14th International Congress of Inborn Errors of Metabolism 2021

'Transforming Rare Disorders'

21-24 November 2021

AEDT	SUNDAY 21 NOVEMBER 2021	AEDT	IST	CET (Saturday)	CST (Saturday)
07:00	**Opening and Welcome Address** Including welcome by the Honourable Minister Greg HUNT MP *Minister for Health and Aged Care, Australia*	07:00	1:30	21:00	14:00
	NEW APPROACHES TO DIAGNOSIS				
07:30-08:12	**Diagnoses and discoveries: Insights from the NIH Undiagnosed Diseases Program** **Dr William A. GAHL** *National Human Genome Research Institute*	07:30	2:00	21:30	14:30
08:12-08:30	**Novel disorders discovered by whole genome sequencing** **Professor Shigeo KURE** *Tohoku University School of Medicine*	08:12	2:42	22:12	15:12
08:30-09:00	**Multi-Omic approaches aid identification of cryptic variants underlying mitochondrial disorders** **Professor David THORBURN** *Murdoch Children's Research Institute*	08:30	3:00	22:30	15:30
09:00-09:18	**Metabolomics** **Professor Bart GHESQUIÈRE** *KU Leuven Metabolomics Expertise Center*	09:00	3:30	23:00	16:00
09:18-09:45	**Metabolics and genetics: Systems biology?** **Professor Henri Gerrit (Han) BRUNNER** *Radboud University Nijmegen Medical Center and Maastricht University Medical Center*	09:18	3:48	23:18	16:18
09:45-10:45	*POSTER VIEWING BREAK*	09:45	4:15	23:45	16:45
	SCREENING				
10:45-11:15	**Reproductive genetic carrier screening for families with inborn errors of metabolism** **Professor Edwin KIRK** *Sydney Children's Hospital, New South Wales Health Pathology Randwick Genomic Laboratory*	10:45	5:15	0:45 (Sunday)	17:45
11:15-11:45	**Population based newborn screening - The future** **Associate Professor Veronica WILEY** *Children's Hospital, Westmead*	11:15	5:45	1:15	18:15
11:45-12:11	**Outcome studies in newborn screening: How lessons learned can guide screening strategy** **Professor Stefan KÖLKER** *Heidelberg University Hospital*	11:45	6:15	1:45	18:45
12:11-12:36	**Ultra-rapid genomic sequencing in the newborn** **Dr David DIMMOCK** *Rady Children's Institute for Genomic Medicine*	12:11	6:41	2:11	19:11
12:36-13:01	**Ethical issues in genomic consent for high-risk patients** **Associate Professor Curtis COUGHLIN** *University of Colorado School of Medicine*	12:36	7:06	2:36	19:36
13:01-13:30	*BREAK*	13:01	7:31	3:01	20:01
	SPONSORED SYMPOSIUMS				
	SYMPOSIUM HALL A / **SYMPOSIUM HALL B**				
13:30-14:30	**Understanding ASMD from our experiences in Gaucher disease: from the science to the clinical practice** **Prof Pramod MISTRY, Dr Michel TCHAN, Prof Maurizio SCARPA** Presented by Sanofi Genzyme SANOFI GENZYME **Newborn Screening for Spinal Muscular Atrophy & Recommended Uniform Screening Panel in the US: from SCID to SMA, and beyond** **Veronica WILEY and Mei BAKER** Presented by Perkin Elmer PerkinElmer	13:30	8:00	3:30	20:30
	PARALLEL SESSIONS				
	CONCURRENT HALL 1 / **CONCURRENT HALL 2** / **CONCURRENT HALL 3**				
	Fat Oxidation and GSD / **Neurotransmitter disorders** / **Clinical Mitochondrial Disorders**				
14:30-16:00	Restoration of interaction between FAO and ETC proteins in VLCAD and VLCAD-deficient mice mitochondria by addition of recombinant VLCAD **Dr Yudong WANG** The first consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies **Prof Thomas OPLADEN** Australian Childhood Mitochondrial Epidemiology Study- Preliminary Results **Dr Carolyn BURSLE**	14:30	9:00	4:30	21:30
	Novel mouse model of Long-chain 3-hydroxyacylCoA dehydrogenase deficiency (LCHADD) retinopathy **Mr Garen GASTON** A phase 1/2, open-label, randomized, intra-subject escalation study to evaluate the safety and efficacy of PTC923 in primary tetrahydrobiopterin deficient subjects with hyperphenylalaninemia **Prof Nicola LONGO** A novel variant in CRLS1 is associated with fatal infantile cardiomyopathy, encephalopathy, sensorineural hearing loss, bull's eye maculopathy and nephrogenic diabetes insipidus **Dr Shanti BALASUBRAMANIAM**				
	Preclinical evidence of efficacy of heptanoate and derivatives as anaplerotic therapy for medium-chain acyl-CoA dehydrogenase deficiency **Prof Al-Walid MOHSEN** Evaluation of 3-O-methyldopa in dried blood spots for the diagnosis of aromatic L-amino-acid decarboxylase (AADC) deficiency in newborn screening and at-risk screening **Prof Thomas OPLADEN** Clinical spectrum of F-box and Leucine-rich repeat protein 4 (FBXL4) related mitochondrial DNA Depletion Syndrome **Dr Rachel WONG**				
