

Day 1: Wednesday 7th June 2023

MORNING

08:30-09:00

REGISTRATION, COFFEE, EXHIBITION STANDS, POSTERS

09:00-12:00

**PARALLEL SESSIONS: WITH COFFEE BREAK
10:30-11:00**

- Dietitians Meeting
 - Clinical & Scientific Trainees Meeting
 - Nurses Meeting
 - Adult IMD Meeting
 - Clinical Pharmacists Meeting
 - Clinical Psychologists Meeting
 - Clinical & Scientific Training Committee Meeting
- *(running in to afternoon)

12:00-13:00

LUNCH AND EXHIBITION STANDS/POSTERS

AFTERNOON

SESSION 1

Adult Inherited Metabolic Disorders Session
CHAIR: Elaine Murphy, UCLH

13:00-13:10

Introduction & housekeeping

13:10-13:40

The role of the gynaecologist in supporting patients with IMD - from menarche to menopause – Ms Melanie Davies.

13:40-14:10

Movement disorders in IMD – Dr Sam Shribman

14:10-14:40

Maggie Lilburn Lecture – Dietary modification in GSD IIIa: a literature review and our experience at the Evelina London Children's Hospital' - Joanna Gribben

14:40-15:00

TEA BREAK , EXHIBITION STANDS, POSTERS

SESSION 2

**Paediatric Inherited Metabolic Disorders Session
Part A.** CHAIR: James Davison, GOSH

15:00-15:30

Optimising transition – Melanie McSweeney

15:30-16:00

Ethical Issues in neonatal screening using whole genome sequencing – Prof Dominic Wilkinson

16:00-16:35

Using patient material to develop therapies for IEMs: PA and MMA as an example - Prof Marshall Summar

16:35-17:10

Flash Posters

3 minute presentations of selected posters

Siyamini Sivananthan, GOSH
(Delayed diagnosis of Niemann-Pick Type C with a deep-intronic mutation)

Nana Ghansah, Neurometabolic Unit London
(The importance of age related reference intervals
for adult metabolite testing in the post genomic era)

Charlotte Ellerton, UCLH
(Audit of maternal iodine intakes during pregnancy in
PKU)

Maria Ines Gama, Birmingham Children's Hospital
(Change in quality of diet and burden of care in children
with PKU treated with sapropterin dihydrochloride: a
longitudinal study)

Sarah Hogg, Cambridge University Hospital
(Audit assessing the quality of dried blood spot
specimens received by UK metabolic laboratories for the
biochemical monitoring of phenylketonuria)

Bethan Clinton, Bristol Children's Hospital
(A single centre review of the investigation and
management of children with Idiopathic Ketotic
Hypoglycaemia)

Ann Bowron, Newcastle Upon Tyne Hospitals NHS Trust
(Do blood samples for ammonia analysis require
transport on ice? A stability study in samples from
healthy volunteers)

Nicola Condon, Queen Elizabeth Hospital Birmingham
(Barriers and facilitators to Physical activity and exercise
in Late Onset Pompe Disease (LOPD): A qualitative
study exploring patients' perspectives and experiences)

EVENING

17:15-18:15

BIMDG AGM - Members only

19:15-22:30

SYMPOSIUM DINNER

Day 2: Thursday 8th June 2023

MORNING

- 07:00-07:45 Early morning activity - Brompton Cemetery Walk
- 08:30-09:00 **REGISTRATION, COFFEE, EXHIBITION STANDS, POSTERS**
- SESSION 3**
- LSD**
CHAIR: Derralynn Hughes/ RFH
- 09:00-09:30 New approaches to assessment of neuronopathic Gaucher disease –
Dr Aimee Donald
- 09:30-10:00 Monitoring cardiac disease in Fabry -Prof Richard Steeds
- 10:00-10:30 Measuring the benefits of new enzymes for Pompe- Dr Mark Roberts
- 10:30-11:00 Gene therapy for lysosomal disorders – Prof Paul Gissen
- 09:00-11:00** ***Parallel Dietetic Session***
- 11:00-11:30 **COFFEE BREAK , EXHIBITION STANDS, POSTERS**
- 11:30-13:00 **Members Papers**
Delegate's oral presentations (8-10 minutes each including 2-4 mins for questions)
- 11:30-11:40 Rohit Hirachan, GOSH
(Evaluation, in an NHS laboratory, of a Digital Microfluidics Platform for Rapid Assessment of Lysosomal Enzyme Activity)
- 11:40-11:50 Christine Newman, National Hospital of Neurology & Neurosurgery London
(The use of continuous glucose monitoring in inherited metabolic diseases with hypoglycaemia – a real world review)
- 11:50-12:00 Nadia Turton, Liverpool James Moores University
(Evidence of Mitochondrial dysfunction and oxidative stress in a neuronal cell model of Phenylketonuria)
- 12:00-12:10 Clare Dale, University Hospital Birmingham
(Do all females with hyperphenylalaninaemia require lifelong follow up and monitoring during pregnancy? A retrospective case series on Phe level monitoring and dietary advice during pregnancy at University Hospitals Birmingham and Guys and St Thomas')
- 12:10-12:20 Rachel Carling, GSTT London
(Comparison of plasma phenylalanine in with volumetrically collected dried blood spot phenylalanine: Implications for monitoring patients with phenylketonuria)

12:20-12:30	Nathan Cantley, Bristol Royal Hospital NHS Trust (The detection of Classical Galactosaemia via the UK PKU newborn screening program 2010-2020: a cross- sectional survey and national audit)
12:30-12:50	<u>Summer studentship presentations</u> (usually 1-2, 8-10 minutes each with 2-4 mins for questions)
13:00-14:00	LUNCH AND EXHIBITION STANDS/POSTERS
AFTERNOON	
SESSION 4	Paediatric Inherited Metabolic Disorders Part B. CHAIR: Spyros Batzios/GOSH
14:00-14:30	Advances in the understanding and treatment of metabolic liver disease – Prof Richard Thompson
14:30-15:00	Advances in management of Pyridoxine dependent epilepsy – Marjorie Dixon and Dr Emma Footitt
15:00-15:30	Advances in argininosuccinic aciduria – Dr Sonam Gurung
CHAIR:	James Davison, Elaine Murphy & Derralynn Hughes
15:30-16:00	Presentation of awards and CLOSE

***PROGRAMME IS SUBJECT TO CHANGE**