BIMDG Programme – ICC Wales June 2024

'Breaking Boundaries'

RCPath accreditation for 11 CPD Credits

Tuesday 11th June

09:00 - 09:30 **Registration, Coffee & Exhibition Stands**

09:30 - 12:00 Parallel sessions

Adult IMD meeting
Clinical and Scientific trainees meeting
Clinical Pharmacists meeting
Dieticians meeting
Nurses meeting
Paediatric CSAC meeting
Paediatric IMD and link clinicians meeting
Psychologists meeting

10:30 - 11:00 **Coffee Break/Exhibition Stands**

12:00 - 13:00 **Lunch/Exhibition Stands**

13:00 - 14:30	**Session 1: The Cutting Edge of IMDs**	**Chairs: Dr Saikat Santra and Dr Duncan Cole**
13:00 - 13:30	Clinical experience with newly approved IMD treatments	Prof Simon Jones, Manchester Children's Hospital
13:30 - 14:00	Givosiran in the Acute Porphyrias	Dr Danja Schulenburg-Brand, University Hospital of Wales, Cardiff
14:00 - 14:30	Update on WGS for Newborn Screening	Dr Karina Stone, Genomics England
14:30 - 14:45	**Tea Break/Exhibition Stands**	
14:45 - 15:15	Challenges in managing very rare disorders	Dr Charlotte Dawson, Queen Elizabeth Hospital, Birmingham

15:15 - 15:39	**Flash Posters – 3 minutes each**	**Chairs: Dr Duncan Cole, Dr Saikat Santra**
	Analysis of the Prevalence of Neurodiversity and Anxiety in UHB Adult PKU patients	Sarah Howe
	Guidance for the Dietetic Management of Adults with Classical Galactosaemia in the UK	Louise Robertson
	Practical considerations for instigating treatment and service user reported outcomes in the management of Guanidinoacetate Methyltransferase (GAMT) deficiency: A dietetic perspective	Sarah Bailey
	Navigating Diagnostic Challenges in a Phenotypically Suspected Case of Niemann-Pick Type C: Exome vs. Biochemical Approaches	Katherine Sloss
	A rare case of secondary carnitine deficiency due to chronic dietary restriction causing hyperammonaemic encephalopathy	Nathan Cantley
	Treatment of Fanconi-Bickel syndrome with SGLT2i	Helen Mundy
	Lipoic acid synthetase deficiency: a possible mimic for non-accidental injury?	James Nurse
	A case report in FBPase and pregnancy	Imogen Hall

15:39 - 16:15 (poster presenters available from 15:45)	**Poster presentations/Tea break/Exhibition Stands**	
16:15 - 17:15	**Session 2: Working across borders**	**Chair: Dr Gisela Wilcox**
16:15 - 16:45	Managing IMD patients across challenging geographies	Prof Chris Hendriksz, FYMCA Medical
16:45 - 17:15	The role of the Advanced Practitioner in IMDs	Anita Inwood, Service Director and Metabolic Nurse Practitioner, Queensland, Australia
17:30 - 18:30	**BIMDG AGM**	**MEMBERS ONLY**
19:30	**Conference Dinner**	

Wednesday 12th June

08:30 - 09:00	**Registration/Coffee & Exhibition Stands**	
09:00 - 10:30	**Parallel Session 3a: Advancements in Diagnostics for IMDs**	**Chair: Prof Stuart Moat**
09:00 - 09:30	The Syndrome Without a Name (SWAN) clinic	Dr Graham Shortland OBE, C&V UHB
09:30 - 10:00	Lipidomics and Metabolomics for Rare Disease Diagnosis	Prof Bill Griffiths, Swansea University
10:00 -10:30	The role of genomics in IMD Diagnosis; panel discussion	
09:00 -10:30	**Parallel Session continued from 11th Dietetics**	
10:30 - 11:00	**Coffee Break/Exhibition Stands**	
11:00 - 13:00	**Maggie Lilburn Lecture and Members' Papers**	**Chair: Dr Julian Raiman**
11:00 - 11:30	Maggie Lilburn Lecture	Fiona White, Willink Unit, Manchester

11:30 - 13:00	**Members' Papers 10 minutes each including questions**	
	Plasma ammonia analysis: is transport of samples on ice required? An investigation in samples from patients with hyperammonaemia	Ann Bowron
	Atypical cases of Hereditary tyrosinaemia type 1	Karolina Witek
	Liberalisation of diet after 6 years of age in Glutaric Aciduria Type 1 (GA1) - variation in practice across UK metabolic centres	Sarah Cawtherley
	Ornithine transcarbamylase deficiency: a multicentre natural history study	Berna Seker Yilmaz
	Blood phenylalanine levels in patients with Phenylketonuria from Europe between 2012 to 2018: is it a changing landscape?	Alex Pinto
	Tolerability of pegunigalsidase alfa across the clinical program: integrated analysis of infusion-related reactions by prior enzyme replacement therapy	Derralynn Hughes
13:00 - 14:00	**Lunch/Exhibition Stands**	
14:30 - 16:00	**Session 4: Transitions**	**Chair: Dr Gisela Wilcox**
14:00 - 14:30	Educating Healthcare Professionals about Rare Diseases	Dr Lucy McKay, CEO Medics 4 Rare Disease

14:30 - 16:00	Transitioning from paediatric to adult care – short presentations and Panel discussion.	Medic – Prof Chris Hendriksz Psychologist – Dr Stuart Rust, Manchester Foundation Trust Nurse - Liz Morris, Lead specialist Nurse for the Lysosomal Disorders Unit, Addenbrooke's Hospital, Cambridge Dietician - Clare Dale
16:00	**Close**	

***** Programme subject to minor changes

BIMDG Conference has been support by an education grant from the following Pharmaceutical Companies. They have had no input into the programme or choice of speakers.

Advanz Pharma; Amicus; Biomarin Europe Ltd; Cenote Pharma; Chiesi; Denali Therapeutics; Immedica; Lucane Pharma; MetaHealthcare; Nutricia Metabolics, Orchard-TX, Sanofi, Takeda, Theravia, Travere Therapeutics Ireland Ltd; Vitaflo International Ltd