



Pre-meeting
Monday 29 June 2026



British Inherited Metabolic Disease Group

Leonardo Royal Hotel, Oxford

Annual Symposium
Tuesday 30 June -
Wednesday 1 July 2026



Poster List

Posters listed by presenting day then alphabetical by Presenting Author Surname.

Poster	Theme	Presenting Day
Bloodspot phenylalanine monitoring frequency in adults with phenylketonuria: A service improvement project Sarah Bailey¹ ¹ All Wales Inherited Metabolic Disorder Service, Cardiff, Wales	Audit	Tuesday
Two cases of taurine transporter (SLC6A6) deficiency presenting with retinopathy and severe hypotaurinaemia Roger Bramley¹ , Dr Sergei Korenev ² , Robert Barski ¹ ¹ Leeds Teaching Hospitals NHS Trust, UK, ² Manchester University NHS Foundation Trust, UK	Clinical	Tuesday
Elevated 2,3-dihydroxy-2-methylbutyric acid on urine organic acid analysis: a case series Roger Bramley¹ , Robert Barski ¹ ¹ Leeds Teaching Hospitals NHS Trust, UK	Clinical	Tuesday
Galactose-1-phosphate uridyltransferase (GALT) testing for Classical Galactosaemia (CG): indications and impact on human milk feeding (HMF) Laura Byers¹ , Dr Anupam Chakrapani ² , Dr James Davison ² , Katie Harvey ⁴ , Kelly Watson ¹ , Camilla Natale ³ , Marjorie Dixon¹ ¹ Dietetics Department, Great Ormond Street Hospital, NHS Foundation Trust, London, UK, ² Department of Metabolic Medicine, Great Ormond Street Hospital, London, UK, ³ MSc Dietetics, UCL Division of Medicine, University College London, London, UK, ⁴ Chemical Pathology, Paediatric Laboratory Medicine, Great Ormond Street Hospital, London, UK	Audit	Tuesday
Determinants of successful human milk feeding in inherited metabolic disorders (IMD) on dietary treatment Laura Byers¹ , Ide Herlihy¹ , Chloe Millington ¹ , Dr Anupam Chakrapani ² , Dr Spyros Batzios ² , Marjorie Dixon ¹ ¹ Dietetics Department, Great Ormond Street Hospital, London, UK, ² Department of Metabolic Medicine, Great Ormond Street Hospital, London, UK	Audit	Tuesday
Protein Substitutes: challenges applying International Dysphagia Diet Standardisation Initiative standards (IDDSI) for an aging PKU community Lisa Gaff¹ , Simone Whiteway ² ¹ Cambridge University Hospitals NHS Foundation Trust, Cambridge, England, ² North Bristol NHS Trust, Westbury-on-Trym, England	Clinical	Tuesday
An unusual presentation of Alpha-methylacyl-CoA racemase deficiency in a patient with elevated creatine kinase Dr Laura Hancox¹ , Dr Wycliffe Mbagaya ¹ , Dr Charlotte Dawson ² ¹ University Hospitals Bristol and Weston NHS Foundation Trust, UK, ² University Hospitals Birmingham NHS Foundation Trust, UK	Clinical	Tuesday

<p>PAH genetics and sapropterin responsiveness in paediatric NI patients Dr Caroline Hart¹, Dr Siobhan O'Sullivan ¹Royal Belfast Hospital for Sick Children, Northern Ireland</p>	Audit	Tuesday
