



The All Wales Inherited  
Metabolic Disease Service

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## **THE ALL WALES INHERITED METABOLIC DISEASE STUDY DAY**

**Date : Wednesday 6 March 2019**

**Venue : Mercure Cardiff Holland Hotel, 24-26 Newport Road,  
Cardiff, CF24 0DD**

**CPD accreditation applied for**

**Target audience: Doctors, nurses, medical trainees, dieticians and biochemists  
with an interest in metabolic disease as well as acute care  
physicians, endocrinologists and paediatricians who wish to update their  
knowledge on acute general metabolic and storage disorders.**

**Registration cost : £25.00 to include all refreshments and lunch.**

**This meeting will be an independent meeting with exhibition stands  
and support from the Pharmaceutical Industry.**

**For further information and a registration form please contact Jacqui McAleer,  
JM Associates, email : [jmassociates1@me.com](mailto:jmassociates1@me.com)**

# PROGRAMME

## 0900 – 0930 REGISTRATION, COFFEE & EXHIBITION STANDS

### MORNING SESSION :

CHAIR : Mike Badminton, Honorary Consultant in Chemical Pathology, Clinical Lead, Cardiff Porphyria Centre, Cardiff

## 0930 – 0945 Introduction to Welsh Paediatric Service

Graham Shortland, Consultant Paediatrician & Clinical Lead for All Wales Paediatric Inherited Metabolic Disease Service

## 0945 – 1035 Approach to the child with metabolic encephalopathy

Graham Shortland, Consultant Paediatrician & Clinical Lead for All Wales Paediatric Inherited Metabolic Disease Service &  
Kath Singleton, Lead Metabolic Dietitian in Wales

## 1035 – 1105 Update on All Wales extended Newborn Screening Program

Stuart Moat, Consultant Clinical Scientist, Director of Newborn Screening Wales

## 1105 – 1125 COFFEE BREAK

CHAIR : Graham Shortland, Consultant Paediatrician & Clinical Lead for All Wales Paediatric Inherited Metabolic Disease Service

## 1125 – 1205 Suspecting, identifying and diagnosing storage disease in children

James Davison, Consultant in Paediatric Metabolic Medicine, Great Ormond Street Hospital NHS Foundation Trust, London

## 1205 – 1250 Keynote Speaker : Practical clinical approach to investigating mitochondrial disorders

Simon Heales, Professor of Clinical Chemistry, Head of Clinical Service, Department of Paediatric Laboratory Medicine, Great Ormond Street Hospital NHS Foundation Trust, London

## 1250 – 1350 LUNCH

### AFTERNOON SESSION :

CHAIR : Duncan Cole, Honorary Consultant in Chemical Pathology and Metabolic Medicine, Clinical Lead All Wales Adult Inherited Metabolic Disease Service

## 1350 – 1400 Introduction to Adult IMD service in Wales

Duncan Cole, Honorary Consultant in Chemical Pathology and Metabolic Medicine, Clinical Lead All Wales Adult Inherited Metabolic Disease Service

## 1400 – 1440 The acute management of the severely ill adult with a metabolic condition

Tarek Hiwot, Consultant in Metabolic Disorders & Honorary Reader in the Institute of Metabolism & Systems Research, University of Birmingham

## 1440 – 1500 TEA BREAK

CHAIR : Danja Schulenburg-Brand, Consultant Chemical Pathologist, Medical Biochemistry and Immunology, University Hospital of Wales, Cardiff

## 1500 – 1540 Not just another heart failure/kidney failure – storage conditions in adults

Duncan Cole, Honorary Consultant in Chemical Pathology and Metabolic Medicine, Clinical Lead All Wales Adult Inherited Metabolic Disease Service

## 1540 – 1610 The ABC of managing acute porphyria for the non-porphyria expert

Mike Badminton, Honorary Consultant in Chemical Pathology, Clinical Lead Cardiff Porphyria Centre

## 1610 SUMMARY & CLOSE OF MEETING