

PROGRAMME

2021 VIRTUAL CONFERENCE – THURSDAY 24TH JUNE **10.00AM – 5.00PM**

Royal College of Pathologists (RCPATH) CPD accreditation approved
for 5 CPD credits

IMPROVING DIAGNOSIS AND TREATMENT

10.00-10.05 INTRODUCTION

DIETETIC SESSION

Chair : Alison Woodall, Metabolic Dietician, Salford Royal NHS Foundation Trust, Salford

10.05-10.35 **Wolman- immunological and dietetic aspects of management**
Fiona White, Metabolic Dietician and Arunabha Ghosh, Consultant Paediatrician, Willink Unit, Manchester

10.35-11.05 **GA1 results of newborn screening and UK dietary consensus**
Marjorie Dixon, Clinical Lead Dietitian – metabolic medicine, GOSH, London & Mildrid Yeo, Consultant in Paediatric IEM, GOSH, London

11.05-11.35 **The management of complex nutritional needs in MNGIE**
Antje Teubner Associate Specialist, Intestinal Failure Unit, Salford

11.35-11.50 BREAK

ADULT SESSION

Co-Chair : Gisela Wilcox, IMD Consultant, Salford Royal NHS Foundation Trust, Salford & Radha Ramachandran, IMD Consultant, Guy's & St Thomas' Hospital, London

Chosen from abstracts submitted :

11.50-12.05 **Developmental consequences of defective ATG7-mediated autophagy in humans**
Professor Rob Taylor, Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle upon Tyne

12.05-12.20 **A case study of a patient with Aicardi-Goutières syndrome presenting as a possible Peroxisomal disorder**
Corey Pritchard, University Hospitals Bristol & Weston

12.20-12.50 **Late-onset MTHFR- a novel disease?**
Fanny Mochel, Consultant Neurologist, Reference Centre for Neurometabolic Disease, Paris

12.50-13.50 LUNCH

13.50-15.20 MEMBER'S ORAL PAPERS CHOSEN FROM ABSTRACTS

Chair : Radha Ramachandran, IMD Consultant, Guy's & St Thomas' Hospital, London

13.50-14.05 **Social, emotional & behavioural outcomes in children born to women with PKU & their relationship with maternal metabolic control**
Charlotte Ellerton, UCLH, London

14.06-14.20 **The use of glycerol phenylbutyrate in adult patients with Urea Cycle Disorders - one centre experience**
Faiza Adrees, Salford Royal NHS Foundation Trust, Salford

14.21-14.35 **Sitosterolaemia effectively managed with dietary modification + Ezetimibe**
Katie Rawlins, GSTT, London

14.36-14.50 **Results of general newborn screening for Isovaleric Acidaemia in the North-West of England**
Bernd Schwahn, Willink Unit, Manchester

14.51–15.05 **Adult-onset generalised dystonia as the main phenotypic feature of alpha-methylacyl-CoA racemase deficiency**
Francesca Magrinelli, UCLH, London

15.06-15.20 **SRD5A3-CDG: Emerging phenotypic features apart from the eye involvement**
Nazreen Banu Kamarus Jaman, GOSH, London

15.20-15.35 Student summer projects summary from 2019 awards :

An in vitro proof of principle study to test the efficacy of translational read-through therapy for mitochondrial disorders
Melissa Kuo, now a 5th year Medical Student at UCL London

Natural protein and phenylalanine tolerance and metabolic control in patients with hereditary tyrosinaemia type I (HTI).
Ozlem Yilmaz, now a PhD Dietetic Student in Turkey

15.35-15.50 BREAK

SCIENTIST SESSION

Co-Chair : Teresa Wu, & Heather Church, Willink Biochemical Genetics Unit, Manchester University NHS Foundation Trust

15.50-16.20 **Laboratory development of clinically useful antibody monitoring in Enzyme Replacement Therapy**
Katherine Brammeier, Senior Biomedical Scientist/Simon Jones, Consultant Paediatrician, Willink Unit, Manchester

16.20-16.50 **How to improve IMD monitoring with patient collected samples during COVID-19 pandemic**
Helena Kemp, Chemical Pathology Consultant, Bristol

16.50-17.00 BREAK

17.00 AGM – Members only