<p>followME Fabry Pathfinders registry: cardiac and renal effectiveness in a multi-national, multi-centre cohort of patients on migalastat treatment for ≥5 years Prof Derralynn Hughes¹, Joseph Giuliano², Prof Aleš Linhart³, Prof Antonio Pisani⁴, Prof Olga Azevedo⁵, Jennifer Hiros⁶, Vipul Jain⁶, Dr Biliana Veleva-Rotse⁶, Dr Peter Nordbeck⁷ ¹Lysosomal Storage Disorders Unit, Royal Free London NHS Foundation Trust and University College London, UK, ²Rare Medical Affairs LLC, USA, ³Second Department of Internal Cardiovascular Medicine, First Faculty of Medicine, Charles University and General University Hospital, Czechia, ⁴Department of Public Health, Nephrology Unit, Federico II University Hospital, Italy, ⁵Cardiology Department, Reference Center of Lysosomal Storage Disorders, MetabERN member, Hospital Senhora da Oliveira, Portugal, ⁶Amicus Therapeutics, Inc., USA, ⁷Internal Medicine I, University Hospital Würzburg, Germany</p>	Clinical	Tuesday
<p>Epidemiology of Inherited Metabolic Disorders in a Specialist Paediatric Metabolic Clinic in India: A Single-Centre Registry Analysis of 349 Cases Dr Gauri Krishna¹ ¹Neurogen Metabolics</p>	Clinical	Tuesday
<p>Hereditary Fructose Intolerance presenting with hepatic dysfunction and hypoglycaemia and prior to exposure to dietary fructose: Diagnostic Pitfalls Parveen Leghari¹, Catherine Zephir¹, Bernd C Schwahn^{1,2} ¹Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester University NHS Foundation Trust, Health Innovation, UK, ²Division of Evolution & Genomic Sciences, School of Biological Sciences, Faculty of Biology, Medicine and Health, University of Manchester, Manchester, UK</p>	Clinical	Tuesday
<p>Cardiac involvement in adults with Glycogen Storage Disease Type IIIa: an observational cohort study Dr Eamon Mccarron¹, Dr Adam Lomas¹, Dr Godfrey Gillett¹, Andrew Oldham², Dr Alison Woodhall², Dr Rob Cooper³, Dr Oliver Watson⁴, Dr Ana Jovanovic², Dr Reena Sharma², Dr Karolina Stepien² ¹Adult Inherited Metabolic Disorders, Sheffield Teaching Hospitals NHS Foundation Trust, UK, ²Adult Inherited Metabolic Disorders, Salford Care Organisation, Northern Care Alliance NHS Foundation Trust, UK, ³Cardiology Department, Liverpool Heart and Chest Hospital NHS Foundation Trust, UK, ⁴Cardiology Department, Northern General Hospital, Sheffield Teaching Hospitals NHS Foundation Trust, UK</p>	Research	Tuesday
<p>From Guidance to Practice: An Audit of PKU Management in Northern Ireland Dr Kerri Munn-bookless¹, Dr Niamh McCool², Dr Caroline Hart¹ ¹Paediatric Metabolic Medicine, RBHSC, Northern Ireland, ²Paediatric Haematology Oncology, RBHSC, Northern Ireland</p>	Audit	Tuesday
<p>Impact on biochemical markers following reduced dose olipudase alfa in adults with acid sphingomyelinase deficiency Dr Elaine Murphy¹, Kirsty White¹, Ana Amado Fondo¹, Dr Robin Lachmann¹ ¹University College London NHS Foundation Trust, UK</p>	Clinical	Tuesday
<p>Understanding the experiences and support needs of families living with Ketotic Hypoglycaemia to inform service delivery Dr Katie Redwood¹, Camille Newby, Naimo Abdullah ¹UHBW NHS, Bristol, UK</p>	Clinical	Tuesday

