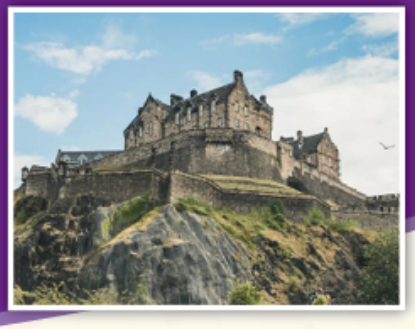
BIMDG

British Inherited Metabolic Disease Group

John McIntyre Conference Centre,
The University of Edinburgh



Pre-meeting
Monday 16 June 2025

Annual Symposium
Tuesday 17 - Wednesday 18 June 2025

Poster list - Both days

Alphabetical by first presenting author surname
Posters marked with ❖ will be presented as a flash presentation

Poster
Adjusting Dietary Therapy using Continuous Glucose Monitoring and Proposed Glycaemic Targets in Glycogen Storage Disease Type Ia: A Case Study **Cameron Arbuckle**[1], Dr Elaine Murphy[1] [1]National Hospital for Neurology and Neurosurgery, UK
Insights into aging among adults affected with Inherited Metabolic Diseases-one tertiary centre experience **Dr John Bassett**[1], Daniel Hand[1], Andrew Oldham[1], Emma Pickering[1], Dr Ana Jovanovic[1], Dr Samreen Safdar[1], Alison Woodall[1], Prof Emma Vardy[2,3], Dr Karolina M Stepien[1] [1]Mark Holland Adult Inherited Metabolic Diseases Unit, Salford Royal Hospital, Northern Care Alliance NHS Foundation Trust, [2]Oldham Care Organisation (part of the Northern Care Alliance NHS foundation trust), Rochdale Road, Oldham, OL1 2JH, [3]Manchester Academic Health Sciences Centre, School of Health Sciences, Faculty of Biology, Medicine and Health, The University of Manchester
Navigating Eliglustat Drug-Drug Interactions Through a Pharmacist-Led MDT Approach **Daniel Bell**[1], Alejandra de la Torre[1], Victoria Morgan[1], M Cherry May Sanchez[2] [1]Cambridge University Hospitals NHS FT, Cambridge, UK, [2]Cancer Research UK Cambridge Centre, Cambridge, UK
Shaping Phenylketonuria research through patient and public involvement **Giana Blaauw**[1], Prof Anita MacDonald[2], Dr Sarah Durnan[1], Prof Jane Coad[3] [1]Nottingham University Hospitals NHS Trust, Nottingham, England, [2]Birmingham Women's and Children's NHS Foundation Trust, Birmingham, England, [3]School of Health Sciences, University of Nottingham and University Hospitals Coventry & Warwickshir, Nottingham and Coventry, England

A prolonged release compared to an amino acid protein substitute in classical PKU effect on morning phenylalanine and tyrosine concentrations

Dr Anne Daly[1], Dr Sharon Evans[1], Dr Alex Pinto[1], Prof Anita MacDonald[1]
[1]Birmingham Children's Hospital, Birmingham, UK

Familial Hypercholesteremia in the Northwest of England: is it diagnosed too late?

Silothabo Dliso[1,2], Dr Clare Van Miert[2], Prof Joanne Blair[1]
[1]Alder Hey Children's NHS Foundation Trust, UK, [2]Liverpool John Moore's University, UK

Review of Patients Diagnosed with Biotin Thiamine Responsive Basal Ganglia disease in our centre

Megan Dorman[1]
[1]Great Ormond Street Hospital, London, UK

5,10-Methenyltetrahydrofolate synthetase deficiency (MTHFS deficiency): expanding the clinical and biochemical phenotype

Dr Sherry Fang[1], **Dr Simon Pope**[2], Dr Yael Hacohen[3], Dr Lara Menzies[4], Dr Preeya Rehsi[1]
[1]Department of Paediatric Inherited Metabolic Disease, Great Ormond Street Hospital for Children NHS, UK, [2]Neurometabolic Unit, Institute of Neurology, Queen Square, UK, [3]Paediatric Neurology, Great Ormond Street Hospital for Children NHS, UK, [4]Department of Clinical Genetics, Great Ormond Street Hospital for Children NHS, UK

