

Annual SOUTH WEST REGIONAL METABOLIC STUDY DAY

THURSDAY 15TH MAY 2014

HOLIDAY INN, TAUNTON

This course aims to continue promoting the awareness and recognition of inherited metabolic disease and to increase the knowledge base about diagnostic pathways, management and the multidisciplinary approach needed for these patients. The day is aimed at clinicians who may be involved in the care of affected children and their families and is of particular relevance to Community Paediatricians, Neurologists, Geneticists, Community nurses, Junior Doctors, Biochemists & Dietitians. The programme will end with an interactive workshop.

Programme

09.00 -09.45	Registration and Coffee
09.45-09.50	Welcome Dr Germaine Pierre, Consultant Metabolic Paediatrician, University Hospitals Bristol NHS Foundation Trust
09.50-10.30	Investigations and management of Hypoglycaemia Dr Effie Chronopoulou, Consultant Metabolic Paediatrician, University Hospitals Bristol NHS Foundation Trust
10.30-11.15	Medium Chain Acyl CoA dehydrogenase Deficiency and Updates on expanded newborn screening Dr Helena Kemp, Director of Newborn Screening Mrs Hilary Sawyer, Principal Clinical Scientist North Bristol NHS Trust
11.15-11.30	Coffee
11.30-12.15	Homocystinurias Dr Germaine Pierre
12.15-13.00	Liver presentations in metabolic disorders Dr Roshni Vara, Consultant Metabolic Paediatrician, Evelina London Children's Hospital
13.00-14.00	Lunch
14.00-15.00	Investigation of the acutely ill neonate and drugs used for metabolic emergencies Dr Germaine Pierre Ms Ceri Manson, Metabolic Pharmacist University Hospitals Bristol NHS Foundation Trust
15.00-15.15	Coffee
15.15-16.30	Interactive workshops on interesting cases (MCADD / galactosaemia / homocystinuria) Workshop1: Diana Webster /Camille Jankowski ; Specialist Metabolic Dietitians Workshop 2:Anny Brown ; Clinical Scientist Workshop 3: Ann Bowron ; Clinical Scientist
16.30	Close and Thanks