<p>7.5-year efficacy outcomes with cipaglucosidase alfa plus miglustat in adults with Pompe disease in ATB200-02, an open-label Phase I/II study Prof Mark Roberts¹, Dr Drago Bratkovic², Dr Barry J Byrne³, Prof Paula R Clemens⁴, Dr Ozlem Goker-Alpan⁵, Prof Priya S Kishnani⁶, Prof Tahseen Mozaffar⁷, Dr Nadine AME van der Beek⁸, Farah Amon⁹, Dr Brian Fox⁹, Dr Fred Holdbrook⁹, Vipul Jain⁹, Prof Benedikt Schoser¹⁰ ¹Salford Royal NHS Foundation Trust, UK, ²PARC Research Clinic, Royal Adelaide Hospital, Australia, ³University of Florida, USA, ⁴Department of Neurology, University of Pittsburgh School of Medicine and VA Pittsburgh Healthcare System, USA, ⁵Lysosomal and Rare Disorders Research and Treatment Center, USA, ⁶Duke University Medical Center, USA, ⁷Department of Neurology, University of California, USA, ⁸Erasmus MC University Medical Center, Netherlands, ⁹Amicus Therapeutics, Inc., USA, ¹⁰Friedrich-Baur-Institute at the Department of Neurology, LMU University Hospital, LMU Munich, Germany</p>	Clinical	Tuesday
<p>Long-term complications in methylmalonic acidaemia in adults Dr Samreen Safdar¹, Mairead Jones¹, Alison Woodall¹, Priscilla Sia¹, Dr Mahad Muhammad Safdar², Faiza Adrees¹, Dr John Bassett¹, Dr Jonathan Meyer¹, Dr Karolina M Stepien¹, Dr Ana Jovanovic¹, Dr Reena Sharma¹ ¹Salford Royal Hospital, Northern Care Alliance NHS Foundation Trust, UK, ²University of Liverpool, UK</p>	Clinical	Tuesday
<p>Multifactorial Progressive Osteoporosis in Adults with PMM2-CDG: A Five-Patient Case Series Dr Samreen Safdar¹, Andrew Oldham¹, Janet Gorton¹, Dr Jonathan Meyer¹, Shaheer Muhammad Safdar², Dr John Bassett¹, Dr Karolina Stepien¹, Dr Ana Jovanovic¹, Dr Reena Sharma¹ ¹Salford Royal Hospital, Northern Care Alliance NHS Foundation trust, Salford, UK, ²University of Lancaster, UK</p>	Clinical	Tuesday
<p>Intrafamilial phenotypic variability in m.3243A>G mitochondrial disease: Implications for cascade testing and multisystem surveillance Dr Samreen Safdar¹, Andrew Oldham¹, Aiza Safdar², Faiza Adrees¹ ¹Salford Royal Hospital, Northern Care Alliance NHS Foundation Trust, UK, ²University of Manchester, UK</p>	Clinical	Tuesday
<p>Factors affecting long-term outcomes in a cohort of adults with LC-FAODs Zona Ismail¹, Dr John Bassett¹, Andrew Oldham¹, Alison Woodall¹, Dr Karolina M Stepien¹, Dr Ana Jovanovic¹, Dr Samreen Safdar¹, Dr Reena Sharma¹ ¹Adult Inherited Metabolic Diseases, Salford Care Organisation, Northern Care Alliance NHS Foundation Trust, Salford, Greater Manchester, Salford, UK</p>	Clinical	Tuesday
<p>When a Simple Test Saves a Life: Neonatal Galactosemia Diagnosed by Benedict Test in a Resource Limited Setting Dr Pavithra Samarakoon¹, Dr Harsha Minasinghe², Dr Thayani Arooran¹, Dr Y Hemika¹, Dr MFS Mihika¹, Dr KD Rammuthupura¹, Dr E Jasinghe¹ ¹Department of chemical Pathology, Colombo, Sri Lanka, ²Department of Paediatrics, Colombo, Sri Lanka</p>	Clinical	Tuesday
<p>Diagnostic Delay and Divergent Phenotypes in Metachromatic Leukodystrophy: Real-World Evidence Supporting Newborn Screening in the Era of Gene Therapy Dr Berna Seker Yilmaz^{1,2}, Dr Anupam Chakrapani³, Dr James Davison³, Dr Helen Mundy², Dr Preeya Rehsi³, Dr Roshni Vara², Prof Paul Gissen^{1,3}</p>	Clinical	Tuesday

<p>Rare Disease Awareness in Young People in the UK: Insights from a Survey Gabriel Tipper¹, Zeynep Ada Yilmaz², Prof Paul Gissen^{3,4}, Dr Berna Seker Yilmaz^{3,5} ¹Department of Biomedical Sciences, University of Exeter, UK, ²School of Film, Media, and Creative Technologies, University of Portsmouth, UK, ³ Genetics and Genomic Medicine Department, Great Ormond Street Institute of Child Health, University College London, UK, ⁴Department of Paediatric Metabolic Medicine, Great Ormond Street Hospital for Children NHS Foundation Trust, UK, ⁵Department of Paediatric Inherited Metabolic Disease, Evelina Children's Hospital, Guy's, and St Thomas' NHS Foundation Trust, UK</p>	Research	Tuesday
