# 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 No | Poster |
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| 1 | **Adjusting Dietary Therapy using Continuous Glucose Monitoring and Proposed Glycaemic Targets in Glycogen Storage Disease Type Ia: A Case Study**<br>**Cameron Arbuckle**[1], Dr Elaine Murphy[1]<br>[1]National Hospital for Neurology and Neurosurgery, UK |
| 2 | **Insights into aging among adults affected with Inherited Metabolic Diseases-one tertiary centre experience**<br>**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]<br>[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 |
| 3 | **Navigating Eliglustat Drug-Drug Interactions Through a Pharmacist-Led MDT Approach**<br>**Daniel Bell**[1], Alejandra de la Torre[1], Victoria Morgan[1], M Cherry May Sanchez[2]<br>[1]Cambridge University Hospitals NHS FT, Cambridge, UK, [2]Cancer Research UK Cambridge Centre, Cambridge, UK |
| 4 | **Shaping Phenylketonuria research through patient and public involvement**<br>**Giana Blaauw**[1], Prof Anita MacDonald[2], Dr Sarah Durnan[1], Prof Jane Coad[3]<br>[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 |

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| **5** | **A prolonged release compared to an amino acid protein substitute in classical PKU effect on morning phenylalanine and tyrosine concentrations**<br>**Dr Anne Daly**[1], Dr Sharon Evans[1], Dr Alex Pinto[1], Prof Anita MacDonald[1]<br>[1]Birmingham Children's Hospital, Birmingham, UK |
| **6** | **Attention to Sapropterin Administration: Can it Bring Clinical Advantage?**<br>Prof Anita MacDonald**[1]**, Dr Martina Tosi[2], Dr Alex Pinto[1], **Dr Anne Daly**[1], Dr Catherine Ashmore[1], Dr Sharon Evans[1]<br>[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 |
| **7** | **Familial Hypercholesteremia in the Northwest of England: is it diagnosed too late?**<br>**Silothabo Dliso**[1,2], Dr Clare Van Miert[2], Prof Joanne Blair[1]<br>[1]Alder Hey Children's NHS Foundation Trust, UK, [2]Liverpool John Moore's University, UK |
| **8** | **Review of Patients Diagnosed with Biotin Thiamine Responsive Basal Ganglia disease in our centre**<br>**Megan Dorman**[1]<br>[1]Great Ormond Street Hospital, London, UK |
| **9** | ❖ **Late-onset argininosuccinic aciduria unmasked by sodium valproate**<br>**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]<br>[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 |
| **10** | ❖ **Adult Refsum Disease: Dietary management is more than only restricting phytanic acid intake**<br>**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]<br>[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 |
| **11** | **Nutritional management of a successful multifetal PKU Pregnancy**<br>**Suzanne Ford**[1], Dr Nathan Cantley[1], Simone Whiteway[1], Dr Elisabeth Summers[1], Dr Helena Kemp[1]<br>[1]North Bristol NHS Trust, Bristol, England |
| **12** | ❖ **Think Ammonia: The Survey**<br>**Jonathan Gibson**[1], Laura Smith van Carroll[1], Pavel Krepelka<br>[1]Metabolic Support UK, England |
| **13** | ❖ **Development of patient education/support for patients with Hereditary Fructose Intolerance (HFI) under the Addenbrooke's Metabolic Service**<br>Lisa Gaff[1], Sarah Donald[1], Ruby Cross[1], **Dr Eleni Leventea**[1], Dr Elizabeth Caller[1]<br>[1]Addenbrookes Hospital, England |
| **14** | **MADDening Results Secondary to Sertraline**<br>Dr Jessica Johnson**[1]**, Dr Mohamed Ahmed[2], Dr Godfrey T. Gillett[1], **Dr Adam Lomas**[1]<br>[1]Sheffield Teaching Hospitals NHS Foundation Trust, UK, [2]York and Scarborough Teaching Hospitals NHS Foundation Trust, UK |