	Genotype-phenotype correlations in CPT1A deficiency detected by newborn screening in Pacific populations **Dr Isaac BERNHARDT** Interim Results of the Vigilan Observational Study: Clinical Characteristics of Creatine Transporter Deficiency **Dr Judith MILLER** Mitochondrial DNA depletion syndromes: a case series of a single center experience **Dr TuĞÇe KURTARANER**				
	Sudden cardiac arrest in young adult patients with long-chain 3-hydroxyacyl CoA dehydrogenase (LCHAD) deficiency, a retrospective case series report **Dr Gabriela ELIZONDO** Comprehensive insights expanding the phenotypic spectrum of inherited disorders of biogenic amines **Dr Oya KUSEYRI HüBSCHMANN** Characterisation of the neurological phenotype and long-term outcome of GLRX5 associated iron-sulfur cluster deficiency in four cases **Dr Bindu Parayil SANKARAN**				
	Complications potentially associated with dietary management of Glycogen Storage Disease Type Ia (GSDIa) **Dr Eliza KRUGER** Stem cell modelling of GTPCH deficiency in neural progenitor cells reveals changes in antioxidant capacity in affected cells **Mrs Lena NEUMANN, Dr Sabine JUNG-KLAWITTER** An exercise intervention in children with mitochondrial skeletal muscle disorders **Ms Nancy VAN DOORN**				
16:00-16:30	**NETWORKING AND TRIVIA BREAK**	16:00	10:30	6:00	23:00
	PARALLEL SESSIONS				
	CONCURRENT HALL 1 / **CONCURRENT HALL 2** / **CONCURRENT HALL 3**				
	Organic acidaemias / **Laboratory LSDs** / **PKU and urea cycle disorders**				
16:30-18:00	Spectrum and characterization of bi-allelic variants in MMAB causing cblB-type methylmalonic aciduria **Dr Patrick FORNY** Newborn screening for LSDs in Brazil: Pilot Study Update **Dr Francyne KUBASKI** Investigating the urinary metabolome in 48 phenylketonuria (PKU) patients by nuclear magnetic resonance (NMR) spectroscopy: association of genotype phenotype value (GPV) with metabolomic results. **A/Prof Friedrich TREFZ**	16:30	11:00	6:30	23:30
	Integrated multi-omic analysis of a rare inborn error of metabolism **Dr Patrick FORNY** Neurofilament light chain and glial fibrillary acidic protein: reliable biomarkers in metachromatic leukodystrophy and the importance of paediatric reference values **Ms Shanice BEEREPOOT** Genotype-based phenotype prediction in phenylketonuria (PKU) patients **Prof Nenad BLAU**				
	Protein load aggravates renal mitochondrial dysfunction in propionic aciduria **Dr Anke SCHUMANN** Quantification of lysosphingomyelin and lysosphingomyelin-509 for the screening of acid sphingomyelinase deficiency **Dr Francyne KUBASKI** Metabolic situation in 26 Chilean PKU adult with compliance and noncompliance treatment **Dr María LEAL-WITT**				
	Persistent CSF biochemical abnormalities in transplanted patients with methylmalonic acidemia: a longitudinal study. **Dr Diego MARTINELLI** Clinical phenotype and molecular analysis for 16 Cases of Galactosialidosis **Dr Toko SHIBUYA** Increase in dietary phenylalanine tolerance following demonstrated response to Sapropterin testing – the Queensland experience of sapropterin-responsive children under 18 years with phenylketonuria **Ms Aoife ELLIOTT**				
	Ethylmalonic acid impairs glutamate oxidation and induces mitochondrial permeability transition in rat cerebellum **Prof Guilhian LEIPNITZ** Iduronate-2-sulfatase transport vehicle restores peripheral and central fluid biomarkers in a mouse model of Hunter Syndrome **Dr Annie ARGUELLO** Efficacy and safety of the recommended pegvaliase dosing regimen in adults with phenylketonuria in the phase 3 PRISM studies **Dr Stephanie SACHAROW**				
	Bezafibrate prevents redox and mitochondrial homeostasis deregulation in brain of glutaryl-CoA dehydrogenase deficient mice **Prof Guilhian LEIPNITZ** Frequency of carriers for rare recessive Mendelian diseases in a Brazilian cohort of 320 patients **Dr Caio Robledo QUAIO** Safety and efficacy of DTX301, an AAV8 gene therapy, in adults with late-onset ornithine transcarbamylase (OTC) deficiency: results from a phase 1/2 clinical trial **Prof Cary O. HARDING**				
	DAY ONE CLOSE				
18:00-20:00	*Welcome reception - In person Delegates only*				