<p>Review of prescribing practice in Urea Cycle Disorders- one adult centre experience Dr Karolina Stepien¹, Dr John Bassett, Dr Jonathan Meyer, Dr Reena Sharma, Dr Ana Jovanovic, Dr Samreen Safdar, Dr Alison Woodall, Faiza Adrees ¹Salford Royal Hospital, Northern Care Alliance NHS Foundation Trust, Salford, UK</p>	Clinical	Tuesday
<p>Pancreatitis in Two Adolescents with Argininosuccinic Aciduria: Expanding the Phenotypic Spectrum Chern Yan Tan¹, Dr Andrew Morris¹ ¹Willink Metabolic Unit, Manchester, UK</p>	Clinical	Tuesday
<p>Natural History and Comorbidities of Childhood Ketotic Hypoglycaemia: Experience from a Regional Specialist Centre Chern Yan Tan¹, Dr Sameera Auckburally², Prof Indi Banerjee², Dr Arunabha Ghosh¹ ¹Willink Metabolic Unit, Manchester, UK, ²Department of Paediatric Endocrinology, Royal Manchester Children's Hospital</p>	Clinical	Tuesday
<p>Timeliness in PKU Care: A National Audit into Turnaround Time of Dried Blood Spots and Frequency of appointments for Adults with PKU in the UK Simon Tapley¹, Benjamin Hayes¹, Natalie Shaw², Bailey Sarah³, Cameron Arbuckle⁴, Louise Robertson⁵, Lisa Gaff⁶, Imogen Hall⁷, Katie Yeung⁷, Nicola McStravick⁸, Alison McKee⁸ ¹University Hospitals Bristol and Weston, UK, ²Newcastle Upon Tyne Hospitals NHS Foundation Trust, UK, ³University Hospital of Wales, UK, ⁴University College London Hospitals, UK, ⁵University Hospitals Birmingham, UK, ⁶Cambridge University Hospitals, UK, ⁷Guy's and St Thomas NHS Foundations Trust, UK, ⁸Belfast Health and Social Care Trust, Northern Ireland</p>	Audit	Tuesday
<p>Short-term use of a slow-release Phenylalanine-free microtablet protein substitute in children and adolescents with Phenylketonuria Dr Martina Tosi¹, Dr Anne Daly¹, Dr Sharon Evans¹, Catherine Ashmore¹, Dr Alex Pinto¹, Prof Anita MacDonald¹ ¹Birmingham Women's and Children's Hospital, UK</p>	Research	Tuesday
<p>Defining the inherited metabolic disorder landscape in numbers using structured databases: proportion and mortality across EU-5 Dr Elise Wilby¹, James Stanway², Laura Smith Van Carroll¹ ¹Metabolic Support, England, ²Keele University, England</p>	Research	Tuesday
<p>Two-Year Sapropterin Outcomes in Phenylketonuria: Longitudinal Study of Metabolic, Dietary, Psychosocial Effects Dr Ozlem Yilmaz Nas^{1,2}, Catherine Ashmore¹, Maria Ines Gama¹, Dr Anne Daly¹, Dr Sharon Evans¹, Dr Alex Pinto¹, Prof Yahya Ozdogan², Prof Anita MacDonald¹ ¹Birmingham Women's and Children's NHS Foundation Trust, UK, ²Department of Nutrition and Dietetics, Ankara Yildirim Beyazıt University, 06760, Turkey</p>	Research	Tuesday

<p>Use of skin testing to advise therapy options in Pompe patients following suspected IgE mediated infusion associated reactions (IAR) Ana Amado Fondo¹, Anna Katherina Cuarentas¹, Kirsty White¹, Tigisti Beyene¹, Dr Joanna Lukawska, Dr Elaine Murphy, Dr Robin Lachmann ¹UCLH, London, UK</p>	Clinical	Wednesday