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| **15** | **Post-baseline outcomes of the UK Early Access to Medicines Scheme registry for cipaglucosidase alfa plus miglustat in late-onset Pompe disease**<br>**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]<br>[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 |
| **16** | ❖ **Does sertraline cause symptomatic non-genetic MADD?**<br>**Dr R.P.M.M.R. Pathirana**[1], Dr Ann Bowron[1], Dr Roy Talbot[1], Dr Yi Shiau Ng[2]<br>[1]Department of Blood Sciences, Royal Victoria Infirmary, The Newcastle Upon Tyne Hospitals, UK, [2]Department of Neurosciences, Royal Victoria Infirmary, UK |
| **17** | ❖ **Healthcare provision and patient's outcomes in adolescents with Phenylketonuria: A UK centre experience**<br>**Alex Pinto**[1], Dr Anne Daly[1], Dr Sharon Evans[1], Catherine Ashmore[1], Prof Anita MacDonald[1]<br>[1]Birmingham Children's Hospital, UK |
| **18** | **5,10-Methenyltetrahydrofolate synthetase deficiency (MTHFS deficiency): expanding the clinical and biochemical phenotype**<br>Dr Sherry Fang[1], **Dr Simon Pope**[2], Dr Yael Hacohen[3], Dr Lara Menzies[4], Dr Preeya Rehsi[1]<br>[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 |
| **19** | **Miglustat: a first-in-class enzyme stabiliser for late-onset Pompe disease**<br>**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]<br>[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 |
| **20** | ❖ **Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD) and sleeve gastrectomy for weight management**<br>**Louise Robertson**[1], Alice Johnson[2], Alistair Sharples[3], Dr Charlotte Dawson[1]<br>[1]University Hospitals Birmingham NHS Foundation Trust, UK, [2]Royal Wolverhampton NHS Trust, UK, [3]University Hospitals of North Midlands NHS Trust, UK |
| **21** | **True Faces of Rare: Preferences for authentic imagery in disorder-specific materials by people living with rare diseases and their communities**<br>**Laura Smith Van Carroll**[1], Pavel Krepelka[1], Kirsty Hoyle[1], Dr Kamran Iqbal[2], Dr Tom Kenny[2]<br>[1]Metabolic Support UK, Manchester, UK, [2]Chiesi UK, Manchester, UK |

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| **22** | **Distinctive phenotype associated with Mitochondrial ATAD3 gene cluster duplication: a case series**<br>**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]<br>[1]Birmingham Women's and Children's Hospital, [2]Coventry Hospital, [3]Singleton Hospital, Swansea |
| **23** | ❖ **Multisystem Involvement and Hypoalbuminemia in N-linked Congenital Disorder of Glycosylation (CDG): A Case Series**<br>**Dr Nirubhan Veeraraghavan**[1], Dr Hugh Lemonde[1], Dr Helen Mundy[1], Dr Dinusha Pandithan[1]<br>[1]Guys and St Thomas Hospital NHS foundation trust, London, UK |
| **24** | **Decompensating Inherited Metabolic Disorders and the Oral Emergency Regimen. The Adult Patient Experience in Bristol**<br>**Simone Whiteway**[1], Suzanne Ford[1]<br>[1]North Bristol NHS Trust |
| **25** | **Elevation of branched chain amino acids due to Branched Chain Amino Acid Transferase 2 deficiency (BCAT2): to treat or not to treat?**<br>**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]<br>[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 No | Poster |
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| 26 | **Difficulties in diagnosing mild peroxisomal biogenesis disorders – a case example**<br>**Sharon Colyer**[1], Claire Hart[1], Jennie Raven[2], John Shepherd[2]<br>[1]Sheffield Children's Hospital, UK, [2]Hull Royal Infirmary, UK |
| 27 | **KS-462-282: An alternative Krabbe Disease biomarker with excellent diagnostic potential**<br>**James Cooper**[1], Kate Neal[1], Christine Egerton[1], Karen Tylee[1], Dr Heather Church[1], Teresa HY Wu[1]<br>[1]Manchester University NHS Foundation Trust, UK |
| 28 | **Evaluating Reference Intervals for Galactose-1-Phosphate Uridyl Transferase: Integrating Biochemical and Genetic Data**<br>**Philip Crook**[1], Catherine Bradford<br>[1]Synnovis Analytics Ltd., London, UK |
| 29 | **The Efficacy of Liquid Valine and Isoleucine Supplements in MSUD**<br>**Dr Anne Daly**[1], Dr Martina Tosi[2], Dr Alex Pinto[1], Dr Catherine Ashmore[1], Dr Sharon Evans[1], Prof Anita MacDonald[1]<br>[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 |
| 30 | **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**<br>**Dr James Davison**[1], Dr Youssef Khalil[2], Dr Catherine Dennis[3], Dr Tess McPherson[4], Professor Peter Clayton[2]<br>[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 |
| 31 | **Evaluation of neurodevelopmental outcome after haematopoietic stem cell transplant (HSCT) in a patient with Triosephosphate isomerase deficiency (TPI-D)**<br>**Carey Eldred**[1], Sian Waller[1], Dr Robert Chiesa[2], Dr James Davison[1]<br>[1]Great Ormond Street Children's Hospital - Metabolic Medicine Department, UK, [2]Great Ormond Street Hospital - Bone Marrow Transplant Department, UK |
| 32 | **An unusual case of argininosuccinic aciduria highlighting the challenges and importance of identification of argininosuccinic acid in amino acid analysis**<br>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]<br>[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 |