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'Transforming Rare Disorders'

21-24 November 2021

AEDT	MONDAY 22 NOVEMBER 2021	AEDT	IST	CET (Sunday)	CST (Sunday)
07:00-07:30	**Online Q&A - New approaches to diagnosis** **Chaired by Eva Morava- Kozicz and Janice Fletcher**	7:00	1:30	21:00	14:00
07:30-08:00	**Online Q&A - Screening** **Chaired by Carmencita Padilla and Dianne Webster**	07:30	2:00	21:30	14:30
	NEW THERAPIES				
08:00-08:27	**Liver targeted gene therapy - an explosion of possibilities** **Professor Ian E. ALEXANDER** *Sydney Children's Hospital Network*	08:00	2:30	22:00	15:00
08:27-08:59	**Treatment of metabolic disorders using genomic technologies** **Professor Charles VENDITTI** *National Institutes of Health*	08:27	2:57	22:27	15:27
08:59-09:29	**CNS targeted gene therapy: Update on gene therapy for lysosomal diseases** **Associate Professor Steven GRAY** *University of Texas Southwestern Medical Center*	08:59	3:29	22:59	15:59
09:29-10:01	**Novel approaches in treatment of CDG: Bringing therapies from bench to bedside** **Professor Eva MORAVA** *Mayo Clinic School of Medicine*	09:29	3:59	23:29	16:29
10:01-10:20	***POSTER VIEWING BREAK***	10:01	4:31	0:01 (Monday)	17:01
10:20-10:45	**NETWORKING AND TRIVIA BREAK**	10:20	4:50	0:20	17:20

