



****PROGRAM SUBJECT TO CHANGE****

Thursday 7 November 2024

08.00	Registration desk open	
08.00 - 08.45	Arrival tea and coffee	
08.45 - 09.00	Conference Opening Room: Leighton Hall Conference Opening, Luminesce Alliance Chair Ms Kathryn Greiner AO Welcome to Country, La Perouse Local Aboriginal Land Council	
09.00 - 10.45	SESSION 1 - Advanced Therapeutics Chair: Adj A/Prof Paula Bray, Sydney Children's Hospitals Network	
9.00	Keynote presentation NSW Health Research Strategy Update Dr Kim Sutherland, OHMR, NSW Health	
9.30	Perspectives from Family Members Interview Session: Emma Maly and A/Prof Wendy Gold	
9.45	Presentation The Kids Advanced Therapeutics Odyssey - Charting progress and Forging Ahead Dr Michelle Lorentzos, Sydney Children's Hospitals Network	
10.00	Panel Session - Interactive Q&A discussion session Opportunities and Challenges for Advanced Therapeutics: clinical translation, access, regulation, commercial and funding landscape Facilitator: Ms Lani Attwood, Sydney Children's Hospitals Network Panel members: Dr Michelle Lorentzos, Sydney Children's Hospitals Network Prof Ian Alexander, Children's Medical Research Institute Dr Paul Fennessy, Paul Fennessy Advisory Vanessa Fitzgerald, NSW Health	
10.45 - 11.15	Morning tea	
11.15 - 13.00	SESSION 2.1 - Precision Therapy for Cancer Therapeutic and Innovative Applications Room: Leighton Hall Chair: Prof Michelle Haber AM, Children's Cancer Institute	SESSION 2.2 - Conference Abstracts Presentations Room: Tyree Room Chair: Dr Laura Fawcett, Sydney Children's Hospitals Network
11.15	Keynote Presentation The Molecular Characterisations Initiative: precision diagnosis for paediatric cancers Prof Elaine Mardis, Institute for Genomic Medicine at Nationwide Children's Hospital	11.15 Treatments for those who have none platform: from Western Australia to nationwide - 100 Dr May Aung-Htut, Murdoch University, Centre for Molecular Medicine & Innovative Therapeutics

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11.50	<p>Setting the Australian scene: what is ZERO? Utilising precision data to drive therapeutic recommendations and to innovate discoveries Prof Michelle Haber AM, Children's Cancer Institute</p>	<p>11.30 Testing gene replacement therapy in retinitis pigmentosa and Leber congenital amaurosis - 101 Dr To Ha Loi, Children's Medical Research Institute</p>
11.55	<p>Translational Tumour Biology – utilising Cas13 technology to identify therapeutic targets using a molecular biology approach A/Prof Paul Ekert, Children's Cancer Institute</p>	<p>11.45 Pre-natal AAV gene therapy for HBSL disease in Dars1 transgenic mice - 102 Elena Venuti, UNSW Sydney</p>
12.10	<p>Innovation driving precision drug targeting discoveries – translating ideas from genetic disease into cancer Dr Antoine de Weck, Children's Cancer Institute</p>	<p>12.00 Gene Therapy for Brittle Bone Disease - 103 A/Prof Aaron Schindeler, Sydney Children's Hospitals Network</p>
12.25	<p>Utilising high-throughput drug screening for therapeutic and innovative outputs Dr Gabor Tax, Children's Cancer Institute</p>	<p>12.15 PeptAlde: Accelerating Antimicrobial Peptide Discovery Using AI Ensemble Techniques - 104 Lauren Lu, Children's Medical Research Institute</p>
12.40	<p>Panel discussion: Transitioning precision therapy research to standard clinical care. Facilitator: Prof Elaine Mardis, Institute for Genomic Medicine at Nationwide Children's Hospital Panel members: A/Prof Paul Ekert, Deputy Director – Research Themes, Children's Cancer Institute Prof David Ziegler, Sydney Children's Hospitals Network Prof Ian Street, Director, Therapeutic Innovations for Kids (THINK), Children's Cancer Institute A/Prof Ness Tyrrell, Children's Cancer Institute</p>	<p>12.30 Modelling PROM1 retinal disease in a mouse model and human retinal organoids - 105 Dr William Yates, Children's Medical Research Institute</p> <p>12.45 Using behavioural theories to implement novel communication resources for healthcare professionals - 106 Amber-Marie Firriolo, Behavioural Sciences Unit (UNSW)</p>

13.00 – 14.00

Lunch



14.00 – 15.30

SESSION 3.1 - Functional Genomics and Precision Therapy: Cell And Gene Therapy Advancements: From Bench to Bedside