<p>Review of genotype-phenotype correlation regarding sapropterin responsiveness in adult patients living with PKU Dr Kylie Beale¹, Simon Tapley¹, Suzanne Ford¹, Dr Wycliffe Mbagaya¹, Dr Nathan Cantley¹ ¹Bristol NHS Group, UK</p>	Clinical	Wednesday
<p>A single-centre patient experience survey of parents and carers of children on low protein diets Lucy Blackstone¹, Victoria Holmes¹, Zixin Luo¹, Leah Paulden¹, Emma Simpson¹, Karen Van Wyk¹, Georgina Wood¹, Catherine Zephir¹ ¹Willink Biochemical Unit, Manchester, England</p>	Research	Wednesday
<p>Variability in dietary management in Isovaleric Acidaemia (IVA): A retrospective longitudinal cohort study Sarah Cawtherley¹, Louise Van Dorp¹, Cameron Arbuckle⁴, Dr Martina Messina^{2,3}, Dr Spyros Batzios², Dr Emma Footitt², Dr Anupam Chakrapani², Rachel Skeath¹, Dr Julein Baruteau², Dr James Davison², Dr Stephanie Grunewald², Dr Alexander Broomfield², Dr Preeya Rehsi², Marjorie Dixon¹ ¹Dietetics, Great Ormond Street Hospital for Children, NHS Foundation Trust, London, UK, London, UK, ²Department of Paediatric Metabolic Medicine, Great Ormond Street Hospital for Children, NHS Foundation Trust, London, UK, London, UK, ³Genetics and Genomic Medicine Department, GOS Institute of Child Health, University College London, London, UK, London, UK, ⁴Charles Dent Metabolic Unit, University College London Hospitals, London, UK, London, UK</p>	Audit	Wednesday
<p>Cholestatic Jaundice with Hypocalcaemia: One Pathway and a Unifying Diagnosis Sharon Colyer¹, Claire Hart¹, Katherine Wright¹, Dr Elizabeth Griffiths² ¹Department of Clinical Chemistry, South Yorkshire & Bassetlaw Pathology, Sheffield Children's Hospital, UK, ²Department of Paediatric Gastroenterology, Sheffield Children's Hospital, UK</p>	Clinical	Wednesday
<p>The holistic management of Late Onset Pompe Disease through nurse and physio led clinics-the Birmingham experience Sarah Steeds¹, Nicola Condon¹, Rachel Curran¹, Dr Charlotte Dawson¹ ¹University Hospitals Birmingham, UK, ²University Hospitals Birmingham, UK, ³University Hospitals Birmingham, UK, ⁴University Hospitals Birmingham, UK</p>	Clinical	Wednesday
<p>Sleep Disturbance in Mucopolysaccharidosis Type III (Sanfilippo Syndrome): A Service Review of Pharmacological Management Efficacy and Safety Dr Daniela Castillo Garcia¹, Clare Hamilton-Kay¹, Sarah Phillippo¹, Rachel Tetlow¹, Dr Sarah Hulley¹, Dr Arubnabha Ghosh¹, Dr Richard Curnock¹, Dr Maria De Castro Lopez¹ ¹St. Mary's Hospital, Manchester NHS Foundation Trust, Manchester, UK</p>	Audit	Wednesday
<p>Peri-operative Management Guide for Inherited Metabolic Diseases Eva Fox¹, Dr Caroline Hart² ¹Ulster University, Northern Ireland, ²Royal Belfast Hospital for Sick Children, Northern Ireland</p>	Clinical	Wednesday
<p>Development of an erythrocyte debrancher enzyme assay to biochemically confirm a diagnosis of GSD IIIb Rohit Hirachan¹, Dr Ralph Wigley¹, Dr Alistair Horman¹ ¹Enzymes Unit, Chemical Pathology, Great Ormond Street Hospital for Children NHS Foundation Trust</p>	Research	Wednesday