	ALLIED HEALTH	ADULT METABOLIC	LABORATORY				
	CONCURRENT HALL 1	**CONCURRENT HALL 2**	**CONCURRENT HALL 3**				
10:45-13:15	Management of GSD, where are we now and where are we heading **Prof Priya KISHANI**	Competencies in adult metabolic medicine **Dr Sandra SIRRS, Dr Annalisa SECHI**	Improving diagnosis and screening: biochemical assays versus genomic applications **Prof Maria FULLER**	10:45	5:15	0:45	17:45
		Late-onset Pompe Disease: a systematic review and meta-analysis **Dr Ida Vanessa SCHWARTZ**					
	Centre based approach to the management of hepatic GSD **Miss Gemma HACK, Ms Joanna GRIBBEN**	Safety of home-based infusion of alglucosidase alfa in late-onset Pompe disease: 13 years of experience from the Erasmus MC university medical center **Miss Imke DITTERS**	Advancing the quality of diagnostic testing for Inherited Metabolic Diseases – the role of ERNDIM EQA schemes **Dr George RUIJTER, Dr Joanne CROFT**				
		Urea cycle disorders: analysis of the outcomes in the adult population **Dr Nicola VITTURI**	Laboratory experience in diagnosing galactosialidosis using matrix-assisted laser desorption ionization time of flight mass spectrometry (MALDI-TOF MS) and post analytical pattern recognition software **Ms Gisele PINO**				
	Center based approach of the management of hepatic GSD **Mrs Foekje DE BOER, Mrs Caroline VAN DER SCHAAF**	Natural history and complications of hereditary fructose intolerance in adult patients. **Dr Nour ELKHATEEB**	IEMbase: An online knowledge base and mini-expert tool for the diagnosis of inborn errors of metabolism **Prof Nenad BLAU**				
	Living with a chronic disease and maintaining quality of life **Dr Laura SMITH**	New condition-specific protein substitutes demonstrate good acceptability with improved compliance, stable tolerance and metabolic control in children and adults with HCU, MSUD and TYR **Ms Diane GREEN**	Functional genomics platform to evaluate the pathogenesis of variants of uncertain significance using CRISPR/Cas9 in HAP1 cells **Mr Gerard MUñOZ-PUJOL**				
	Role of continuous glucose monitoring in personalized glycemic management of hepatic glycogen storage disorders **Dr Ambika GUPTA, Dr Neerja GUPTA**	Use Of Sglt2 In Neutropenia In Gsd 1b, A UK Experience **Mr Antonio OCHOA**	Solving the unsolved: Implementing new -Omics approaches to enable diagnosis of virtually all patients with mitochondrial disease **Ms Sumudu AMARASEKERA**				
	Hepatocellular adenoma development in glycogen storage disease type Ia patients is associated with G6PC1 genotype and sex: a nationwide observational cohort study. **Mr Fabian PEEKS**	Nutritional adequacy of low protein diets in a sample of adult patients with Inborn Errors of Intermediary Protein Metabolism **Dr Nicola VITTURI**	Plasma C24:0 and C26:0 lysophosphatidylcholines are reliable biomarkers for the diagnosis of peroxisomal β-oxidation disorders. **Mr Blai MORALES-ROMERO**				
	The impact on the family and child pre and post-transplant **Mrs Julia**	Role of GLA variants in Parkinson's Disease **Dr Merve KOç YEKEDüZ**	DNL310 corrects lysosomal lipid accumulation and biomarkers of skeletal dysplasia in patients with neuronopathic MPS II: An exploratory analysis of fluid-based biomarkers **Dr Akhil BHALLA**				
13:15-13:30	***BREAK***			13:15	7:45	3:15	20:15

	SPONSORED SYMPOSIUM						
	SYMPOSIUM HALL A	**SYMPOSIUM HALL B**					
13:30-14:30	**Diagnose. Understand. Treat. A Glance at a Global NPC Cohort** Prof. Peter BAUER M.D Presented by Centogene	**Expediting diagnosis in rare metabolic diseases – examples from neuronal ceroid lipofuscinosis type 2 (CLN2) and mucopolysaccharidosis (MPS)** Dr Carolina FISCHINGER, Dr D. Scott Thomas DEMAREST, Dr Cathleen Louise RAGGIO Presented by BioMarin *Restricted to Health Care Professionals only - This sessions is not intended for US Healthcare Professionals			13:30	8:00	3:30	20:30
	CENTOGENE THE RARE DISEASE COMPANY	BIOMARIN					

