FOURTH INTERNATIONAL HOMOCYSTINURIAS PATIENT EXPERT VIRTUAL MEETING

30 NOVEMBER - 1 DECEMBER 2021

PROGRAM

HCU Network Australia

EHOD
European Network and Registry for Homocystinurias and Methylation Defects

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WELCOME

The Organising Committee welcomes you to the first ever virtual International Homocystinurias Patient-Expert (iHOPE) Meeting. We do hope this virtual meeting will make it accessible to people who have not been able to attend previous in-person events. Meeting times have been set to allow people to join from around the world. We look forward to bringing together renowned researchers, clinicians in genetic and metabolic disorders, dieticians, patients and caregivers. The hope is that the meeting will help to improve the treatment of homocystinurias as we hear about potential emerging treatments and practical suggestions to help patients and caregivers manage and live with homocystinuria.

We take this opportunity to thank each delegate and speaker for making the time to join us for iHOPE 2021 and we hope you are able to connect with, and learn from, others in the field of homocystinuria. We would also like to acknowledge and thank our sponsors; without them today would not be possible.

The Organising Committee

Prof. Henk Blom

Prof. Martina Huemer

Prof. Viktor Kožich

Dr. Andrew Morris

Tara Morrison

HCU Network Australia

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A MESSAGE FROM YOUR CO-HOST

People impacted by a rare disease, like homocystinuria, are empowered by knowledge. Patients and caregivers have a strong desire to learn how to manage and live with HCU successfully and they draw hope from scientific advancement and emerging treatments. Parents of newly diagnosed children want to know more about the disorders and what are the best practices for their child's care. Older patients and experienced caregivers require information on managing HCU day-to-day and adult patients feel strongly about supporting research and gaining access to clinical trials.

As a parent of 2 children with Classical Homocystinuria our personal journey would have been quite different if awareness were greater, and my children were detected by screening. When our family faced the diagnosis of Homocystinuria we were left with many unanswered questions about this disorder. My hope is to unite the HCU community and try and change this experience for others.

Tara Morrison
HCU Network Australia

OVERVIEW

The iHOPE Meeting promotes engagement and networking in the field of homocystinuria and provides a platform for the exchange of patient experiences and needs. This exchange of information is paramount when promoting improved health outcomes for individuals affected by these disorders. This platform brings patient needs to front of mind and fosters collaboration between all stakeholders.

The aims:

- Provide stakeholders with updates on research and new and emerging treatments.
- Strengthen networking between all stakeholders including enhanced peer- support for patients and caregivers.
- Improve knowledge on these disorders by exchanging experiences and best practices on presentation, diagnosis and treatment.
- Provide education and resources for patients, caregivers and healthcare professionals.
- Enhance stakeholder focus, knowledge and enthusiasm in these disorders.
- Share experience of patients and caregivers living with these disorders.

HCU Network Australia

HCU NETWORK AUSTRALIA ARE A DRIVING FORCE IN THE JOURNEY TO A CURE, IMPROVING QUALITY OF LIFE ALONG THE WAY.

HCU Network Australia is focused on improving the health outcomes for people impacted by homocystinuria and helping them to live with HCU successfully. We support patients and their families, raise awareness, and support and encourage research to improve the diagnostic path, expand treatment options and ultimately find a cure.

HCUNetworkAustralia.org.au

E-HOD

European Network and Registry for
Homocystinurias and Methylation Defects

E-HOD is an international collaboration of clinicians and scientists; though it started as a European project, it now has a worldwide membership.

E-HOD aims to improve the diagnosis and management of homocystinurias and methylation defects.

Its core activities are developing guidelines, maintaining a register of patients and sharing knowledge through international meetings.