<p>Complications of Central Venous Access in Metabolic Patients Undergoing Hemofiltration Dr Nazreen Banu Kamarus Jaman¹, Dr James Davison¹, Dr Preeya Rehsi¹, Megan Dorman¹, Dr Emma Footitt¹, Dr Rola Zaatra¹ ¹Great Ormond Street Hospital, UK</p>	Audit	Wednesday
<p>Development of a selective HPPD inhibitor without an off-target liability Domen Kämpjut¹, Petros Tyrakis¹, Jonathan Lindström¹, Nigel Ramsden², Oliver Maddocks¹ ¹Faeth Therapeutics, Cambridge, UK, ²Rxcelerate, Cambridge, UK</p>	Research	Wednesday
<p>Sleep study severity as a predictor of intraoperative airway difficulty in Mucopolysaccharidosis patients: a single-centre experience Dr Christos Lazaridis¹, Dr Lois Mackley², Dr Marie-Claire Raphael², Dr Riona Tully², Dr Elena Targa², Mathilda Antonini¹, Rebecca Whiteley¹, Megan Keating¹, Dr Preeya Rehsi¹, Dr James Davison¹, Dr Anupam Chakrapani¹, Dr Emma Footitt¹, Dr Elaine Chan³, Dr Amaki Sogbodjor², Prof Paul Gissen^{1,4}, Dr Alexander Broomfield¹ ¹Department of Paediatric Metabolic Medicine, Great Ormond Street Hospital for Children NHS Foundation Trust, UK, ²Department of Paediatric Anaesthetic Medicine, Great Ormond Street Hospital for Children NHS Foundation Trust, UK, ³Department of Paediatric Respiratory Medicine, Great Ormond Street Hospital for Children NHS Foundation Trust, UK, ⁴UCL Great Ormond Street Institute of Child Health, UK</p>	Research	Wednesday
<p>Continuous glucose monitoring in assessment of susceptibility to ketotic hypoglycaemia in children diagnosed with spinal muscular atrophy in infancy on disease-modifying therapies Dr Sophie Manoy¹, Dr Marita Smith¹, Kate Munro², Antonia Wright², Dr Anita Cairns², Dr Matthew Lynch^{1,2} ¹Queensland Lifespan Metabolic Medicine Service, Queensland Children's Hospital, Australia, ²Department of Neurosciences, Queensland Children's Hospital, Australia</p>	Research	Wednesday
<p>Glutathione Synthetase Deficiency: a rare differential diagnosis of neonatal metabolic acidosis Dr Hebatallah Mansour^{1,2}, Dr Chern Tan^{1,2}, Dr Bernd Schwahn^{1,2,3} ¹Royal Manchester Children's Hospital, UK, ²Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester University NHS Foundation Trust, Health Innovation Manchester, UK, ³Division of Evolution & Genomic Sciences, School of Biological Sciences, Faculty of Biology, Medicine and Health, University of Manchester, UK</p>	Clinical	Wednesday
<p>Determinants of Bone Density Change in Gaucher Disease: A Multisite Mixed-Effects Analysis Dr Carmen Minea¹, Dr Boliang Yu¹, Dr Patrick B. Deegan¹ ¹Cambridge University Hospitals, England</p>	Research	Wednesday
<p>Future proofing The Metabolic Service: MCADD Clinic – a nurse led approach Claire Mizler¹, Nichola Cowie¹, Hannah Dixon¹, Rebecca Herbert¹ ¹Sheffield Children's Hospital Foundation Trust, UK</p>	Clinical	Wednesday
<p>A homozygous intronic variant in NDUFAF8 causes optic atrophy and learning difficulties Dr Maria Moustakis¹, K Kavanagh², LS Taylor^{3,4}, D Brosnahan⁵, S Chamney^{5,6}, K McCreery⁶, M O'Regan⁷, D O'Rourke⁸, RW Taylor^{3,4}, CL Alston^{3,4}, C Howard¹ ¹National Centre for Inherited Metabolic Disorders, CHI at Temple Street, Dublin 1, Ireland, ²National Centre for Inherited Metabolic Disorders, Mater Misericordiae University Hospital, Dublin 7, Ireland, ³Newcastle Highly