	PARALLEL SESSIONS						
	CONCURRENT HALL 1	**CONCURRENT HALL 2**	**CONCURRENT HALL 3**				
	LSD Therapeutics	**Novel Therapeutics and Mechanisms**	**Disease Mechanisms**				
14:30-16:00	Sebelipase alfa enzyme replacement therapy in Wolman disease allows long-term survival with high health-related quality of life in a retrospective study with up to ten years follow-up **Dr Tanguy DEMARET**	Discovery of CDX-6512, a gastrointestinal-stable methionine-gamma-lyase as a potential orally-administered enzyme therapy for homocystinuria **Dr Kristen SKVORAK**	Dominant variants at the catalytic site of STT3A result in a CDG with a variable phenotype including intellectual disability, dysmorphic symptoms, short stature, skeletal dysplasia and cramps **Dr Matthew WILSON**	14:30	9:00	4:30	21:30
	Switching from agalsidase alfa to pegunigalsidase alfa to treat patients with Fabry disease: 1 year of treatment data from BRIDGE, a phase 3 open-label study **Prof Ales LINHART**	Discovery of a gastrointestinal-stable bacterial leucine decarboxylase as a potential orally-administered enzyme therapy for maple syrup urine disease **Dr Kristen SKVORAK**	Gm3 synthase deficiency is associated with increased mitochondrial respiration in the brain **Dr Eric GOETZMAN**				
	Effect of alglucosidase alfa dosage on survival and walking ability in patients with classic infantile Pompe disease: A study of the European Pompe Consortium. **Miss Imke DITTERS**	Allosteric, specific GALK1 inhibitors for development as substrate reduction therapy for classic galactosemia. **Dr Sabrina MACKINNON**	Abnormalities of Mitochondrial Dynamics and Bioenergetics in Neuronal Cells from CDKL5 Deficiency Disorder **Dr Nicole VAN BERGEN**				
	Nature history of CNS manifestation of infantile onset Pompe disease: a prospective cohort observational study using a quantitative scoring to reveal the progressive evolution of white matter changes **Dr Wen-Chin WENG**	Patient characteristics in the pivotal phase 3 PEACE trial of pegzilarginase human enzyme therapy for Arginase 1 Deficiency **Dr George DIAZ**	Impaired cofactor-dependent maturation of mitochondrial sulfite oxidase represents a new class of sulfite oxidase deficiency **Prof Guenter SCHWARZ**				
	Structure of Human Muscle Glycogen Synthase Guides Inhibitor design for Pompe Disease **Dr Thomas MCCORVIE**	Association between early treatment and neurodevelopmental outcome in 18 families with PDE-ALDH7A1 **Prof Clara VAN KARNEBEEK**	Loss of function of the phosphoinositide kinase PI4KA causes hypomyelinating leukodystrophy **Prof Aurora PUJOL**				
	Venglustat Pharmacokinetics (PK) and Biomarker Responses in Patients with Gaucher Disease Type 3 or GBA-associated Parkinson Disease **Dr M. Judith PETERSCHMITT**	Correction of mitochondrial dysfunction in ataxia-telangiectasia (A-T): an anaplerotic approach to therapy **Prof David COMAN**	Early prediction of disease severity in cytosolic Urea Cycle Disorders **Dr Matthias ZIELONKA**				
16:00-16:15	***REFRESH BREAK***			16:00	10:30	6:00	23:00
16:15-17:00	**Garrod Lecture** Simplifying the clinical classification of polymerase gamma (POLG) disease based on age of onset; studies using a cohort of 155 cases **Dr Laurence BINDOFF**			16:15	10:45	6:15	23:15

	SPONSORED SYMPOSIUM						
	SYMPOSIUM HALL A	**SYMPOSIUM HALL B**	**SYMPOSIUM HALL C**	**SYMPOSIUM HALL D**			
17:00-18:00	**Rare neurometabolic disorders: A focus on AADC deficiency** Dr. Wuh-Liang Hwu, Prof. Thomas Opladen, Prof. Agathe Roubertie Presented by PTC Therapeutics *restricted to Health Care Professionals only	**20 years of the Fabry Outcome Survey: understanding the importance of organ protection in patients with Fabry disease** Prof. Kathy Nicholls, Prof. Aleš Linhart, Prof. Christoph Kampmann, Ms. Mary Pavlou Presented by Takeda *restricted to Health Care Professionals only	**Translating theory into practice in Fabry disease: evidence from clinical trials to real-world** Dr Michel Tchan, Professor Daniel Bichet, Professor Roser Torra Presented by Amicus Therapeutics *restricted to Health Care Professionals only - This sessions is not intended for US Healthcare Professionals	**Pursuing More Effective Diagnosis and Treatment of Pompe Disease: Applying New Advances into Practice** Priya S. Kishnani, David Kronn, Kenneth I. Berger Presented by Sanofi Genzyme	17:00	11:30	7:00	0:00 (Monday)
	PTC THERAPEUTICS	Takeda	Amicus Therapeutics	SANOFI GENZYME				
	CLOSE OF DAY TWO						