e-hod.org

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PROGRAM

SESSION 1	PRESENTER	AEDT	CET	EST
Opening	Tara Morrison	Tues 30 Nov 5.00pm	Tues 30 Nov 7.00am	Tues 30 Nov 1.00am
Classical Homocystinuria				
Tips to stay on diet for classical homocystinuria	Brooke Pinsent	Tues 30 Nov 5.05pm	Tues 30 Nov 7.05am	Tues 30 Nov 1.05am
Nutricia Metabolics continued support of the HCU community	Amira Ramadan Renee Wells	Tues 30 Nov 5.35pm	Tues 30 Nov 7.35am	Tues 30 Nov 1.35am
Observations from newborn screening for homocysteine in NSW	Kaustuv Bhattacharya	Tues 30 Nov 6.00pm	Tues 30 Nov 8.00am	Tues 30 Nov 2.00am
Homocystinuria and bones	Michel Tchan	Tues 30 Nov 6.30pm	Tues 30 Nov 8.30am	Tues 30 Nov 2.30am
Break & Networking		Tues 30 Nov 7.00pm	Tues 30 Nov 9.00am	Tues 30 Nov 3.00am
Research Updates				
The EHOD project: updates on guidelines and research	Martina Huemer	Tues 30 Nov 7.30pm	Tues 30 Nov 9.30am	Tues 30 Nov 3.30am
MTHFR deficiency: influence of early initiation of therapy on long-term outcome	Mathilde Yverneau	Tues 30 Nov 8.00pm	Tues 30 Nov 10.00am	Tues 30 Nov 4.00am
Homocystinuria patient and caregiver survey: experiences of diagnosis and patient satisfaction	Andrew Morris	Tues 30 Nov 8.30pm	Tues 30 Nov 10.30am	Tues 30 Nov 4.30am
B12 dose escalation in cblC deficiency: experience from a US natural history study	Irini Manoli	Tues 30 Nov 9.00pm	Tues 30 Nov 11.00am	Tues 30 Nov 5.00am
Clinical presentation and treatment practice in patients with CBS deficiency: update on E-HOD registry analysis	Viktor Kozich	Tues 30 Nov 9.30pm	Tues 30 Nov 11.30am	Tues 30 Nov 5.30am
Close		Tues 30 Nov 10.00pm	Tues 30 Nov 12.00pm	Tues 30 Nov 6.00am

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SESSION 2	PRESENTER	AEDT	CET	EST
Panel Discussions				
Panel Q & A **CblC and Severe MTHFR Deficiency** Moderator: Kaustuv Bhattacharya	Matthias Baumgartner Martina Huemer Irini Manoli David Rosenblatt	Wed 1 Dec 7.00am	Tues 30 Nov 9.00pm	Tues 30 Nov 3.00pm
Panel Q & A **Classical homocystinuria** Moderator: Kaustuv Bhattacharya	Andrew Morris Viktor Kozich Harvey Levy	Wed 1 Dec 7.45am	Tues 30 Nov 9.45pm	Tues 30 Nov 3.45pm
Research announcement	Karolina Stepien	Wed 1 Dec 8.30am	Tues 30 Nov 10.30pm	Tues 30 Nov 4.30pm
Break & Networking		Wed 1 Dec 8.40am	Tues 30 Nov 10.40pm	Tues 30 Nov 4.40pm
Novel Treatments Progress				
Clinical Trial Recruitment: COVID , challenges & where we are today	Mark Bechter	Wed 1 Dec 9.00am	Tues 30 Nov 11.00pm	Tues 30 Nov 5.00pm
Discovery of CDX-6512, a GI-stable methionine-gamma-lyase as a potential orally-administered enzyme therapy for homocystinuria	Kristen Skvorak	Wed 1 Dec 9.30am	Tues 30 Nov 11.30pm	Tues 30 Nov 5.30pm
Natural history studies and why they are needed in rare disease drug development	Jalé Güner Sagar A. Vaidya	Wed 1 Dec 10.00am	Tues 30 Nov 12.00am	Tues 30 Nov 6.00pm
Strategies to correct CBS deficiency: studies in mice	Warren Kruger	Wed 1 Dec 10.30am	Tues 30 Nov 12.30am	Tues 30 Nov 6.30pm
SYNB1353: An Investigational Methionine-consuming Synthetic Biotic Medicine for the Treatment of Homocystinuria	Mylène Perreault	Wed 1 Dec 11.00am	Tues 30 Nov 1.00am	Tues 30 Nov 7.00pm
Break & Networking		Wed 1 Dec 11.10am	Tues 30 Nov 1.10am	Tues 30 Nov 7.10pm
HCU Network America & HCU Network Australia Research Grant Awardees				
Evaluation of benefits of thiol-based reductants in classical homocystinuria	Tomas Majtan	Wed 1 Dec 11.30am	Tues 30 Nov 1.30am	Tues 30 Nov 7.30pm
Altered folate metabolism in HCU: opportunities for improved therapy	Ken Maclean	Wed 1 Dec 12.00pm	Tues 30 Nov 2.00am	Tues 30 Nov 8.00pm
Patient Organisation and Collaboration Updates				
RARE-X: Homocystinuria patient's data powering progress	Vanessa Vogel-Farley	Wed 1 Dec 12.30pm	Tues 30 Nov 2.30am	Tues 30 Nov 8.30pm
HCU Network America: Advocating for improved NBS	Margie McGlynn	Wed 1 Dec 1.00pm	Tues 30 Nov 3.00am	Tues 30 Nov 9.00pm
Acknoweledgements & Close	Tara Morrison	Wed 1 Dec 1.30pm	Tues 30 Nov 3.30am	Tues 30 Nov 9.30pm