Specialised Mitochondrial Diagnostic Service, Newcastle upon Tyne Hospitals NHS Foundation Trust, UK, ⁴Mitochondrial Research Group, Translational and Clinical Research Institute, Faculty of Medical Sciences., Newcastle, UK, ⁵Department of Paediatric Ophthalmology, CHI at Crumlin, Dublin 12, Ireland, ⁶Department of Paediatric Ophthalmology, CHI at Temple Street, Dublin 1, Ireland, ⁷Department of Paediatric Neurology, CHI at Crumlin., Dublin 12, Ireland, ⁸Department of Paediatric Neurology, CHI at Temple Street., Dublin 1, Ireland</p>	Clinical	Wednesday

<p>Enhancing Enzyme Replacement Therapy in Lysosomal Storage Disorders: Faster Infusion Rates Across the UK Antonio Ochoa-ferraro¹, Zoe Taylor², Faiza Adrees³, Daniel Bell⁴, Susan McMahon⁵, Evelyn Frank⁶, Chin Gan⁷, Siema Akram⁸, Niamh Finnegan⁹, Robert Dugdale¹⁰, Clara Monaghan¹¹, Thomas Wyllie¹² ¹University Hospitals Birmingham NHS Foundation Trust, Birmingham, UK, ²All Wales Adult Inherited Metabolic Disease Service (AWIMDS), University Hospital of Wales, Cardiff and Vale University Health Board, UK, ³Salford Royal NHS Foundation Trust, Northern Care Alliance NHS Foundation Trust (NCA), UK, ⁴Cambridge University Hospitals NHS Foundation Trust, UK, ⁵Inherited Metabolic Disorders Service, Royal Hospital for Children, UK, ⁶University College London Hospitals NHS Foundation Trust, UK, ⁷Great Ormond Street Hospital NHS Foundation Trust, UK, ⁸Birmingham Children’s Hospital, Birmingham Women’s and Children’s NHS Foundation Trust, UK, ⁹Royal Free London NHS Foundation Trust, UK, ¹⁰Royal Manchester Children’s Hospital, Manchester University NHS Foundation Trust, UK, ¹¹Royal Belfast Hospital for Sick Children, Belfast Health and Social Care Trust, UK, ¹²Noah’s Ark Children’s Hospital for Wales, Cardiff and Vale University Health Board, UK</p>	Clinical	Wednesday
<p>Metabolic Pharmacist Roles Across the UK: Current Practice and Service Provision Antonio Ochoa-ferraro¹, Robert Dugdale², Daniel Bell³, Georgina Smith⁴, Siema Akram⁵, James Roddick⁶, Priya Dattani⁷, Clayton Wong⁸, Clara Monaghan⁹, Faiza Adrees¹⁰, Trinh Huynh¹¹, Chin Gan¹², Alice Ruston¹³, Thomas Wyllie¹⁴ ¹University Hospitals Birmingham NHS FT, UK, ²Royal Manchester Children’s Hospital, Manchester University NHS FT, UK, ³Cambridge University Hospitals NHS FT, UK, ⁴Nottingham University Hospitals NHS Trust, UK, ⁵Birmingham Children’s Hospital, Birmingham Women’s and Children’s NHS FT, UK, ⁶Inherited Metabolic Disorders in Scotland (IMD Scotland), NHS Greater Glasgow and Clyde, UK, ⁷University Hospitals of Leicester NHS Trust, UK, ⁸Royal Free London NHS Foundation Trust, UK, ⁹Royal Belfast Hospital for Sick Children, UK, ¹⁰Salford Royal NHS Foundation Trust, Northern Care Alliance NHS Foundation Trust (NCA), UK, ¹¹Evelina London Children’s Hospital, Guy’s and St Thomas’ NHS FT, UK, ¹²Great Ormond Street Hospital For Children NHS FT, UK, ¹³Sheffield Teaching Hospitals NHS FT, UK, ¹⁴Noah’s Ark Children’s Hospital for Wales, Cardiff and Vale University Health Board, UK</p>	Clinical	Wednesday
<p>Service improvement: Clinical Nurse Specialist (CNS) led clerking of Neuronal Ceroid Lipofuscinosis (CLN2) Batten disease patients Katie Raven¹, Anja Keating¹, Dr Laura Guilder¹ ¹UHBW, Bristol, UK</p>	Clinical	Wednesday