❖ **Late-onset argininosuccinic aciduria unmasked by sodium valproate**

Robert Field[1,2], Hongying Chen[3], Aine Redmond[3], Alex Dudley[3], David Lewis-Smith[3], Eavan McGovern[3], Stephen Ryan[3,4], Loai Shakerdi[1], James O'Byrne[1]
[1]National Centre for Inherited Metabolic Disorders, Mater Misericordiae University Hospital, Ireland, [2]Department of Neurology, Mater Misericordiae University Hospital, Ireland, [3]Department of Neurology, Beaumont Hospital, Ireland, [4]Department of Neurology, Connolly Hospital Blanchardstown, Ireland

❖ **Adult Refsum Disease: Dietary management is more than only restricting phytanic acid intake**

Sarah Firman[1,2], Jennifer Cook[1,2], Imogen Hall[1,2], Katie Yeung[1,2], Steve King[1,2], Dr Raphael Buttigieg[2], Dr Fiona Vaz[2], Dr Anthony S. Wierzbicki[2], Dr Radha Ramachandran[2]
[1]Department of Nutrition and Dietetics, Guy's and St Thomas' NHS Foundation Trust, London, UK, [2]Adult Inherited Metabolic Diseases, Guy's and St Thomas' NHS Foundation Trust, London, UK

Nutritional management of a successful multifetal PKU Pregnancy

Suzanne Ford[1], Dr Nathan Cantley[1], Simone Whiteway[1], Dr Elisabeth Summers[1], Dr Helena Kemp[1]
[1]North Bristol NHS Trust, Bristol, England

❖ **Think Ammonia: The Survey**

Jonathan Gibson[1], Laura Smith van Carroll[1], Pavel Krepelka
[1]Metabolic Support UK, England

MADDening Results Secondary to Sertraline

Dr Jessica Johnson[1], Dr Mohamed Ahmed[2], Dr Godfrey T. Gillett[1], Dr Adam Lomas[1]
[1]Sheffield Teaching Hospitals NHS Foundation Trust, UK, [2]York and Scarborough Teaching Hospitals NHS Foundation Trust, UK

❖ **Development of patient education/support for patients with Hereditary Fructose Intolerance (HFI) under the Addenbrooke's Metabolic Service**

Lisa Gaff[1], Sarah Donald[1], Ruby Cross[1], **Dr Eleni Leventea**[1], Dr Elizabeth Caller[1]
[1]Addenbrookes Hospital, England

Attention to Sapropterin Administration: Can it Bring Clinical Advantage?

Prof Anita MacDonald[1], Dr Martina Tosi[2], Dr Alex Pinto[1], Dr Anne Daly[1], Dr Catherine Ashmore[1], Dr Sharon Evans[1]

[1]Department of Dietetics, Birmingham Women's and Children's Hospital, UK, [2]Department of Health Sciences, University of Milan; Department of Pediatrics, Vittore Buzzi Children's Hospital, Italy

Post-baseline outcomes of the UK Early Access to Medicines Scheme registry for cipaglucosidase alfa plus miglustat in late-onset Pompe disease

Dr Elaine Murphy[1], Dr Duncan Cole[2], Dr Patrick Deegan[3], Prof Tarekegn Geberhiwot[4], Prof Derralynn Hughes[5], Dr Robin Lachmann[6], Dr Reena Sharma[7], Vipul Jain[8], Elizabeth Moffat[9], Jasmine Rutecki[8], Dr Sophie Clarke[9], Prof Mark Roberts[10]

[1]Charles Dent Metabolic Unit, National Hospital for Neurology and Neurosurgery, University College London Hospitals, UK, [2]University Hospital of Wales, UK, [3]Lysosomal Disorders Unit, Addenbrooke's Hospital, UK, [4]Institute of Metabolism and Systems Research, University of Birmingham, UK, [5]Lysosomal Storage Disorders Unit, Royal Free London NHS Foundation Trust and University College London, UK, [6]National Hospital for Neurology and Neurosurgery, University College London Hospitals, UK, [7]The Mark Holland Metabolic Unit, Salford Royal Foundation NHS Trust, UK, [8]Amicus Therapeutics, Inc., USA, [9]Amicus Therapeutics UK Ltd, UK, [10]Salford Royal NHS Foundation Trust, UK