SPEAKERS

PROF. MATTHIAS BAUMGARTNER

Switzerland

Matthias Baumgartner, MD is professor of metabolic diseases at the University of Zurich, head of the Division of Metabolism, Medical Director of the Swiss Newborn Screening Program and

Director of Research & Education at the University Children's Hospital Zurich, Switzerland. Since 2012 he leads the clinical research priority program "Rare Disease Initiative Zurich – radiz" at the University of Zurich. He is an internationally known metabolic paediatrician and scientist with a main research interest in disorders of intracellular cobalamin metabolism

including the homocystinurias and methylmalonic acidurias and an editor of the Journal of Inherited Metabolic Disease. He is a cofounder and steering committee member of the European networks and registries for Intoxication type Metabolic Diseases (E-IMD, www.e-imd.org) and for Homocystinurias and remethylation disorders (E-HOD, www.e-hod.org).

MARK BECHTER, B.M.

United Kingdom

Dr. Mark Bechter joined the pharma industry in 2004 after a clinical career in Anesthesiology and Emergency Medicine. Through a series of roles in Medical Affairs, at both country, regional and global levels, he has extensive experience in both pharmaceuticals and medical devices, with a focus on rare diseases. Prior to Aeglea, Mark served as a Global Therapeutic lead at Baxter and VP of medical affairs at Amicus Therapeutics, where he established medical affairs teams that successfully launched products internationally. He has also served in various medical roles at Novartis, Chiesi Group, Synageva BioPharma (acquired by Alexion) and was CMO of the Precision Medicine Catapult, UK Government funded project. Mark received his medical degree (Bachelor of Medicine) from the University of Southampton in the UK and continues to be registered with the General Medical Council of the UK as a Pharmaceutical Physician.

DR. KAUSTUV BHATTACHARYA

Australia

Senior Staff Specialist, Sydney Children's Hospitals Network, Sydney, Australia Conjoint Senior Lecturer, Discipline of Child and Adolescent Health and Genomics, Sydney University

Kaustuv Bhattacharya is a metabolic paediatrician trained in the UK. His metabolic training was in London at Great Ormond Street Hospital for Children. Having completed a fellowship in metabolic medicine and newborn screening in Sydney, Australia, he completed clinical research in glycogen

storage diseases with the late Dr Philip Lee and obtained a research MD in clinical nutrition for this work from the University of London. His work also led to a new carbohydrate therapy for glycogen storage disease type I being developed and marketed.

Dr Bhattacharya is a senior staff specialist at Sydney Children's Hospitals Network and conjoint senior lecturer at Sydney University. He is continuing clinical research projects in several inborn errors of metabolism, allied to the New South Wales newborn screening programme and is the principal investigator for several clinical trials. He has published 50 papers in peer-reviewed journals ranging from dietary therapy in inborn errors of metabolism to haematopoietic stem cell transplants in lysosomal storage disorders. He will be the convenor of International Congress of Inborn Errors of Metabolism (ICIEM) 2021 in Sydney.

JALÉ GÜNER

United States

Ms. Jalé Güner is the Executive Director of Clinical Operations and is the pegtibatinase (TVT-058) Development Program Lead at Travere Therapeutics. She has been with Travere since 2019 and is responsible for overseeing the strategy and execution of pegtibatinase through its development life cycle. Jalé has specialized her clinical development and operations career in oncology and rare disease, working at small and large biopharma companies in California. Jalé received her B.S degrees in Biochemistry and Molecular Cell Biology at UCSD and currently resides with her family and dog in San Diego, CA.

PROF. MARTINA HUEMER, MD

Switzerland & Austria

Martina Huemer, MD and Psychologist, works as Consultant at the Children's Hospital in Zürich and heads the specialist outpatient clinic for inborn errors of metabolism at University Children's Hospital in Basel, Switzerland, as well as in Vorarlberg, Austria.

She received her degree in Psychology in 1987 and her MD in 1994. After receiving her post-doctoral lecturing qualification for Paediatrics in 2009, she was appointed guest professor at the University of Vienna in 2015 and titular professor at the University of Zürich in 2016. Her research focuses on the Homocystinurias and methylation disorders and on psychological aspects of patient care in inborn errors of metabolism. She is author to more than 80 peer reviewed publications including treatment guidelines for a number of inborn errors of metabolism, including cystathionine synthase deficiency and the remethylation defects. Martina Huemer is chair of the European network and registry for homocystinurias and methylation defects (E-HOD) project. Beyond her research activities around the homocystinurias and within E-HOD she leads several projects on health-related quality of life, patient education and patient reported outcomes in children with metabolic diseases.