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AEDT	TUESDAY 23 NOVEMBER 2021			AEDT	IST	CET (Monday)	CST (Monday)
07:00-07:30	**Online Q&A - New therapies** **Chaired by John Christodoulou and Roberto Guigliani**			07:00	1:30	21:00	14:00
07:30-08:00	**Online Q&A - Allied health** **Chaired by Anita Inwood and Rebecca Halligan**	**Online Q&A - Adult metabolic** **Chaired by Michel Tchan and Yusof Rahman**	**Online Q&A - Laboratory** **Chaired by Veronica Wiley and Adviye Tolun**	07:30	2:00	21:30	14:30
	INTEGRATED CARE						
08:00-08:05	**The patient experience of research** Mrs Sarah King			08:00	2:30	22:00	15:00
08:05-08:34	**Patient advocacy in guiding policy development for metabolic disorders** Nicole Millis *Rare Voices Australia* **** This presentation is supported by Menarini***			08:05	2:35	22:05	15:05
08:34-09:02	**Design and outcome of Rare Disease Registries in PKU** Dr Susan Berry *University of Minnesota*			08:34	3:04	22:34	15:34
09:02-09:28	**Consumer consultation to provide focused family centered-care** Dr Terry Derks *University of Groningen, Beatrix Children's Hospital*			09:02	3:32	23:02	16:02
09:28-10:00	**Treatment of the rare and Indigenous** Dr Callum Wilson *National Metabolic Service, Starship Childrden's Hospital*			09:28	3:58	23:28	16:28
10:00-10:20	***POSTER VIEWING BREAK***			10:00	4:30	0:00 (Tuesday)	17:00
10:20-10:42	**NETWORKING AND TRIVIA BREAK**			10:20	4:50	0:20	17:20
	LATE BREAKING NEWS						
10:42-11:45	Long-term functional correction of cystathionine beta-synthase deficiency in mice by adeno-associated viral gene therapy **Prof Warren Kruger** Mini-COMET: Safety and efficacy after ≥97 weeks' avalglucosidase alfa in infantile-onset Pompe disease (IOPD) patients previously treated with alglucosidase alfa who had demonstrated clinical decline **Dr David Kronn** Phenylbutyrate Reduces Metabolite Accumulation and Improves Bioenergetics in Combined D,L-2-Hydroxyglutaric Aciduria **Dr Yu Leng Phua** Reduction in Plasma Phenylalanine Levels in Patients with Phenylketonuria with Live Bacterial Therapeutic SYNB1618 **Prof Jerry Vockley**			10:42	5:12	0:42	17:42
	PARALLEL SESSIONS						
	LSD Therapeutics	**Vitamins and Cofactors**	**General Metabolism**				
11:45-13:15	Two-year outcomes of the first trial of olipudase alfa enzyme replacement therapy in children with chronic acid sphingomyelinase deficiency show continued improvements in clinical parameters **Prof Roberto GIUGLIANI**	Complications, Hospital Admissions, And Healthcare Costs During Pharmacologic Treatment For Wilson Disease **Dr Eliza KRUGER**	The Impact of SARs-CoV2 infection on paediatric patients with Inherited Metabolic Diseases in the UK: The first 14 months. **Dr Sarah HULLEY**	11:45	6:15	1:45	18:45
	Olipudase alfa enzyme replacement therapy improves liver and lipid parameters in adults and children with chronic acid sphingomyelinase deficiency: 1-year results of the ASCEND and ASCEND-Peds trials **Prof Maurizio SCARPA**	Identification of novel pathogenic MOCS1 variants predicts molybdenum cofactor deficiency type A disease incidence **Prof Guenter SCHWARZ**	Treatable Inherited Metabolic Disorders causing Intellectual Disability: 2021 Review and App. **Prof Clara VAN KARNEBEEK**				
	Ex vivo autologous haematopoietic stem cell gene therapy in mucopolysaccharidosis type IIIA **Dr Simon JONES**	Increased Survival in MoCD Type A Patients Treated with Fosdenopterin when Compared to a Natural History Cohort **Dr Liza SQUIRES**	CAMLG-CDG: a novel glycosylation and membrane trafficking defect **Dr Matthew WILSON**				
	Interim Results of Transpher A, a Multicentre, Single-Dose, Phase 1/2 Clinical Trial of ABO-102 Gene Therapy for Sanfilippo Syndrome Type A (Mucopolysaccharidosis IIIA) **Dr Nicholas SMITH**	5,10-methenyltetrahydrofolate synthetase (MTHFS) deficiency: an extreme rare defect of folate metabolism. **Prof Henk BLOM**	Decipher N-linked glycosylation changes in the plasma from CDG patients **Dr Miao HE**				