❖ **Does sertraline cause symptomatic non-genetic MADD?**

Dr R.P.M.M.R. Pathirana[1], Dr Ann Bowron[1], Dr Roy Talbot[1], Dr Yi Shiau Ng[2]

[1]Department of Blood Sciences, Royal Victoria Infirmary, The Newcastle Upon Tyne Hospitals, UK, [2]Department of Neurosciences, Royal Victoria Infirmary, UK

❖ **Healthcare provision and patient's outcomes in adolescents with Phenylketonuria: A UK centre experience**

Alex Pinto[1], Dr Anne Daly[1], Dr Sharon Evans[1], Catherine Ashmore[1], Prof Anita MacDonald[1]

[1]Birmingham Children's Hospital, UK

Miglustat: a first-in-class enzyme stabiliser for late-onset Pompe disease

Professor Mark Roberts[1], Professor Tahseen Mozaffar[2], Dr Barry J Byrne[3], Professor Mazen M Dimachkie[4], Dr Robert J Hopkin[5], Professor Priya S Kishnani[6], Professor Benedikt Schoser[7], Professor Ans T van der Ploeg[8], Dr Jon Brudvig[9], Dr Brian Fox[9], Dr Fred Holdbrook[9], Mr Vipul Jain[9], Mr Franklin Johnson[9], Dr Jennifer Zhang[9], Professor Giancarlo Parenti[10]

[1]Salford Royal NHS Foundation Trust, UK, [2]University of California, USA, [3]University of Florida, USA, [4]University of Kansas Medical Center, USA, [5]Cincinnati Children's Hospital Medical Center, USA, [6]Duke University Medical Center, USA, [7]Friedrich-Baur-Institute, LMU University Hospital, LMU Munich, Germany, [8]Erasmus MC University Medical Center, Netherlands, [9]Amicus Therapeutics, Inc., USA, [10]Metabolic Unit, University of Naples Federico II, Italy

❖ **Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD) and sleeve gastrectomy for weight management**

Louise Robertson[1], Alice Johnson[2], Alistair Sharples[3], Dr Charlotte Dawson[1]

[1]University Hospitals Birmingham NHS Foundation Trust, UK, [2]Royal Wolverhampton NHS Trust, UK, [3]University Hospitals of North Midlands NHS Trust, UK

True Faces of Rare: Preferences for authentic imagery in disorder-specific materials by people living with rare diseases and their communities

Laura Smith Van Carroll[1], Pavel Krepelka[1], Kirsty Hoyle[1], Dr Kamran Iqbal[2], Dr Tom Kenny[2]

[1]Metabolic Support UK, Manchester, UK, [2]Chiesi UK, Manchester, UK

Distinctive phenotype associated with Mitochondrial ATAD3 gene cluster duplication: a case series

Dr Srividya Sreekantam[1], Dr Suresh Vijayaraghavan[1], Dr Saikat Santra[1], Dr Julian Raiman[1], Ms Louise Simmons[1], Dr Jane Cassidy[1], Chris Stockdale[1], Dr Mrinalini Rajimwale[2], Dr Sreedhara Nittur[3], Dr Swati Naik[1]

[1]Birmingham Women's and Children's Hospital, [2]Coventry Hospital, [3]Singleton Hospital, Swansea

❖ **Multisystem Involvement and Hypoalbuminemia in N-linked Congenital Disorder of Glycosylation (CDG): A Case Series** **Dr Nirubhan Veeraraghavan**[1], Dr Hugh Lemonde[1], Dr Helen Mundy[1], Dr Dinusha Pandithan[1] [1]Guys and St Thomas Hospital NHS foundation trust, London, UK
Decompensating Inherited Metabolic Disorders and the Oral Emergency Regimen. The Adult Patient Experience in Bristol **Simone Whiteway**[1], Suzanne Ford[1] [1]North Bristol NHS Trust
Elevation of branched chain amino acids due to Branched Chain Amino Acid Transferase 2 deficiency (BCAT2): to treat or not to treat? **Dr Guido Zago**[1], Dr Marios Kaliakatsos[2], **Dr Nazreen Kamarus Jaman**[1], Dr Martina Messina[1], Alice Dianin[3], Dr Laura Rubert[3], Dr Andrea Bordugo[3], Dr Giulia Rodella[3], Dr Elena Piccoli[3], Dr Preeya Rehsi[1] [1]Department of Paediatric Metabolic Medicine, Great Ormond Street Hospital NHS Trust, London, UK, [2]Department of Paediatric Neurology, Great Ormond Street Hospital, London, UK, [3]Inherited Metabolic Diseases Unit and Regional Centre for Newborn Screening, Diagnosis and Treatment of Inherited Metabolic Diseases and Congenital Endocrine Diseases, Azienda Ospedaliera Universitaria Integrata, Verona, Italy, Verona, Italy