<p>Galactosaemia and cheese: cheese analysis for lactose and galactose content in the UK, 2022–2026 Louise Robertson^{1,2}, Sue Bevington², Karen Ford², Simon Tapley³, Prof Anita MacDonald⁴ ¹University Hospitals Birmingham NHS Foundation Trust, UK, ²Galactosaemia Support Group, UK, ³University Hospitals Bristol and Weston NHS Foundation Trust, UK, ⁴Birmingham Women’s and Children’s Foundation Trust, UK</p>	Research	Wednesday
<p>An Exercise and Dietary Induced Fat-Free Mass Increase in Two Patients with Phenylketonuria (PKU) Sarah Howe¹, Louise Robertson², Clare Dale³ ¹University Hospitals Birmingham, Birmingham, UK, ²University Hospitals Birmingham, Birmingham, UK, ³University Hospitals Birmingham, Birmingham, UK</p>	Clinical	Wednesday
<p>Characterization of Maple Syrup Urine Disease in a Chilean Cohort: Genotypic Variations and Metabolic Control Maria Florencia Salazar¹, Verónica Cornejo¹, Felipe Peñaloza¹, Eugenia Rojas-Agurto¹, Catalina Arancibia¹, Carolina Arias¹, Juan Francisco Cabello^{1,2}, Pilar Peredo¹, Vicente Quiroz¹, María Jesús Leal-Witt¹ ¹Laboratory Of Metabolic Diseases, Inta, University of Chile, Chile, ²Department of Neurology, Boston Children’s Hospital, USA</p>	Research	Wednesday

<p>Estimating prevalence of long chain fatty acid oxidation disorders in the UK Saikat Santra¹, Erru (Christy) Yang², Lay Ean Tan², Nin Dass² ¹Birmingham Women's and Children's NHS Foundation Trust, UK, ²Ultragenyx Pharmaceutical Inc., UK</p>	Research	Wednesday
<p>Multiple acyl CoA dehydrogenase deficiency (MADD): single-centre review of paediatric clinical, dietetic and biochemical disease severity spectrum Rachel Skeath¹, Erin Emmett¹, Alistair Horman¹, Ralph Wigley¹, Spyros Batzios¹, Laura Byers¹, Anupam Chakrapani¹, Sarah Cawtherley¹, Marjorie Dixon¹, Megan Dorman¹, Jennifer Dunne¹, Emma Footitt¹, Ide Herlihy¹, Mel McSweeney¹, Chloe Millington¹, Sophie Taylor¹, Karin Tuschl¹, James Davison¹ ¹Great Ormond Street Hospital for Children NHS Foundation Trust, UK</p>	Clinical	Wednesday
<p>Cognitive and Behavioural Profiles of Children and Young People with Inherited Metabolic Disorders: A Retrospective Clinical Audit Molly Smith¹, Magdalini Hadjikalli¹, Dr Clare Williams¹, Dr Emma Matthews¹ ¹Great Ormond Street Hospital NHS Foundation Trust, UK</p>	Audit	Wednesday
<p>Paediatric Aldolase A deficiency: Case report, treatment & biomarkers of an ultra-rare inherited metabolic disorder Dr Dexter Tarr¹, Victoria Whiteley¹, Prof Judith JM Jans, James Cooper¹, Oliver Parkes¹, Dr Bernd Schwahn¹ ¹Manchester Foundation Trust, UK, ²UMC Utrecht, Netherlands</p>	Clinical	Wednesday
<p>Lived Experience: Insights into Familial Chylomicronaemia Syndrome Dr Elise Wilby¹, Jill Prawer², Laura Smith van Carroll¹ ¹Metabolic Support, UK, ²Action FCS, UK</p>	Research	Wednesday
<p>Single-centre experience of parenteral nutrition when acutely managing methylmalonic acidaemia and propionic acidaemia Georgina Wood¹, Dr Dexter Tarr¹, Dr Arunabha Ghosh¹ ¹Willink Biochemical Genetics Unit, St Mary's Hospital, Manchester University NHS Foundation Trust, UK</p>	Clinical	Wednesday