PROF. VIKTOR KOŽICH

Czech Republic

Professor Kožich graduated from the School of General Medicine, Charles University in Prague in 1985. Since his graduation he has been working in the Institute of Inherited Metabolic Diseases and he specialized in clinical biochemistry and medical genetics, in 2012 he became the full Professor of Medical Genetics.

His main interests are genetic, biochemical, clinical, epidemiological and ethical aspects of inherited metabolic disorders with a special interest in disorders of homocysteine metabolism and namely in cystathionine beta-synthase deficiency—a disease in which he became interested in 1991-1992 during his fellowship in the laboratory of Prof.Jan P.Kraus (University of Colorado School of Medicine in Denver, USA). Prof.Kožich is also involved in organization of neonatal screening and serves as a Chairman of the national Coordination Center on Neonatal Screening in the Czech Republic.

Professor Kožich has been a tutor of graduate and postgraduate students, he is an author of over 100 publications in peer reviewed international journals, several chapters in books, and of articles and chapters in Czech medical literature; he has been an invited speaker at various international and national conferences. He is a member of councils of several international learned societies (SSIEM, ERNDIM, and ESHG) and he is active in peer review system at both the national and international levels.

DR. WARREN KRUGER

United States

Dr. Warren Kruger received his Ph.D. in Biochemistry and Genetics from the University of California at San Francisco, working in the lab of Dr. Ira Herskowitz studying transcriptional control of the yeast HO gene. He went on to do post-doctoral work at Stanford in human genetics in the lab of Dr. David Cox, where he developed yeast functional assays to study the functional effects of human cystathionine beta-synthase mutations. Since 1995 he has been a researcher at the Fox Chase Cancer Center in Philadelphia, where he is currently a full professor in the Molecular Therapeutics Program. Dr. Kruger's work focuses on the role of dysregulated methionine metabolism in human disease. He has had continuous NIH funding in this area since 1997 and is a recognized leader in the 1-carbon metabolism field, having been an invited speaker and session chair several times at both the International Homocysteine Meeting and the FASEB summer conference on Folate, Methionine, and B12 Metabolism. His lab has made numerous contributions to the study of CBS deficiency including the cloning of gene, creation of functional models in yeast and mice, and key discoveries on the regulations of the enzyme. He enjoys scientific collaboration and the mice created in his lab are used to study homocysteine related disease throughout the world.

DR. HARVEY LEVY

United States

I am a Senior Physician in Medicine/Genetics at Boston Children's Hospital in the Division of Genetics and Genomics and Professor of Pediatrics at Harvard Medical School. I have published over 400 articles on metabolic disorders and on the newborn screening of these disorders. I have been the fortunate recipient of a number of awards for my work.

The first metabolic disorder I encountered during my post-doctoral fellowship was the classic homocystinuria (HCU). This was 1967 and the first case in the United States of HCU identified by newborn screening. This infant had an older sister with the complications of HCU who was born before newborn screening for HCU had been added to the Massachusetts program. Shortly thereafter, I encountered an infant with markedly increased homocysteine but whose other biochemical characteristics were very different from HCU. This led to a collaboration with Dr. Harvey Mudd of the NIH and the discovery of the B12 metabolic disorder now known as cobalaminC (cblC) and the realization that elevated homocysteine (i.e. homocystinuria) was not one disorder but the key feature of many disorders. Thus, in my over 50 years of study in the metabolic and transport disorders, including the diagnosis, treatment, and investigation of PKU, galactosemia, Hartnup disorder, and others as well as newborn screening, the homocystinurias, including HCU, have been at the forefront.

DR. KEN MACLEAN

United States

Dr Ken Maclean is a Professor of Pediatrics at the University of Colorado School of Medicine and holds the Ehst-Hummel-Kaufman Family Endowed chair in Inherited Metabolic Disease. Dr Maclean gained his undergraduate degree and PhD at the University of Greenwich in London, UK. This was followed by an EMBO post-doctoral fellowship at the Hungarian Academy of Sciences in Szeged, Hungary and a further post-doctoral fellowship at the Royal London-St Bartholomew's Hospital trust in the UK.

In 1998, Dr. Maclean came to Colorado to work on the transcriptional and post-translational regulation of cystathionine beta-synthase (CBS) in the laboratory of Dr. Jan Kraus.

Since establishing his own laboratory in 2002, Dr. Maclean's work has centered on using transgenic and knockout/in mouse models to investigate the pathobiology of CBS deficient homocystinuria (HCU), homocysteine remethylation defects, Down Syndrome, propionic acidemia, and non-ketotic hyperglycinemia. Additional collaborative projects in the Maclean lab have focused on investigating the mechanisms involved in hepatic fibrosis, atherosclerosis, diabetes, nonalcoholic steatohepatitis, and alcoholic steatohepatitis.