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Room: Leighton Hall

Chair: Prof Roger Reddel AO

14.00 **Cell therapy pipeline, past, present, and future success**

A/Prof Anai Gonzalez-Cordero, Children's Medical Research Institute

14.15 **The world of avatar technology for cystic fibrosis and beyond**

Dr Shafagh Waters, UNSW Sydney

14.30 **Case study: Rett syndrome – from diagnosis to treatment**

A/Prof Wendy Gold, University of Sydney

14.45 **Overcoming the challenges posed by pre-existing immunity to AAV viral vectors**

A/Prof Grant Logan, Children's Medical Research Institute

SESSION 3.2 - Innovation and Commercialisation Opportunities: Paediatric Cancer Drug Discovery Innovation and the Road to Commercialisation

Room: Tyree Room

Chair: Emeritus Prof Richard Henry, UNSW Sydney

Why is novel drug discovery needed for paediatric cancer?

Dr Emily Mould, Children's Cancer Institute.

Collaborative drug discovery models: how can drug discovery for paediatric cancers be accomplished collectively?

Prof Ian Street, Children's Cancer Institute

What are the challenges in finding novel drugs for paediatric cancers?

A/Prof Antoine De Weck, Children's Cancer Institute

14:45 **Panel Session**
Barriers, challenges, and opportunities to gene and cell therapy research translation and commercialisation
Facilitator: A/Prof Wendy Gold, University of Sydney

15:00 **Panel members:**
 A/Prof Anai Gonzalez-Cordero, Children’s Medical Research Institute
 Mr Brett Anslow, Consumer Stem Cell Academic TBC
 Commercialisation experience TBC

14.45 Panel Session
Overcoming the roadblocks and achieving a sustainable model of paediatric cancer drug discovery and commercialisation.
Facilitator: Emeritus Prof Richard Henry

Panel members:
 A/Prof Greg Arndt, Head of Drug Discovery Biology, THINK, Children’s Cancer Institute
 Prof David Ziegler, Sydney Children’s Hospitals Network
 Dr Julia Warning, NSW Health
 Brandon Capital Partners representative
 Industry representative TBC

15.30 - 16.00 Afternoon tea



16:00 – 17:30 **SESSION 4.1 No one left behind: truly engaging families in precision medicine**
Room: Leighton Hall
Chair: Prof Claire Wakefield, UNSW Sydney

SESSION 4.2 Innovation and Commercialisation Opportunities
Room: Tyree Room
Chair: Lani Attwood, Sydney Children’s Hospitals Network

16:00 **Co-designing and co-imagining the future of precision medicine – optimal patient-centred care**
 Kris Pierce, Child Unlimited

16:00 **NAT Network – Non-animal technology network**
 Dr Shafagh Waters, UNSW Sydney

16:15 **Psychosocial Enabling Platform – purpose, current and future activities**
 Dr Kate Hetherington, UNSW Sydney

16:15 **Health biobanking in NSW- maximising efficiency and research support**
 Prof Jennifer Byrne, NSW Health Pathology

16:30 **Psychosocial Precision Medicine – Addressing the Social Determinants of Health**
 Prof Sue Woolfenden, Sydney Local Health District and University of Sydney

16:30 **Clinical trials and commercialisation**
 Anne O’Neill, Director, Enterprise, International Partnerships and Clinical Trials, NSW Health

16:45 **Panel Session**
Doing a better job of communicating with families: addressing challenges and driving solutions
Facilitator: Prof Claire Wakefield
Panel members:
 Emeritus Prof Les White, Cancer Institute NSW
 Kris Pierce, Child Unlimited
 Seaneen Wallace, Sydney Children’s Hospitals Network
 Dr Clarissa Schilstra, UNSW Sydney

16:45 **AAV development and translation**
 A/Prof Leszek Lisowski, Children’s Medical Research Institute

17:10 **Rapid Fire Presentations (5 minutes each)**
Adapting SIBS-ONLINE for Australian siblings of patients with genetic epilepsy - 107
 Dr Lauren Kelada, UNSW Sydney
Partnering with consumers to advance research and impact for inherited retinal diseases - 108
 Dr Eden Robertson, UNSW Sydney
Trio germline sequencing in childhood cancer: Healthcare professionals' perspectives from PREDICT - 109
 Jacqueline Hunter, University of Melbourne/University of New South Wales

17:00 **Functional Genomics Enabling Platform Showcase: Facility, Laboratory Services, and Equipment to Deliver New Research Capacity**
 Josh Studdert, Children’s Medical Research Institute

17:30 **Close**



Friday 8 November 2024

08:00 **Registration desk open**

08.00 - 08.45 **Arrival tea and coffee**

08.45 - 10.30 **SESSION 5.1 Precision medicine informed by real world data**

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Room: Leighton Hall

Chair: Dr Teresa Anderson AM, Chief Executive, SDPR Implementation Authority

08.45 **Practicalities of using real-world electronic health data for research: insights from the UK, Great Ormond Street Hospital clinical informatics research program**