	RGX-121 gene therapy for the treatment of severe mucopolysaccharidosis type II (MPS II): Interim analysis of data from the first in human study **Prof Roberto GIUGLIANI**	Eladocagene Exuparvovec Gene Therapy Improves Motor Development in Patients With Aromatic l-Amino Acid Decarboxylase Deficiency **Dr Paul W-L HWU**	De novo DHDDS variants cause a neurodevelopmental and neurodegenerative disorder in the spectrum of myoclonus syndromes intersecting glycosylation and organelle disorders **Dr Serena SERENA GALOSI**				
	Clinical data trends from Phase 1 and Phase 2 studies for AVR-RD-01, an investigational lentiviral gene therapy for Fabry disease **Prof Mark THOMAS**	Eladocagene Exuparvovec Improves Body Weight and Reduces Respiratory Infections in Patients With Aromatic l-Amino Acid Decarboxylase Deficiency **Dr Paul W-L HWU**	Identification of the first patient with Guanidinoacetate Methyltransferase Deficiency by newborn screening. **Prof Marzia PASQUALI**				
13:15-13:30	***BREAK***			13:15	7:45	3:15	20:15
	SPONSORED SYMPOSIUM						
	SYMPOSIUM HALL A	**SYMPOSIUM HALL B**	**SYMPOSIUM HALL C**				
13:30-14:30	**Advancing the patient experience during ex vivo lentiviral gene therapy for lysosomal disorders** Dr. Robert Wynn, Dr. Chris Mason, Dr. Mark Thomas, Dr. Kathleen Nicholls, Dr. Ben Carnley Presented by Avrobio *restricted to Health Care Professionals only	**Targeting Toxic Galactitol for the Treatment of Classic Galactosemia** Shoshana Shendelman, Laura Saltonstall, Riccardo Perfetti Presented by Applied Therapeutics	**A tale of two Phe states: PKU neurocognitive burden and the Palynziq experience** Mark Walterfang, Cary Harding Symposium sponsored and funded by BioMarin *restricted to Health Care Professionals only - This sessions is not intended for US Healthcare Professionals	13:30	8:00	3:30	20:30
	AVROBIO	APPLIED THERAPEUTICS	BIOMARIN				
	SPECIAL AWARD PRESENTATIONS						
14:30-15:30	**Long-term, sustained efficacy and safety from a phase ½ clinical trial of an AAV8-mediated liver-directed gene therapy in adults with glycogen storage disease type Ia.** Dr Terry G. J. Derks **Nanoparticles with β-cyclodextrin as a technological advantage to Niemann-Pick C treatment: preliminary results of biodistribution and cholesterol reduction** Dr Carmen Regla Vargas **The Australian Genomic Health Alliance (AGHA) Mitochondrial Flagship: Delivering Mitochondrial Diagnoses at a National Level.** Prof John Christodoulou **A systematic study of a panel of mitochondrial functional testing in fibroblasts shows strong clinical utility** Prof Johan Van Hove			14:30	9:00	4:30	21:30
15:30-16:20	**Danks Lecture** **Transforming Rare Disorders** Introduction by: Jim McGill Award Recipient: John Christodoulou			15:30	10:00	5:30	22:30
16:20-16:30	***REFRESH BREAK***			16:20	10:50	6:20	23:20
	SPONSORED SYMPOSIUM						
	SYMPOSIUM HALL A	**SYMPOSIUM HALL B**	**SYMPOSIUM HALL C**	**SYMPOSIUM HALL D**			
16:30-17:30	**Introducing HST5040: A Systemic Small Molecule Approach for MMA and PA** Carlo Dionisi-Vici, Kimberly A Chapman, Gerald F. Cox Presented by Hemoshear *restricted to Health Care Professionals only	**The story of the United Kingdom National Alkaptonuria (AKU) Centre and its global influence on research, changing the face of AKU management.** Shirley Judd, Prof Lakshminarayan Ranganath Presented by VitaFlo	**Burden of treatment and Quality of Life in Patients with Urea Cycle Disorders** Karolina Stepien, Francis Fatoye, Gillian Yeowell, Danielle Burns Presented by Immedica *restricted to Health Care Professionals only	**Glucosylceramide Synthase Inhibition and its Critical Role in the Glycosphinigolipid Hub of Lysomal Storage Disorders** Associate Professor Carolyn Ellaway, Professor Tim Cox, Professor Hans Aerts Presented by Sanofi Genzyme	16:30	11:00	6:30	23:30
	HEMOSHEAR THERAPEUTICS	Vitaflo	Immedica	SANOFI GENZYME				
	CLOSE OF DAY THREE						
19:00-00:00	***Conference Dinner - In person registrations only***						

