Rare Metabolic Disorders: Detection, Research, Management and Treatment (London, UK - 20th-22nd September 2016)

Description:
This conference will discuss rare metabolic disorders, their detection, current research, disease management and treatment.

From congenital disorders to inherited metabolic diseases, this event will investigate recent developments in an informal academic setting, with an atmosphere conducive to debate and discussion.

With a number of sessions over three days, many aspects of the metabolome will be covered, bringing together those working in academia, medicine, biotechnology and pharmaceuticals.

Talks Include:

- Mobile application for balance training, in people with familial dysautonomia: three case reports:
  Rosalee Gefen, The Zinman College of Physical Education & Sport Sciences, Wingate Institute, Netanya, Israel

- Recent advances in Familial Dysautonomia:
  Professor Horacio Kaufmann, MD FAAN, Medicine and Pediatrics. Axelrod Chair for Dysautonomia Research, New York University School of Medicine, NY, USA

- Enzyme replacement therapy for lysosomal storage disorders: the pharmacology of marginal gains
  Dr Robin Lachmann, National Hospital for Neurology and Neurosurgery, London, United Kingdom

- Mitochondrial Biogenesis in the Pediatric Cardiomyopathy called Barth Syndrome: A Search for Novel Drug Targets
  Dr. Ashim Malhotra, Pacific University Oregon, USA, School of Pharmacy, College of Health Professions, Hillsboro, United States

- Managing Familial Dysautonomia in a North London DGH: A paediatrician's steep learning curve
  Dr Su Laurent, Royal Free London NHS Foundation Trust, Barnet Hospital, Barnet, Hertfordshire, United Kingdom

- Alkaptonuria - Metabolic response to treatment with Nitisinone.
  Dr Anna M Milan, Royal Liverpool and Broadgreen University Hospitals, Liverpool, United Kingdom

- Talk title from Professor Chani Maayan, Hadassah Medical Center, Kiryat Hadassah, Jerusalem, Israel to be confirmed

- Talk title from Ana Amado Fondo, Metabolic CNS, Charles Dent Metabolic Unit, London, United Kingdom to be confirmed

- Autonomic Dysfunction in Familial Dysautonomia
  Dr Ellen Merete Hagen, Consultant Neurologist, Autonomic Unit, National Hospital for Neurology & Neurosurgery, London, United Kingdom

- The implications of the emerging insights in metabolic and genomic science for patients and primary care
  Dr Paul Michael Knapton, British Heart Foundation, London, United Kingdom

- DevelopAKUre: a patient-led clinical trial for a rare disease
  Oliver Timmis, AKU Society, Cambridge, United Kingdom

- The role of Open Innovation in development of new therapies for rare diseases
  Dr Martino Picardo, Stevenage Bioscience Catalyst, Stevenage, Herts, United Kingdom

- The impact of genomics on rare disease research
Dr Jonathan Milner, Deputy Chairman, Abcam plc, Cambridge, United Kingdom
- Biochemical insights into Primary hyperoxaluria type III.
Dr Kerry Loomes, School of Biological Sciences, University of Auckland, Auckland, New Zealand
- Steroid profiling for rare diseases
Dr Bruno Vogt, Inselspital, University Hospital of Bern, Bern, Switzerland
- Plasma miRNA signatures for following the neoplastic process in hereditary tyrosinemia
Professor Robert M. Tanguay, Université Laval, Québec, Canada
- B12 deficiency and Gestational diabetes - an update
Dr Vimal Karani S, University of Reading, Food & Nutritional Sciences, School of Chemistry Food & Pharmacy, Reading, United Kingdom
- GRK2 as a new integrative node in obesity and insulin resistance: multi organ effects
Dr Cristina Murga, Centro de Biología Molecular Severo Ochoa (CSIC-UAM), Madrid, Spain
- Biliary transporter mutations; implications for gestational liver disease
Professor Catherine Williamson, King's College London, London, United Kingdom
- Gene and Cell therapy approaches for Rare Diseases
Dr Takis Athanasopoulos, University of London, Egham, United Kingdom
- Role and regulation of the RNA-binding protein Bicc1 in cystic kidney diseases
Professor Daniel Constam, Ecole Polytechnique Fédérale de Lausanne, Lausanne, Switzerland
- Computational approaches to targeted drug design in metabolic diseases
Dr Adina Milac, Biochemistry of the Romanian Academy, București, Romania
- The molecular basis of childhood-onset mitochondrial diseases’ for ~30 minutes
Professor Shamima Rahman, UCL Institute of Child Health, London, United Kingdom
- Talk title from Associate Professor Christina Bark, Karolinska Institutet, Stockholm, Sweden to be confirmed
- Next Generation Sequencing (NGS) approach to discovery of rare neuro-metabolic disorders
Dr. Maja Tarailo-Graovac, Centre for Molecular Medicine and Therapeutics, Vancouver, Canada
- Clinical gait analysis in Alkaptonuria
Dr. Gabor Barton, Liverpool John Moores University, Liverpool, United Kingdom
- Innovative treatment of the porphyrias
Dr. Elisabeth Minder, Stadtspital Triemli - Stadt Zürich, Zürich, Switzerland
- Diagnosis and Discovery of Treatable Neurometabolic Diseases
Dr. Van Karnebeek, University of British Columbia, Vancouver, Canada
- Talk title to be confirmed
Dr. Germaine Pierre, UHBT, Education Centre, Bristol, United Kingdom
- Cystic fibrosis: molecular aspects of disease, current strategies for therapy
Associate Professor Valerie Chappe, Dalhousie University, Nova Scotia, Canada

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