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| **33** | **FollowME Fabry Pathfinders registry: patient-reported outcomes in a cohort of patients on migalastat treatment for median 4 years**<br>**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]<br>[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 |
| **34** | **Retrospective Analysis of Vitamin B12 Dose Optimization in Methylmalonic acidaemias and Cobalamin Deficiency-Related Disorders: Insights from a Tertiary Metabolic Centre in the UK**<br>**Dr Nazreen Kamarus Jaman**[1], Dr Guido Zago[1], Dr James Davison[1], Dr Stephanie Grunewald[1], Melanie Mcsweeny[1], Megan Dorman[1], Marjorie Dixon[1], Rachel Skeath[1]<br>[1]Great Ormond Street Hospital, London, UK |
| **35** | **Continuous Glucose Monitoring in the GSD cohort of Addenbrooke's Metabolic Service**<br>**Dr Eleni Leventea**[1], Sarah Donald[1], Lisa Gaff[1]<br>[1]Addenbrooke'a Hospital, Cambridge, UK |
| **36** | **Barriers and facilitators to clinical trial participation: improving accessibility, logistics, and awareness**<br>**Helen Lycett**[1], Samantha Wiseman[1], Alexandra Morrison[1]<br>[1]Rare Disease Research Partners, Amersham, UK |
| **37** | **Primary and secondary multiple acyl-coA dehydrogenase deficiency (MADD): clinical insights and treatment response**<br>**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 Estelle Healy[1], Dr Grainne Connolly[1], Dr Michael Kinney[1], Dr John McConville[3], Dr Ashley Elliott[1,4]<br>[1]Royal Victoria Hospital Belfast, Northern Ireland, [2]Sheffield Children's Hospital, England, [3]Ulster Hospital, Northern Ireland, [4]Wellcome-Wolfson Institute of Experimental Medicine, Queen's University Belfast, Northern Ireland |
| **38** | **Role of tele monitoring in rare disease management**<br>**Alison Mckee**[1], Nicola Mcstravick[1], Sophie Houghton[1], Dr Grainne Connolly[1], Dr Emma Murray[1], Laura Murphy[1], Joy Mcdonald[1]<br>[1]Belfast Health and Social Care Trust, Belfast, Northern Ireland |
| **39** | **Phenylketonuria (PKU) - A Transition Model for Rare Disease**<br>**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]<br>[1]Belfast Health and Social Care Trust, Belfast, UK |
| **40** | **Glycogen Storage Disease type 3 & Ketogenic Diet Therapy – Case Report**<br>**Camille Newby**[1], **Abigail Robotham**[1], Victoria Wilkins[1], Sam Whiting[1], Catherine Armstrong[1], Germaine Pierre[1], Effie Chronopoulou[1]<br>[1]Bristol Royal Hospital for Children, UK |
| **41** | **Disorder or distraction: Considering the significance of an ACADSB gene variant in a young person with developmental delay**<br>Dr Mahdia Sami Obeda[1], **Dr James Nurse**[1], Dr Paul Cook[1], Dr Hugh Lemonde[2], Dr Katherine Lachlan[1]<br>[1]Southampton General Hospital, UK, [2]Evelina Children's Hospital, UK |

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| **42** | **How good is blood Phe control in Maternal PKU in Europe: results from 102 pregnancies**<br>**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]<br>[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 |
| **43** | **Are Cystatin-C measurements useful for the surveillance of chronic kidney disease in Paediatric MMA patients?**<br>**Dr Preeya Rehsi**[1], Ms Gemma Meenan[1], Dr Matko Marlais[2], Ms Melanie McSweeney[1]<br>[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 |
| **44** | **Managing the untreatable - the broader role of rapid whole genome sequencing in neurological regression**<br>Dr Saule Gasiunas[1], **Dr Dexter Tarr**[1], Dr Richard Curnock[1]<br>[1]Willink Unit, St. Mary's Hospital, UK |
| **45** | **Optimising Ketogenic Therapy in Multiple Acyl-CoA Dehydrogenase Deficiency: A Case Report**<br>**Dr Fiona Vaz**[1], **Sarah Firman**[1], Roqsana Ara[1], Jennifer Cook[1], Imogen Hall[1], Katie Yeung[1], Dr Radha Ramachandran[1]<br>[1]Guy's and St Thomas' NHS Foundation Trust, UK |
| **46** | **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**<br>**Leoni Walker**[1]<br>[1]Lloyds Clinical, Harlow, UK |
| **47** | **Discrepant urine biochemistry in a paediatric patient with Alkaptonuria**<br>**Courtney Watt**[1]<br>[1]NHS Greater Glasgow and Clyde, Scotland |
| **48** | **A Rare cause of rhabdomyolysis in a 12 year old girl**<br>**Dr Ralph Wigley**[1], Dr Alistair Horman[1], Dr Julien Baruteau[1]<br>[1]Great Ormond Street Hospital, UK |
| **49** | **A case of X-linked form of combined Methylmalonic aciduria and Homocysteinemia - Cbl X**<br>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]<br>[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 |