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AEDT	WEDNESDAY 24 NOVEMBER 2021	AEDT	IST	CET (Tuesday)	CST (Tuesday)
07:30-08:00	**Online Q&A - Late Breaking news** **Chaired by Kimitoshi Nakamura and Jim Bonham**	07:30	2:00	21:30	14:30
08:00-08:30	**Online Q&A - Special award presentations** **Chaired by Ute Spiekerkoetter and Shawn McCandless**	08:00	2:30	22:00	15:00
08:30-09:00	**Online Q&A - Integrated care** **Jose Abdenur and Anita Inwood**	08:30	3:00	22:30	15:30
09:00-09:30	**Society Updates** SIMD - Gerry Berry SLEIMPN - Ceila Perez de Ferrán SSIEM - Gajja Salomons and Ute Spiekerkoetter ASIEM - Veronica Wiley **Presentation for ICIEM 2025** JSIMD - Kimitoshi Nakamura **Congress Close** Kaustuv Bhattacharya	09:00	3:30	23:00	16:00

14TH INTERNATIONAL CONGRESS OF INBORN ERRORS OF METABOLISM

ICIEM 2021

21-23 NOVEMBER 2021, SYDNEY, AUSTRALIA

VALUED MAJOR PARTNERS

OPAL PARTNERS

PTC THERAPEUTICS

measured by moments

SANOFI GENZYME

PLATINUM PARTNERS

Amicus Therapeutics

APPLIED THERAPEUTICS

AVROBIO

HEMOSHEAR THERAPEUTICS

Takeda

Vitaflo

Enhancing Lives Together

GOLD PARTNERS

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SILVER PARTNERS

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PREKULAB

TRAVERE THERAPEUTICS

BRONZE PARTNERS

APR APPLIED PHARMA RESEARCH

BRUKER

metaX INSTITUT FÜR DIÄTETIK

NUTRICIA METABOLICS LIFE-TRANSFORMING NUTRITION

origin a bridgebio company

Orpharma A ray of hope

REGENXBIO

PROGRAM PARTNERS

BIOMARIN

CENTOGENE THE RARE DISEASE COMPANY

Immedica pharma

MENARINI

PerkinElmer For the Better