The Maclean laboratory uses transcriptomic, metabolomic, proteomic, and behavioral approaches combined with a range of biochemical and molecular strategies to understand the biological mechanisms that result in the clinical sequelae found in these diseases with a view towards developing improved treatments. Dr Maclean recently co-directed an FDA-funded multi-site clinical trial of the use of taurine as a novel treatment for HCU. This treatment resulted in significant improvement in HCU patient endothelial function and constitutes the first advance in treatment for this disease in over 40 years.

DR. TOMAS MAJTAN

United States

Tomas Majtan received his PharmD in 2003 from the Faculty of Pharmacy, Comenius University in Bratislava studying novel antimicrobial compounds and disinfectants using Salmonella pathogens. He then pursued postgraduate training in microbiology at Slovak Medical University and in 2006 he received a PhD in molecular biology working at the Institute of Molecular Biology of the Slovak Academy of Sciences. During this period, he studied epidemiology and genetics of Salmonella virulence factors and antibiotic resistance markers and deciphered gene expression of bacteriophage during infection of an important industrial strain producing amino acid lysine. In 2007, he started postdoctoral training on enzymology and biochemistry of cystathionine beta-synthase (CBS) in Professor Jan Kraus group at the University of Colorado School of Medicine, Aurora, Colorado. In 2013, he was promoted to Assistant Research Professor at the Department of Pediatrics, Section of Genetics and Metabolism, University of Colorado School of Medicine and continues working on understanding molecular mechanisms underlying inborn errors of metabolism and developing new treatments with focus on homocystinuria.

His contributions to the field include uncovering the mechanism of how missense pathogenic mutations impair CBS function, understanding how CBS cofactors work and affect folding and stability of the WT and mutant enzyme, solving several crystal structures of full-length human CBS to gain structural insight into homocystinuria or clarifying the role of CBS as hydrogen sulfide producing enzyme in health and disease. He utilized his intimate knowledge about CBS and has been working with Professor Jan Kraus and Orphan Technologies on enzyme therapy for homocystinuria since 2010. His enzyme therapy for homocystinuria named pegtibatinase (developed by Travere Therapeutics) is currently advancing in a Phase 1/2 clinical trial evaluation.

Dr. Majtan has been a mentor and supervisor of several graduate and postgraduate students or research associates. He is an author of over 50 peer-reviewed papers published in international journals as well as several patents, book chapters and monographs. He regularly presents his research on various conferences and meetings. In addition, he serves in editorial board of MDPI's Biology journal as well as a reviewer for multiple scientific journals and several funding agencies.

DR. IRINI MANOLI, M.D.,P.H.D.

United States

Dr Manoli is a physician scientist and Clinician Associate Investigator of the Medical Genomics and Metabolic Genetics Branch in the National Human Genome Research Institute (NHGRI), National Institutes of Health (NIH), Bethesda, MD, USA. She earned her medical degree, a Masters in Pediatric Endocrinology and PhD in Medical Sciences from the University of Athens in Greece and completed her pediatrics residency training at John Radcliffe Hospital in Oxford, England and University Children's Hospital in Athens. She subsequently pursued postdoctoral and fellowship training in clinical and biochemical genetics through the combined NHGRI and Children's National Medical Center Training program and was ABMGG board certified in 2009. She continued as a Staff Clinician in the Medical Genomics and Metabolic Genetics Branch and became a Clinician Associate Investigator in 2016.

She combines work on animal models and clinical studies with the aim to develop new therapies for methylmalonic acidemias (MMA) and defects of intracellular cobalamin metabolism. Her clinical research led to the re-evaluation of long-standing dietary guidelines for MMA and cobalamin C deficiency. Dr Manoli is the recipient of a number of grants and awards, including an NIH Bench to Bedside grant to study isotope methods for metabolic phenotyping of organic acidemias, and served as a co-PI for a U01 grant to study the genetics of Moebius syndrome. She serves as faculty of the Metropolitan Washington D.C. Medical Genetics Fellowship Training Program, is a scientist member of the NIH IRB, and a co-inventor of seven patents related to biomarkers and therapeutics for organic acidemias.

MARGARET MCGLYNN

United States

Margie McGlynn is President of the Board of HCU Network America, a patient advocacy organization she co-founded to provide support for patients and families affected by homocystinuria. She is also President of the Hempling Foundation for Homocystinuria Research, a fund she established to support research on new therapies for HCU in honor of her late sisters, who passed from homocystinuria in the early 1970s. Margie is committed to finding a cure for homocystinuria so that someday no no one will suffer like her sisters did, and no families will need to deal with the impact of this devastating illness on their family members or the fear of passing the disease along to additional offspring.

