



****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, Luminesce Alliance Chair Ms Kathryn Greiner AO	
	Room: Leighton Hall	
	Welcome to Country, Aunty Maxine Ryan, La Perouse Local Aboriginal Land Council	
09.00 - 09.02	Ms Kathryn Greiner AO - Introduction of Keynote Speaker	
9.02	Keynote presentation	
	NSW Health Research and Innovation Strategy	
	Dr Kim Sutherland, OHMR, NSW Health	
9.37	Perspectives from Family Members	
	Interview Session: Mrs Emma Maly and A/Prof Wendy Gold, University of Sydney	
09.54	SESSION 1 - Advanced Therapeutics	
	Chair: Adj A/Prof Paula Bray, Sydney Children's Hospitals Network	
09.56	Presentation	
	The Kids Advanced Therapeutics Odyssey - Charting progress and Forging Ahead	
	Dr Michelle Lorentzos, Sydney Children's Hospitals Network	
10.06	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	
	Ms Vanessa Fitzgerald, NSW Health	
10.45 - 11.15	Morning tea	
	Sponsored by NOVARTIS	
11.15 - 13.00	SESSION 2.1 - Precision Therapy for Cancer Therapeutic and Innovative Applications	SESSION 2.2 - Conference Abstracts Presentations
	Session Sponsor	Room: Tyree Room
	EQUINIX	Chair: Dr Laura Fawcett, Sydney Children's Hospitals Network
	Room: Leighton Hall	
	Chair: Prof Michelle Haber AM, Children's Cancer Institute	
11.17	Keynote Presentation	11.15 Treatments for those who have none platform:

Session sponsor



	<p>The Molecular Characterisations Initiative: precision diagnosis for paediatric cancers</p> <p>Prof Elaine Mardis, Institute for Genomic Medicine at Nationwide Children's Hospital</p>	<p>from Western Australia to nationwide - 100</p> <p>Dr May Aung-Htut, Murdoch University, Centre for Molecular Medicine & Innovative Therapeutics</p>
11.52	<p>Setting the Australian scene: what is ZERO? Utilising precision data to drive therapeutic recommendations and to innovate discoveries</p> <p>Prof Michelle Haber AM, Children's Cancer Institute</p>	<p>11.30 Testing gene replacement therapy in retinitis pigmentosa and Leber congenital amaurosis - 101</p> <p>Dr To Ha Loi, Children's Medical Research Institute</p>
11.57	<p>Translational Tumour Biology – utilising Cas13 technology to identify therapeutic targets using a molecular biology approach</p> <p>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</p> <p>Miss Elena Venuti, UNSW Sydney</p>
12.10	<p>Innovation driving precision drug targeting discoveries – translating ideas from genetic disease into cancer</p> <p>A/Prof Antoine de Weck, Children's Cancer Institute</p>	<p>12.00 Gene Therapy for Brittle Bone Disease - 103</p> <p>A/Prof Aaron Schindeler, Sydney Children's Hospitals Network</p>
12.23	<p>Utilising high-throughput drug screening for therapeutic and innovative outputs</p> <p>Dr Gabor Tax, Children's Cancer Institute</p>	<p>12.15 PeptAlde: Accelerating Antimicrobial Peptide Discovery Using AI Ensemble Techniques - 104</p> <p>Ms Lauren Lu, Children's Medical Research Institute</p>
12.36	<p>Introduction from Mark Cowley to Kieran Habojan, Equinix</p>	
12.40	<p>Panel discussion: Transitioning precision therapy research to standard clinical care</p> <p>Facilitator: Prof Elaine Mardis, Institute for Genomic Medicine at Nationwide Children's Hospital</p> <p>Panel members:</p> <p>A/Prof Paul Ekert, Deputy Director – Research Themes, Children's Cancer Institute</p> <p>Prof David Ziegler, Sydney Children's Hospitals Network</p> <p>Prof Ian Street, Director, Therapeutic Innovations for Kids (THINK), Children's Cancer Institute</p> <p>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</p> <p>Dr William Yates, Children's Medical Research Institute</p> <p>12.45 Using behavioural theories to implement novel communication resources for healthcare professionals - 106</p> <p>Miss Amber-Marie Firriolo, Behavioural Sciences Unit (UNSW)</p>
13.00 – 14.00	<p>Lunch Sponsored by  NOVARTIS</p>	
14.00 – 15.30	<p>SESSION 3.1 - Functional Genomics and Precision Therapy: Cell And Gene Therapy Advancements: From Bench to Bedside</p> <p>Session Sponsor</p>  <p>Room: Leighton Hall</p> <p>Chair: Prof Roger Reddel AO</p>	<p>SESSION 3.2 - Innovation and Commercialisation Opportunities: Paediatric Cancer Drug Discovery Innovation and the Road to Commercialisation</p> <p>Room: Tyree Room</p> <p>Chair: Emeritus Prof Richard Henry AM, UNSW Sydney</p>
14.02	<p>Cell therapy pipeline, past, present, and future success</p> <p>A/Prof Anai Gonzalez-Cordero, Children's Medical Research Institute</p>	<p>14:02 Why is novel drug discovery needed for paediatric cancer?</p> <p>Dr Emily Mould, Children's Cancer Institute.</p>
14.15	<p>The world of avatar technology for cystic fibrosis and beyond</p> <p>Dr Shafagh Waters, UNSW Sydney</p>	<p>14.17 Developing New Therapeutics for Kids with Cancer</p> <p>Prof Ian Street, Children's Cancer Institute</p>