Poster list - Wednesday only posters

Alphabetical by first presenting author surname

Poster
Difficulties in diagnosing mild peroxisomal biogenesis disorders – a case example **Sharon Colyer**[1], Claire Hart[1], Jennie Raven[2], John Shepherd[2] [1]Sheffield Children's Hospital, UK, [2]Hull Royal Infirmary, UK
KS-462-282: An alternative Krabbe Disease biomarker with excellent diagnostic potential **James Cooper**[1], Kate Neal[1], Christine Egerton[1], Karen Tylee[1], Dr Heather Church[1], Teresa HY Wu[1] [1]Manchester University NHS Foundation Trust, UK
Evaluating Reference Intervals for Galactose-1-Phosphate Uridyl Transferase: Integrating Biochemical and Genetic Data **Philip Crook**[1], Catherine Bradford [1]Synnovis Analytics Ltd., London, UK
The Efficacy of Liquid Valine and Isoleucine Supplements in MSUD **Dr Anne Daly**[1], Dr Martina Tosi[2], Dr Alex Pinto[1], Dr Catherine Ashmore[1], Dr Sharon Evans[1], Prof Anita MacDonald[1] [1]Department of Dietetics, Birmingham Women's and Children's Hospital, UK, [2]Department of Health Sciences, University of Milan; Department of Pediatrics, Vittore Buzzi Children's Hospital, Italy
Elevated 4-methylsterol and 4,4-dimethylsterol in microcephaly, congenital cataract and psoriasiform dermatitis syndrome due to methylsterol monooxygenase 1 (MSMO1) deficiency provides diagnostic and treatment response biomarker **Dr James Davison**[1], Dr Youssef Khalil[2], Dr Catherine Dennis[3], Dr Tess McPherson[4], Professor Peter Clayton[2] [1]Metabolic Medicine, Great Ormond Street Hospital, UK, [2]UCL GOS Institute of Child Health, UK, [3]Clinical Genetics, Oxford University Hospitals, UK, [4]Dermatology, Oxford University Hospitals, UK
Evaluation of neurodevelopmental outcome after haematopoietic stem cell transplant (HSCT) in a patient with Triosephosphate isomerase deficiency (TPI-D) **Carey Eldred**[1], Sian Waller[1], Dr Robert Chiesa[2], Dr James Davison[1] [1]Great Ormond Street Children's Hospital - Metabolic Medicine Department, UK, [2]Great Ormond Street Hospital - Bone Marrow Transplant Department, UK
FollowME Fabry Pathfinders registry: patient-reported outcomes in a cohort of patients on migalastat treatment for median 4 years **Derralynn Hughes**[1], Ulla Feldt-Rasmussen[2], Gere Sunder-Plassmann[3], Aneal Khan[4], Biliana O. Veleva-Rotse[5], Joseph D. Giuliano[5], Hai Jiang[5], Vipul Jain[5], Jasmine Rutecki[5], Aleš Linhart[6] [1]Royal Free London NHS Foundation Trust and University College London, UK, [2]Rigshospitalet, Copenhagen University Hospital, Denmark, [3]Medical University of Vienna, Austria, [4]M.A.G.I.C. (Metabolics and Genetics in Canada) Clinic Ltd., University of Calgary, Cumming School of Medicine, Canada, [5]Amicus Therapeutics, Inc., USA, [6]Charles University and General University Hospital, Czech Republic

Retrospective Analysis of Vitamin B12 Dose Optimization in Methylmalonic acidaemias and Cobalamin Deficiency-Related Disorders: Insights from a Tertiary Metabolic Centre in the UK