Margie received a BS in Pharmacy, MBA in Marketing and honorary doctorate in sciences from the University at Buffalo. Margie spent 26 years at Merck where she served in leadership roles in marketing, new product development, and managed care, last serving as President, Global Vaccines and Anti-Infectives. After retiring from Merck, Margie served for 4 years as President and Chief Executive Officer of the International AIDS Vaccine Initiative (IAVI), a non-profit product development partnership focused on HIV vaccine development. Margie also serves on the boards of Vertex Pharmaceuticals, Amicus Therapeutics, and Novavax.

DR. ANDREW MORRIS

United Kingdom

Dr. Andrew Morris is a Consultant and Senior Lecturer in Paediatric Metabolic Medicine. He works on the Willink Metabolic Unit, which is part of the Genomic Medicine in the Central Manchester University Hospitals.

Dr. Morris has extensive experience in inborn errors or metabolism in children and some experience with adults. He has considerable experience of managing patients with classical homocystinuria and remethylation disorders.

Dr. Morris is on the Steering Group for E-HOD, the European Network and Registry for Homocysinurias and Methylation Defects; he is Leader of the Classical Homocystinuria Guidelines Group and the Work Group on Evaluation of the Project.

Dr. Morris is currently a scientific advisor for the HCU Network Australia and most recently is the first named author on the recently published Guidelines for the diagnosis and management of cystathionine beta-synthase deficiency from the Journal of Metabolic Disease.

DR. MYLÈNE PERREAULT

United States

Dr. Mylène Perreault is Head of Biology at Synlogic and is responsible for the pre-clinical development strategy and operations for metabolic and immunology-related disorders. Mylene is a PhD scientist with over 19 years of experience in the pharmaceutical industry leading platform and drug development programs, in addition to lead multiple early programs to sustain the portfolio. Prior to joining Synlogic, Mylene served as an Associated Research Fellow and Laboratory Head in the Internal Medicine department at Pfizer, where she led pre-clinical projects and pharmacology operations for several indications, including type 2 diabetes, obesity, non-alcoholic liver disease and cancer cachexia. Mylene started her career in pre-clinical development at Wyeth Pharmaceuticals where she led the pharmacology efforts for metabolic disease programs. She earned a Ph.D. in physiology-endocrinology from Laval University in Québec city, Canada, and a B.S. in Biology also from Laval University. Mylene has co-authored numerous manuscripts and abstracts focused on metabolic-related disorders.

BROOKE PINSENT

Australia

Brooke Pinsent is a clinical dietitian working at The Royal Children's Hospital Melbourne with five years of experience in paediatrics. She has specialised in the nutrition management of inborn errors of metabolism for the past year. She is a current member of HGSA and ASIEM.

AMIRA RAMADAN

Australia

For the last 10 years, Amira has been a leader in the Medical Nutrition industry for inherited metabolic disorders (IMD), she is a pharmacist with more than 19 years' of experience in the healthcare sector managing various other therapeutic areas (neurology, faltering growth, oncology, immunology and cardiology). She has a played a pivotal role in progressing the management of IMD's through implementing the newborn screening program for PKU and other IMD in several (developing) countries worldwide and by providing customized product portfolio, educational tools and services. Amira believes in the power of Nutricia specialised medical nutrition to provide "Best Care" when working together with the community, healthcare professionals and authorities. Amira joined Nutricia Metabolics team in Australia in 2019 to continue her journey in supporting the metabolic community.

DAVID ROSENBLATT

Canada

David Rosenblatt is a Canadian medical geneticist, pediatrician, and professor in the departments of Human Genetics, Medicine, Pediatrics, and Biology at McGill University in Montreal, Quebec, where he was the chairman of the Department of Human Genetics from 2001 to 2013. He is known for his contributions to the field of inborn errors of folate and vitamin B12 metabolism.

DR. KRISTEN SKVORAK

United States

Dr. Kristen Skvorak is an in vivo pharmacologist who strongly believes the patient voice should guide drug development, from ideation through approval. Her research focuses on the discovery and pre-clinical validation of novel therapies of Inborn Errors of Metabolism (IEM), a passion that started in 2003 with her doctoral thesis on Maple Syrup Urine Disease (MSUD) at the University of Pittsburgh School of Medicine. Dr. Skvorak has published on the creation and characterization of mouse models of MSUD, novel cell transplant therapies for IEM, which progressed to clinical development in two countries, and generation of a minipig model of Phenylketonuria (PKU). She was a multiple-year awardee of the National PKU Alliance Research Fellowship for her postdoctoral work at the Children's Hospital of Pittsburgh, and is currently a Translational Scientist, and the first Patient Ambassador, at Codexis, Inc., an enzyme engineering biotechnology company in the Bay Area of California, USA.