14.28	<p>Case study: Rett syndrome – from diagnosis to treatment</p> <p>A/Prof Wendy Gold, University of Sydney</p>	<p>14.32 What are the challenges in finding novel drugs for paediatric cancers?</p> <p>A/Prof Antoine De Weck, Children’s Cancer Institute</p>
14.41	<p>Overcoming the challenges posed by pre-existing immunity to AAV viral vectors</p> <p>A/Prof Grant Logan, Children’s Medical Research Institute</p>	
14:55	<p>Panel Session</p> <p>The Family Journey: Challenges and Opportunities in Rare Genetic Disease Care</p> <p>Facilitator: A/Prof Wendy Gold, University of Sydney</p> <p>Panel members:</p> <p>Mr Brett Anslow, Parent</p> <p>Lauren Jensen, Parent</p> <p>Ms Kris Pearce, Parent and Child Unlimited Director</p>	<p>14.47 Panel Session</p> <p>Overcoming the roadblocks and achieving a sustainable model of paediatric cancer drug discovery and commercialisation.</p> <p>Facilitator: Emeritus Prof Richard Henry AM</p> <p>Panel members:</p> <p>A/Prof Greg Arndt, Head of Drug Discovery Biology, THINK, Children’s Cancer Institute</p> <p>Prof David Ziegler, Sydney Children’s Hospitals Network</p> <p>Dr Julia Warning, NSW Health</p> <p>Dr Jess Smith, Brandon Capital Partners</p>
15.30 - 16.00	<p>Afternoon tea Sponsored by  NOVARTIS</p>	
16:00 – 17:30	<p>SESSION 4.1 No one left behind: truly engaging families in precision medicine</p> <p>Room: Leighton Hall</p> <p>Chair: Prof Claire Wakefield, UNSW Sydney</p>	<p>SESSION 4.2 Innovation and Commercialisation Opportunities</p> <p>Room: Tyree Room</p> <p>Chair: Ms Lani Attwood, Sydney Children’s Hospitals Network</p>
16.02	<p>Transforming patient care through genuine partnerships with families in precision medicine</p> <p>Ms Kris Pierce, Child Unlimited</p>	<p>16:00 NAT Network – Non-animal technology network</p> <p>Dr Shafagh Waters, UNSW Sydney</p>
16.17	<p>Psychosocial Enabling Platform – purpose, current and future activities</p> <p>Dr Kate Hetherington, UNSW Sydney</p>	<p>16:15 Health biobanking in NSW- maximising efficiency and research support</p> <p>Prof Jennifer Byrne, NSW Health Pathology</p>
16.32	<p>Psychosocial Precision Medicine – Addressing the Social Determinants of Health</p> <p>Prof Sue Woolfenden, Sydney Local Health District and University of Sydney</p>	<p>16:30 Clinical trials and commercialisation</p> <p>Anne O’Neill, Director, Enterprise, International Partnerships and Clinical Trials, NSW Health</p>
16.47	<p>Panel Session</p> <p>Doing a better job of communicating with families: addressing challenges and driving solutions</p> <p>Facilitator: Prof Claire Wakefield</p> <p>Panel members:</p> <p>Emeritus Prof Les White, Cancer Institute NSW</p> <p>Ms Kris Pierce, Child Unlimited</p> <p>Ms Seaneen Wallace, Sydney Children’s Hospitals Network</p> <p>Dr Clarissa Schilstra, UNSW Sydney</p>	<p>16:45 AAV development and translation</p> <p>A/Prof Leszek Lisowski, Children’s Medical Research Institute</p>
17.12	<p>Rapid Fire Presentations (5 minutes each)</p> <p>Adapting SIBS-ONLINE for Australian siblings of patients with genetic epilepsy - 107</p> <p>Dr Lauren Kelada, UNSW Sydney</p> <p>Partnering with consumers to advance research and impact for inherited retinal diseases - 108</p>	<p>17:00 Functional Genomics Enabling Platform Showcase: Facility, Laboratory Services, and Equipment to Deliver New Research Capacity</p> <p>Josh Studdert, Children’s Medical Research Institute</p>



