMINATE

The Luminesce Alliance Newsletter

Issue 1 – March 2020

Welcome to Luminesce Alliance: Innovation for Children's Health

Message from the Chair

As Chair, it is my pleasure to introduce you to Luminesce Alliance's first newsletter and at the same time present you with our new name and visual identity.

Formerly, known as Paediatrio, the Luminesce Alliance name was inspired by the organisation's intent to significantly illuminate paediatric research to improve health outcomes for children through pioneering research partnerships.

Our three founding partners, indeed all our researchers and clinicians, thought it was timely to reflect our capacity to grow our partnerships and demonstrate the power of our collaborations. We believe that this is the only way we can expand our knowledge of paediatric health, so that all our children can live healthy and productive lives. It is for this reason, we have embraced a name that captures how we are working with our partners, and engaging new partners as we grow, in order for us to become a world leader in paediatric translational medicine.

Our early beginnings were definitely pioneering. Established as cooperative joint venture by Sydney Children's Hospitals Network; the Children's Medical Research Institute; and the, Children's Cancer Institute, to bring collaborative interactions between researchers and clinicians in translational paediatric research, we led the way by being one of the first in Australia. Over the last year, Luminesce Alliance has extended its track record of successful collaboration to embrace strong affiliations with the University of Sydney and the University of New South Wales. We are now, no longer a "trio" in "paediatrics" as was implied in our former name "Paediatrio". Having now achieved a wider coalition, we are capitalising on the world class research strengths of these partners; and, developing important collaboration pathways, all of which are leading to improved health and welfare outcomes for children and young people in NSW and beyond.

It is an exciting phase for Luminesce Alliance, particularly as we see momentum across the array of research programs we are supporting through a $20 M grant from NSW Health, specifically:

- paediatric precision medicine program for cancer and rare diseases;
- screening program for newborns for Spinal Muscular Atrophy and Primary Immunodeficiency in NSW and ACT;
- establishment of small-scale production capacity for gene and cell therapy vectors;
- INFORM 2 a clinical trial which will allow Australian children access to a novel drug treatment cancer therapy in high-risk relapsed/refractory malignancies.

In this newsletter, and subsequent bimonthly issues, our aim is to keep you informed of these achievements; introduce you to our leading researchers and clinicians all of whom are part of this journey; and keep you informed of upcoming events.

I also encourage you to visit our recently launched Luminesce Alliance website at www.luminesce.org.au to find out more about the Alliance.

Kathryn Greiner AO

In the Spotlight

Paediatric Precision Medicine

Luminesce Alliance is proudly supporting the Paediatric Precision Medicine (PPM) program which will transform children's health care in NSW by bringing precision medicine to all children with cancer and rare genetic diseases.

The Precision Medicine program is made up of 6 streams across pre-clinical, clinical and health systems research, which together will build capacity and enable new technologies to be integrated into personalised clinical care.

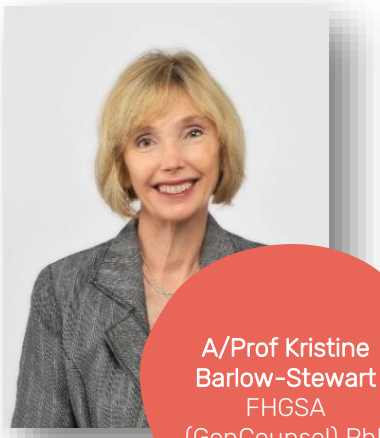
We will be highlighting the exciting research that is currently being delivered across these streams in our regular feature section, *In the Spotlight*.

Read more about each of the precision medicine streams including information on Lead Investigators, Team Members and Collaborators, by visiting our website, **www.luminesce.org.au**

Paediatric Rare Disease Predisposition Screening Program (PPM3b)

By nature, and by definition, a rare genetic disease is extremely uncommon, difficult to detect and difficult to treat. What happens when a child is suspected of having a rare genetic disease? Do clinicians know what to do? Are they aware of the latest advances in precision medicine? And what information and services are provided to the parents?

These are some of the questions being answered by the team behind PPM3b. One project under PPM3b and being led by Associate Professor Kristine Barlow-Stewart, is looking at sub-specialities across the Sydney Children's Hospitals Network (SCHN) where a model of care which incorporates precision medicine exists; and, those sub-specialities where a model of care needs to be developed.



A/Prof Kristine Barlow-Stewart FHGSA (GenCounsel) PhD

For each sub-speciality at the two SCHN campuses, Westmead and Randwick, it will look at how models of care unique to particular sub-specialties are currently implemented and how they were developed, and the role of clinical geneticists and the genetic counselling team in the optimal patient journey. It will also look at barriers and challenges in a model's current and future implementation and differences between the campuses. The findings may inform best practice in implementation of precision medicine across sub-specialities at the two campuses. They may also guide future developments with sub-specialities where such models currently do not exist. Read more.

Model of care
The way health services are delivered. Outlines best practice and maps out the patient journey.

The psychosocial implications of genetic testing and precision medicine for children and their families & the healthcare professionals who care for them (PPM6)

Children with high-risk cancers tend to have limited treatment options, with often devastating consequences. Precision medicine, which uses new technologies to better understand the causes of each child's cancer and to test new treatment options, is offering new survival hope for children and their families.

However, with any radical shift in medicine, challenges can emerge as healthcare professionals try to add new approaches into their practice. These challenges were analysed as part of a study led by one of our Alliance researchers, Brittany McGill, with the study recently published in the *Journal of Personalized Medicine* The hopes and challenges of precision medicine for children with cancer.