DR. MICHEL TCHAN (BMEDSC, MBBS, PHD, FRACP)

Australia

Dr. Tchan is a clinical and metabolic geneticist looking after adults with genetic disorders and inborn errors of metabolism. He is currently the Head of Department, Genetic Medicine at Westmead Hospital in Sydney, and a senior lecturer at the University of Sydney.

He is currently responsible for the NSW Adult Genetic Metabolic Disorders Clinic. His research interests include clinical aspects of the lysosomal storage disorders, the neurological consequences of phenylketonuria in adults, and clinical trials of novel therapies for genetic disorders.

SAGAR A. VAIDYA

United States

Dr. Sagar Vaidya is a physician-scientist and rare disease drug developer who has served as the Vice President of Clinical Development at Travere Therapeutics since 2019. Dr. Vaidya is responsible for overseeing clinical development for the company's metabolic pipeline. Previously, Dr. Vaidya served in roles in clinical development at Sangamo Therapeutics and at BioMarin Pharmaceuticals, and he remains committed to driving the rapid development of novel treatments for rare diseases with unmet medical need. Dr. Vaidya completed his Infectious Diseases fellowship at Massachusetts General Hospital, his Internal Medicine and Pediatrics residency at the Icahn School of Medicine at Mount Sinai Hospital, and received his M.D. and Ph.D. degrees from the David Geffen School of Medicine at UCLA.

VANESSA VOGEL-FARLEY

United States

Vanessa is the Research and Data Governance Platform Lead for RARE-X and also serves as the Executive Director of the Dup15q Alliance. Previously, she was at the University of Minnesota Center for Neurobehavioral Development. She also served as the Clinical Research Coordinator for the Division of Developmental Medicine Laboratory of Cognitive Neuroscience, Boston Children's Hospital, working on collaborations with scientists from MIT and Harvard examining several clinical populations, including autism. She has 15 years of experience in data collection methods, as well as expertise in non-profit strategy, patient advocacy and research operations.

RENEE WELLS

Australia

For the last 5 years, Renee has been a metabolic specialist at Nutricia supporting the medical nutrition products and services for the inherited metabolic disorders (IMD) community. She a dietitian with over 19 years' experience working in a variety of roles in both the community and hospital environment in Australia and the United Kingdom in different specialised areas (burns, intensive care, infectious diseases, paediatrics, metabolics and Ketogenic diets). She joined the Nutricia Metabolics team in Australia in 2016 and has been a crucial link between Nutricia and the healthcare professionals working in metabolic medicine that support and manage people with HCU. She loves creating a meal out of fridge ingredients such as roasting up vegetables for a delicious salad.

MATHILDE YVERNEAU

France

Mathilde Yverneau completed her medical studies and pursued her pediatric training in Rennes (France), where she was certified as a pediatrician in 2020. Since, she is a pediatric fellow at Rennes University Hospital in the Department of Child and Adolescent Medicine. Her enthusiasm for nutrition research was evident early in her training: she completed a Master 2 through a year of research training in NuMeCan Institute (INRAE, INSERM, Rennes 1 University), evaluating the impact of postnatal nutrition on the intestinal endocrine function. Specialised in neonatalogy, she pursued her research training in a clinical study examining the clinical presentation of early-onset MTHFR deficiency under the supervision of Prof. Manuel Schiff. This study is integrated into the E-HOD ongoing larger research program

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At Travere Therapeutics, we are in rare for life.

We come together every day to help patients, families and caregivers of all backgrounds as they navigate life with a rare disease. On this path, we know the need for treatment options is urgent — that is why our global team works with the rare disease community to identify, develop and deliver life-changing therapies. In pursuit of this mission, we continuously seek to understand the diverse perspectives of rare patients and to courageously forge new paths to make a difference in their lives and provide hope — today and tomorrow.