Prof Neil Sebire, Great Ormond Street Hospital Institute of Child Health

09.15 **Advanced genome and transcriptome analytics - using comprehensive molecular profiling techniques accelerating the uptake of precision medicine**

A/Prof Mark Cowley, Children's Cancer Institute

09.30 **Advanced clinical data analytics: creating an ecosystem to access, extract and interpret clinical eMR and administrative data**

Prof Tom Snelling, University of Sydney

09.45 **Kids Link: Utilising data linkage to advance data-driven healthcare to strengthen our health system**

Prof Natasha Nassar, University of Sydney

10.00 **NCI Australia's Precision Medicine Infrastructure and Clinic Genomics Services**

Dr Warren Kaplan, NCI

10.10 **Panel Session**

Harnessing real-world data for patient benefit

Facilitator: Dr Teresa Anderson AM, Chief Executive, SDPR Implementation Authority

Panel members:

Prof Neil Sebire, Great Ormond Street Hospital Institute of Child Health

A/Prof Mark Cowley, Children's Cancer Institute

Prof Tom Snelling, University of Sydney

Prof Natasha Nassar, University of Sydney

SESSION 5.2 Kids Advanced Therapeutics Pipeline
N=1 clinical trials in paediatric advanced therapies

Room: Tyree Room

Chair/Facilitator: Adj. Associate Professor Paula Bray, Sydney Children's Hospitals Network

The session will explore the opportunities and challenges of N=1 clinical trials in paediatric advanced therapies, including clinical translation, patient access, ethical considerations, and scientific innovations

Panel members:

A/Prof Leszek Lisowski, Children's Medical Research Institute

Prof Adam Jaffe, Sydney Children's Hospitals Network

Dr Richard Webster, Sydney Children's Hospitals Network

Dr May Aung-Htut, Monash University

A/Prof Sarah Garnett, Sydney Children's Hospitals Network

Ms Kris Pierce, Child Unlimited



Posters:

- P101 Steps to resolution of variants of uncertain significance in RPE65**
Dr Steven Eamegdool, Children's Medical Research Institute
- P102 Genetic editing applications for variant classification and therapy development for cone-rod dystrophies**
Miss Reeva Nadkar, Children's Medical Research Institute
- P103 Estimating population prevalence of rare diseases in children using administrative health data**
Dr Carolyn Arico, The University of Sydney
- P104 Systematic review of population-based studies investigating rare diseases in children**
Dr Carolyn Arico, The University of Sydney
- P105 AAV-mediated DARS1 gene replacement in preclinical models of the leukodystrophy HBSL**
Miss Elizabeth Kalotay and Dr Dominik Fröhlich, UNSW Sydney
- P106 Preclinical assessment of gene therapy for RARS2-related early onset epileptic encephalopathy**
Ms Puja Thapa, The University of Sydney
- P107 PRECISE – Co-designing a genomics and precision medicine program for primary care**
Dr Alan Ma, The University of Sydney and Sydney Children's Hospitals Network
- P108 Newborn Screening for Duchenne muscular dystrophy: Development of a scoping study**
Miss Sarah Shin, Sydney Children's Hospital Network
- P109 From recommendation to access: Improving procurement of precision-guided treatments with implementation science**
Mr Patrick Nay, Kids Cancer Centre, Sydney Children's Hospital
- P110 Co-developing positive psychology resources with caregivers of children with developmental epileptic encephalopathies**
Dr Suzanne Nevin, UNSW Sydney
- P111 A pilot study of creating a long-term follow-up clinical registry and biorepository of patients undergoing cell and gene therapies**
Dr Laszlo Irinyi, NSW Health Pathology
- P112 Gene therapy proof-of-concept for spastic paraplegia 56**
Miss Richa Chaluvadi and Dr Dominik Froehlich, UNSW Sydney

11.00-13.00

SESSION 6.1 HSIE and Precision Therapy - Delivering outcomes focused sustainability for children in the era of precision medicine

Room: Leighton Hall

Chair: Adj A/Prof Cathryn Cox PSM, Sydney Children's Hospitals Network

11.00

The need to evaluate health services innovations at scale: challenges and opportunities in the US and Australia

Prof Gary L Freed, University of Michigan

11.30

Health systems implementation and health economics –Child Health services research and innovations