Balancing expectations with actual realities

Brittany McGill, Research Fellow, Behavioural Sciences Unit, School of Women & Children's Health, UNSW

In this study, a broad group of doctors (including paediatric oncologists, pathologists and genetics professionals) and scientists (including laboratory scientists and bioinformaticians) working on PRecISion Medicine for Children With Cancer (PRISM) were interviewed.

Most professionals interviewed were excited about the potential of precision medicine over the long-term, but were mindful of the importance of managing families' expectations in the early stages of the trial. Key challenges for doctors, especially oncologists, included sharing genomic results with families. Doctors who did not specialise in cancer genetics reported they felt unsure talking with families about results which might suggest that cancers 'run in the family'. From the conversations with scientists, the overwhelming impression was that being part of such a 'high-stakes' trial made them feel new emotional pressures and experience new workload demands.

The evidence from this world-first study will now be used to inform our understanding of how best to support doctors and scientists striving for new treatment options for children with cancer.

PPM Project Lead

Overseeing the project management for the Precision Medicine Program is Quoc Nguyen, who joined Luminesce Alliance last October.

Quoc Nguyen,
Luminesce Alliance
Project Manager

Quoc has over 20 years' experience in health and medical research management. He has managed multicentred epidemiological studies, clinical trials, casemix funding modelling, medical research laboratories and translational research projects. He also been involved in the foundation establishment of several research centres and groups including, the National Exceptional Case Coordination Unit (NECCU), The Australian Candidaemia Study Group, the Colorectal Cancer Research Consortium, Pharmacogenomic Research for Individualised Medicine (PRIMe), and the Australian Prostate Cancer Research Centre - NSW (APCRC -NSW).

Shine Brightly

Dianne Sylvester PhD

After working for many years as a diagnostic scientist in a cytogenetics laboratory analysing tumours from childhood cancer patients, I was inspired to pursue a career investigating the underlying genetic changes that might contribute to the development of cancer in children.

Shine Brightly
An occasional piece highlighting our dedicated researchers who are looking for ways to expand our knowledge of paediatric health.

Dianne Sylvester
PhD Research Officer
Children's Cancer Research Unit
SCHN

I recently submitted my PhD thesis titled *'Genes underpinning predisposition to childhood cancer'*, which found that almost a quarter of childhood cancer patients with features indicative of an underlying genetic susceptibility to cancer may carry pathogenic germline variants in cancer predisposition genes.

Fortunately, I now have the opportunity to continue my research as part of the *Paediatric Cancer Predisposition Screening Program (PPM3a)*.

For every child diagnosed with cancer in NSW, our project will investigate whether underlying genetic variants may have contributed to the patient's cancer diagnosis. We will identify the proportion of childhood cancer patients with pathogenic germline variants in cancer predisposition genes and associate these genetic findings with specific childhood cancer diagnoses. This will lead to the transition of appropriate clinical genetic testing into the paediatric oncology clinic.

The Torch

A new guide for collaborative research

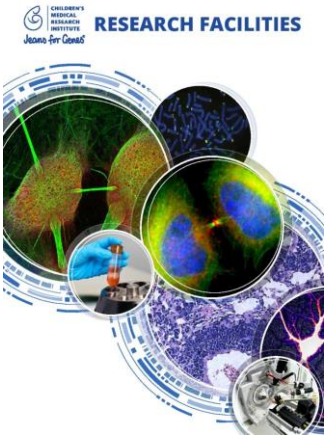
Universities Australia, the Australian Research Council and the National Health and Medical Research Council has recently published the Collaborative Research: A guide supporting the Australian Code for Responsible Conduct of Research. This guide aims to assist researchers and institutions adhere to the Australian Code for Responsible Conduct of Research when engaging in collaborative research with other Australian and International researchers and institutions.

Newborn Screening Program Abstract Accepted

Congratulations to Michelle Farrar and the Newborn Screening Program team on the acceptance of their abstract *"Real world impact of newborn screening for spinal muscular atrophy: 2 year Australian pilot"* for the 16th International Child Neurology Congress, San Diego, USA. The congress is in October 2020, so here's hoping that travel restrictions will be lifted by then.

A/Prof Michelle Farrar
Paediatric Neurology
School of Women's & Children's Health UNSW &
Lead Neuromuscular Diseases Clinical & Research Program SCHN

Children's Medical Research Institute (CMRI) Facilities



Luminesce Alliance Partner, CMRI, welcomes visiting scientists to use their equipment and core facilities, some of which include: BioResources; Stem Cell & Organoid; CellBank Australia; Vector & Genome Engineering; Biomedical Proteomics; Single Cell Analytics; Peptide Synthesis; Drug Screening; Advanced Microscopy & ATAC; Bioinformatics. Read more about CMRI Facilities.

About Luminesce Alliance

At Luminesce Alliance we aim to facilitate and drive paediatric translational medicine. Working with our partners, and engaging new partners as we grow, we seek to become a world leader in paediatric translational medicine. We will achieve this by holding true to our:

Vision - To create Australia's most ground breaking, innovative and translational paediatric research alliance that will change children's health around the world.

Mission - To have a voice on a national and international platform so that our research is disseminated and implemented to directly improve the health of children.

Purpose - To empower our partners to work together in changing the global landscape in paediatric research.

Find out more about the Alliance, what we do, who we are and keep up to date on news and events at www.luminesce.org.au We welcome your feedback and any news items you would like to be featured, simply email us at: info@luminesce.org.au

Our Partners

The Sydney children's Hospitals Network — care, advocacy, research, education
Children's Cancer Institute
Children's Medical Research Institute — Jeans for Genes®
The University of Sydney
UNSW Sydney

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