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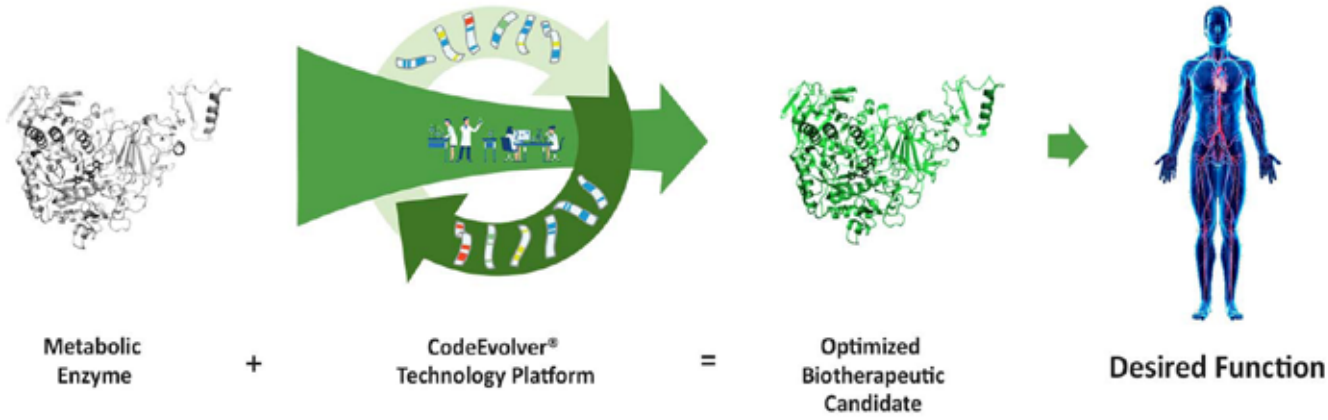
CODEXIS®

Codexis is a leading enzyme engineering company dedicated to the development of novel biotherapeutics and high-performance enzymes. Codexis is proud to support the Homocystinuria community, as well as other rare disease communities, through action. Our goals are to build disease awareness, improve quality of life for patients, and discover better treatments.
For more information, visit: www.codexis.com/therapeutics/

Designing Therapeutics for Patients

Therapeutic Targets

- Oral administration with meals
- Highly specific for the amino acid of interest
- Designed to work in the GI-tract



SHARE YOUR RECIPE

TO GO INTO THE DRAW TO

WIN

A NUTRIBULLET®

lowproteinconnect.com.au

WE WANT TO KNOW YOUR FAVOURITE LOPROFIN CULINARY CREATIONS!

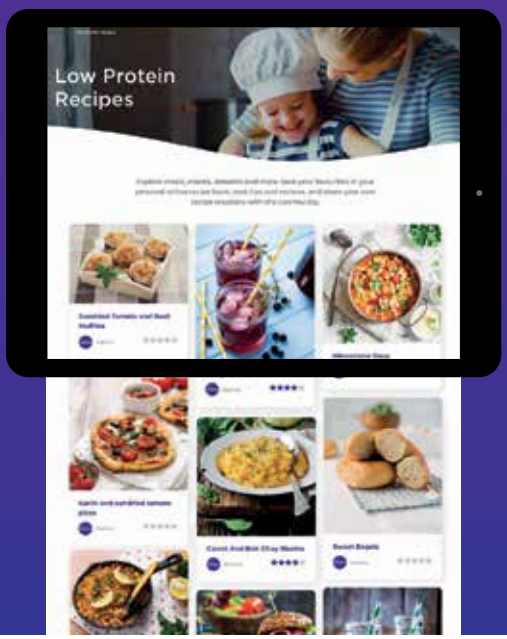
Scan the QR code to be inspired.

FOR YOUR CHANCE TO WIN 1 OF 10 NUTRIBULLET® BLENDERS or bonus product vouchers, tell us how you "Feed Your Possible" by sharing an original low protein recipe with the PKU community.

It could be your best breakfast, tastiest lunch, restaurant worthy dinner or all three!

TO ENTER:

1. Register on lowproteinconnect.com.au
2. Submit your original Loprofin low protein recipe via the Recipe Hub
3. Add a photo
4. Enter as many times as you like



Follow @lowproteinconnectanz #feedyourpossible

Loprofin is a food for special medical purposes. Use under medical supervision.

Recipe submission must be your own unique recipe and not already exist on the Nutricia website. Nutribullet® is not a participant in or sponsor of this promotion. Competition Entry Period is 20 September 2021 at 12:01am AEST to 13 December 2021 at 11:59pm AEST. Read full terms and conditions at lowproteinconnect.com.au/share-your-recipe-terms-and-conditions/. Image file size must not exceed 500kb and must be in png or jpeg format.

Nutricia Australia Pty Limited, Talavera Corporate Centre, Lvl 4, Building D, 12-24 Talavera Road, Macquarie Park, NSW 2113
Danone Nutricia NZ Ltd, 56-58 Aintree Avenue, Airport Oaks, Auckland 2022, New Zealand | lowproteinconnect.com.au

NUTRICIA METABOLICS
LIFE-TRANSFORMING NUTRITION

Delivering Metabolic Harmony. Transforming Life.

We are proud to support HCU Network Australia in its mission to help people living with Homocystinuria. Together, we can create hope for a better tomorrow.

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www.aeglea.com
Twitter: @aegleabio

THANK YOU TO THE **iHOPE 2021** SUPPORTERS

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cblC Brasil
Por Amor à Vida.

MOM
mães metabólicas
ASSOCIAÇÃO