Prof Raghu Lingam, UNSW Sydney

11.45

What is PEACH and PEACH-E? Current Update and Next Steps

Prof Karen Zwi & Ms Seaneen Wallace, Sydney

11:00 – 13:00

SESSION 6.2 Conference Abstracts Presentations

Room: Tyree Room

Chair: Dr Radoslaw (Radek) Szmyd, Children's Medical Research Institute

11.00 Harnessing liquid biopsy to unveil somatic RAS-MEK pathway variants in extracranial AVMs - 202

Dr Smadar Kahana-Edwin, The Children's Hospital at Westmead

11.15 Cryptico: A Computational Approach for Exploring Cryptons in Search of Novel Therapeutics - 203

Dr Pablo Acera, Children's Cancer Institute

11.30 Inferring the life history of NTRK-fusion paediatric tumours - 204

Dr Piyushkumar Mundra, Children's Cancer Institute

12.00 Panel Session
Thinking outside the silos: innovations to optimise the health system for children
Facilitator: Adj A/Prof Cathryn Cox PSM, Sydney Children's Hospitals Network
Panel members:
 Prof Raghu Lingam, UNSW Sydney
 Prof Gary L Freed, University of Michigan
 Prof Sue Woolfenden, University of Sydney
 Dr Helen Goodwin, NSW Health
 Dr Sophy Shih, Kirby Institute

12.30 Abstract Presentations
The consumer group landscape: identifying advocacy gaps and improving precision medicine outcomes - 200
 Tiana Kittos, Child Unlimited
Paediatric service navigation to address life challenges: a mixed-methods study - 201
 Rebecca Bosward, Sydney Local Health District

11.45 CBL mutations in CNS and solid tumours - a new therapeutic opportunity? - 205
 Dr Lauren Brown, Children's Cancer Institute

12.00 Identifying non-coding lncRNA dependencies in high-risk MLL-rearranged acute myeloid leukaemia - 206
 Steven He, Children's Cancer Institute

12.15 Individualized-tumour-informed panel approach enables ultra-sensitive ctDNA-based minimal disease monitoring for paediatric cancers - 207
 Dr Wenhan Chen, Children's Cancer Institute

12.30 A Data Lakehouse for Childhood Cancer Precision Medicine - 208
 James Bradley, Children's Cancer Institute


12.45 PREDICT and Validate: Investigating Germline Alterations in Paediatric Cancer Predisposition [RAPID] - 209
 Safaa Al Haj Hussein, The Children's Hospital At Westmead, The University Of Sydney

13.00 - 14.00 **Lunch**



14.00 - 15.30 SESSION 7.1 The road ahead in genomics and genetic testing
Room: Leighton Hall
Chair: Prof Adam Jaffe, UNSW Sydney
14.00 The future direction for genomics testing: a global perspective on improving access to genomics knowledge and technology
 Tiffany Boughtwood, Australian Genomics
14.15 Genomic testing - what is the roadmap for Australia
 Dr Emma Palmer, Sydney Children's Hospitals Network
14.30 The State of Rare Diseases in Australia
 Prof Adam Jaffe, UNSW Sydney
14.45 Abstract Presentation: A collaborative implementation science approach to genomics and precision medicine multidisciplinary teams - 210
 Dr Alan Ma, University of Sydney/Sydney Children's Hospitals Network
15.10 Panel Session
Challenges of sustained translation in genomic screening and testing: battle scars, lessons learned, and priority actions to move forward
Facilitator: Prof Adam Jaffe, UNSW Sydney
Panel members:
 Tiffany Boughtwood, Australian Genomics
 A/Prof Ness Tyrrell, Children's Cancer Institute
 Prof Claire Wakefield, UNSW Sydney
 Adj A/Prof Paula Bray, Sydney Children's Hospitals Network

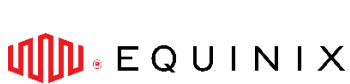
SESSION 7.2 - Health Systems Implementation and Economics (HSIE) & Psychosocial Workshop
Room: Tyree Room
Facilitator: Prof Raghu Lingam, UNSW Sydney.
 Summary: Health systems and interventions are complex, with complex humans at the core as both providers and receivers.
 Prof Lingam will provide an engaging session on health systems implementation research, exploring frameworks for the development and evaluation of complex health interventions. Ms Wallace, Dr Ostojic alongside two research advisors will explore the intricacies and key considerations of 'co-design' and involvement of consumers in the planning, design and evaluation of health interventions.
 Prof Raghu Lingam, UNSW Sydney
 Seaneen Wallace, Sydney Children's Hospitals Network
 Dr Katarina Ostojic, University of Sydney
 Consumer research advisors - Mr Jack Calderan (Young Adult Research Advisor) and Ms Alunya Wilkinson (Parent Research Advisor)
 Prof Gary L Freed, University of Michigan

	Dr Emma Palmer, Sydney Children's Hospitals Network	
15.30 - 16.00	Afternoon tea	
16.00 - 16.30	<p>Panel discussion: Reflections and next steps for paediatric precision medicine</p> <p>Facilitator: Kathryn Greiner AO</p> <p>Panel members:</p> <p>Prof Michelle Haber AM</p> <p>Prof Roger Reddell AO</p> <p>Adj A/Prof Cathryn Cox PSM</p> <p>Prizes</p>	
16.45	Conference close	

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