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**
Session Sponsor



Room: Leighton Hall

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

08.49 **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

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

A/Prof Mark Cowley, Children's Cancer Institute

09.37 **Accessing, extracting and interpreting eMR data**

Prof Tom Snelling, University of Sydney

09.50 **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

08.45 – 10.30

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

Room: Tyree Room

Chair/Facilitator: Adj. A/Prof 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:

Dr Andrea Perez-Iturralde, 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, Murdoch University

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

Ms Kris Pierce, Child Unlimited

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

10.30 – 11.00

Morning tea and Poster Presentations

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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 – 13:00

SESSION 6.2 Conference Abstracts Presentations

Room: Tyree Room

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

11.02

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

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

	Prof Gary L Freed, University of Michigan	Westmead
11.37	Strengthening Child Health Systems Prof Raghu Lingam, UNSW Sydney	11.15 Cryptico: A Computational Approach for Exploring Cryptons in Search of Novel Therapeutics - 203 Dr Pablo Acera, Children's Cancer Institute
11.55	What is PEACH and PEACH-E? Current Update and Next Steps Prof Karen Zwi & Ms Seaneen Wallace, Sydney Children's Hospitals Network	11.30 Inferring the life history of NTRK-fusion paediatric tumours - 204 Dr Piyushkumar Mundra, Children's Cancer Institute
12.08	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	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 Mr 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
12.33	Abstract Presentations The consumer group landscape: identifying advocacy gaps and improving precision medicine outcomes - 200 Ms Kris Pierce, Child Unlimited Paediatric service navigation to address life challenges: a mixed-methods study - 201 Ms Rebecca Bosward, Sydney Local Health District	12.30 A Data Lakehouse for Childhood Cancer Precision Medicine - 208 Mr James Bradley, Children's Cancer Institute 12.45 PREDICT and Validate: Investigating Germline Alterations in Paediatric Cancer Predisposition [RAPID] - 209 Miss Safaa Al Haj Hussein, The Children's Hospital at Westmead, The University of Sydney
13.00 - 14.00	Lunch Sponsored by	
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 - 15.30 SESSION 7.2 - Health Systems Implementation and Economics (HSIE) & Psychosocial Workshop Room: Tyree Room
14.03	The future direction for genomics testing: a global perspective on improving access to genomics knowledge and technology Tiffany Boughtwood, Australian Genomics	Facilitator: Prof Raghu Lingam, UNSW Sydney Summary: Health systems and interventions are complex, with complex humans at the core as both providers and receivers.
14.18	Genomic testing - what is the roadmap for Australia Dr Emma Palmer, Sydney Children's Hospitals Network	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.
14.33	The State of Rare Diseases in Australia Prof Adam Jaffe, UNSW Sydney	Prof Raghu Lingam, UNSW Sydney Ms Seaneen Wallace, Sydney Children's Hospitals Network Dr Katarina Ostojic, University of Sydney
14.48	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	Consumer research advisors - Mr Jack Calderan (Young Adult Research Advisor) and Ms Alunya Wilkinson (Parent Research Advisor)
15.03	Panel Session Challenges of sustained translation in genomic screening and testing: battle scars, lessons learned, and priority actions to move forward	Prof Gary L Freed, University of Michigan

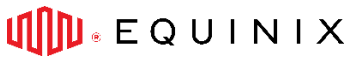
Facilitator: Prof Adam Jaffe, UNSW Sydney
Panel members:
 Ms Tiffany Boughtwood, Australian Genomics
 A/Prof Ness Tyrrell, Children's Cancer Institute
 Prof Claire Wakefield, UNSW Sydney
 Adj A/Prof Paula Bray, Sydney Children's Hospitals Network
 Dr Emma Palmer, Sydney Children's Hospitals Network

15.30 - 16.00	Afternoon tea	Sponsored by	
16.00 - 16.30	Panel discussion: Reflections and next steps for paediatric precision medicine Facilitator: Ms Kathryn Greiner AO Panel members: Prof Michelle Haber AM Prof Roger Reddell AO Prizes	Sponsored by	
16.45	Conference close - Chair Ms Kathryn Greiner AO		

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