Dr Nazreen Kamarus Jaman[1], Dr Guido Zago[1], Dr James Davison[1], Dr Stephanie Grunewald[1], Mrs Melanie Mcsweeny[1], Miss Megan Dorman[1], Mrs Marjorie Dixon[1], Miss Rachel Skeath[1]

[1]Great Ormond Street Hospital, London, UK

Continuous Glucose Monitoring in the GSD cohort of Addenbrooke's Metabolic Service

Dr Eleni Leventea[1], Sarah Donald[1], Lisa Gaff[1]

[1]Addenbrooke'a Hospital, Cambridge, UK

Barriers and facilitators to clinical trial participation: improving accessibility, logistics, and awareness

Helen Lycett[1], Samantha Wiseman[1], Alexandra Morrison[1]

[1]Rare Disease Research Partners, Amersham, UK

Primary and secondary multiple acyl-coA dehydrogenase deficiency (MADD): clinical insights and treatment response

Dr Eamon Mccarron[1], Dr Emma Murray[1], Neil Gilmore[1], Dr Gillian Hamilton[1], Sharon Coyler[2], Joanne Croft[2], Camilla Scott[2], Clarie Hart[2], Dr Grainne Connolly[1], Dr Michael Kinney[1], Dr John McConville[3], Dr Ashley Elliott[1,4]

[1]Royal Victoria Hospital Belfast, Northern Ireland, [2]Sheffield Childrens Hospital, England, [3]Ulster Hospital, Northern Ireland, [4]Wellcome-Wolfson Institute of Experimental Medicine, Queen's University Belfast, Northern Ireland

Role of tele monitoring in rare disease management

Alison Mckee[1], Nicola Mcstravick[1], Sophie Houghton[1], Dr Grainne Connolly[1], Dr Emma Murray[1], Laura Murphy[1], Joy Mcdonald[1]

[1]Belfast Health and Social Care Trust, Belfast, Northern Ireland

Phenylketonuria (PKU) - A Transition Model for Rare Disease

Nicola McStravick[1], Alison McKee[1], Sophie Houghton[1], Dr Grainne Connolly[1], Anne Grimsley[1], Nicky Courtney[1], Dr Siobhan O'Sullivan[1], Dr Caroline Hart[1], Emma Tracey[1]

[1]Belfast Health and Social Care Trust, Belfast, UK

Glycogen Storage Disease type 3 & Ketogenic Diet Therapy – Case Report

Camille Newby[1], **Abigail Robotham**[1], Victoria Wilkins[1], Sam Whiting[1], Catherine Armstrong[1], Germaine Pierre[1], Effie Chronopoulou[1]

[1]Bristol Royal Hospital for Children, UK

Disorder or distraction: Considering the significance of an ACADSB gene variant in a young person with developmental delay

Dr Mahdia Sami Obeda[1], **Dr James Nurse**[1], Dr Paul Cook[1], Dr Hugh Lemonde[2], Dr Katherine Lachlan[1]

[1]Southampton General Hospital, UK, [2]Evelina Children's Hospital, UK

How good is blood Phe control in Maternal PKU in Europe: results from 102 pregnancies

Alex Pinto[1], Dr Kirsten Ahring[2], Manuela Ferreira Almeida[3], Catherine Ashmore[1], Sarah Bailey[4], Dr Amaya Bélanger-Quintana[5], Prof Alberto Burlina[6], Dr Duncan Cole[4], Clare Dale[7], Dr Anne Daly[1], Esther van Dam[8], Dr Charlotte Dawson[7], Dr Sharon Evans[1], Sarah Firman[9], Diane Green[10], Dr Tarekegn Hiwot[7], Yteke Hoekstra[8], Sarah Howe[7], Dr Fatma Ilgaz[11], Christian Loro[6], Nicola McStravick[12], Katie Rawlins[9], Louise Robertson[7], Prof Júlio César Rocha[13], Iris Rodenburg[8], Dr Danja Schulenburg-Brand[4], Prof Francjan van Spronsen[8], Dr Gisela Wilcox[10], Alison Wodall[10], Prof Anita MacDonald[1]

[1]Birmingham Children's Hospital, UK, [2]Copenhagen University Hospital, Rigshospitalet, Copenhagen, Denmark, [3] Centro de Referência na área de Doenças Hereditárias do Metabolismo, Unidade Local de Saúde de Santo António, E.P.E. (ULSSA), , Portugal, [4]University Hospital of Wales, Wales, [5]Hospital Universitario Ramón y Cajal, , Spain, [6]Padova University Hospital, , Italy, [7]University Hospitals Birmingham , , UK, [8]University Medical Centre Groningen, University of Groningen, , The Netherlands, [9]Guy's & St Thomas' NHS Foundation Trust, UK, [10]Salford Royal Foundation Trust, UK, [11]Hacettepe University, Turkey, [12]Royal Victoria Hospital, UK, [13]NOVA Medical School (NMS), Faculdade de Ciências Médicas, (FCM), Universidade Nova de Lisboa, Portugal

Are Cystatin-C measurements useful for the surveillance of chronic kidney disease in Paediatric MMA patients?

Dr Preeya Rehsi[1], Ms Gemma Meenan[1], Dr Matko Marlais[2], Ms Melanie McSweeney[1]

[1]Department of Paediatric Inherited Metabolic Disease Great Ormond Street Hospital NHS Foundation Trust and Institute for Child Health London UK, [2]Department of Paediatric Nephrology, Great Ormond Street Hospital for Children, London, UK

Managing the untreatable - the broader role of rapid whole genome sequencing in neurological regression

Dr Saule Gasiunas[1], **Dr Dexter Tarr**[1], Dr Richard Curnock[1]

[1]Willink Unit, St. Mary's Hospital, UK

Optimising Ketogenic Therapy in Multiple Acyl-CoA Dehydrogenase Deficiency: A Case Report

Dr Fiona Vaz[1], **Sarah Firman**[1], Roqsana Ara[1], Jennifer Cook[1], Imogen Hall[1], Katie Yeung[1], Dr Radha Ramachandran[1]

[1]Guy's and St Thomas' NHS Foundation Trust, UK

Optimising Peripheral Intravenous Catheter Insertion and Management in Out-of-Hospital Settings: Addressing Patient and Technical Factors, Enhancing Nurse Competency, and Promoting Evidence-Based Practices for Improved Outcomes

Leoni Walker[1]

[1]Lloyds Clinical, Harlow, UK

An unusual case of argininosuccinic aciduria highlighting the challenges and importance of identification of argininosuccinic acid in amino acid analysis

Dr Vicki Warburton[1], Rebecca Hopkins[1], Kinga Krzywdzinska[1], Kris-Mae Dela Isla[1], Tim Thorpe[1], Dr Paul Cook[2], Dr Efstathia Chronopoulou[3], Dr Germaine Pierre[3]

[1]University Hospitals Bristol and Weston NHS Foundation Trust, UK, [2]University Hospital Southampton NHS Foundation Trust, UK, [3]Bristol Royal Hospital for Children, UK

Discrepant urine biochemistry in a paediatric patient with Alkaptonuria

Courtney Watt[1]

[1]NHS Greater Glasgow and Clyde, Scotland

A Rare cause of rhabdomyolysis in a 12 year old girl

Dr Ralph Wigley[1], Dr Alistair Horman[1], Dr Julien Baruteau[1]

[1]Great Ormond Street Hospital, UK

A case of X-linked form of combined Methylmalonic aciduria and Homocysteinemia - Cbl X

Natalia Olkhovich[1,2], **Yuliia Zhyvytsia**[1], Maryna Patsora[1], Natalia Samonenko[1], Natalia Pichkur[1], Natalia Mytsyk[1], Oksana Barvinska[1], Iryna Hrehul[1], Svitlana Kormoz[1], Yuliia Tymruk[1], Olena Kutsyk[1], Maryna Shulga[1], Natalia Petrenko[1], Lina Moshkivska[1], Anna Shilova[1] Tetiana Ivanova[1] Natalia Gorovenko[2,3]

[1]National Ukranian Childrens Hospital OKHMATDYT, [2]Institute of Genetic and Regenerative Medicine, Kyiv, Ukraine, [3]National University of Health named P.L. Shupyka, Kyiv, Ukraine