

CDT 2023 - PROGRAM AT A GLANCE - (Lectures / Practical Sessions)

| Sunday – July 30 | | Monday – July 31 | Tuesday – August 1 | Wednesday – August 2 | Thursday – August 3 |
|--|-------------|--|---|---|---|
| | 8:30-10:00 | <p><i>Inborn Errors or Metabolism: General Overview</i> R Giugliani</p> <p><i>Clinical approach of patients with suspected metabolic diseases</i> D Weinstein</p> | <p><i>Treatment of Intermediate Metabolism Diseases - Overview</i> C Fischinger</p> <p>Case Report 3 - F Poswar & CDT team</p> | <p>Practical Session I <i>Interview with patients to discuss diagnosis and management.</i></p> <p>C Fischinger & CDT Faculty</p> | <p><i>The IEM in the Era of Genomic Medicine</i> F Kok</p> <p><i>Neonatal Screening for IEM</i> I Schwartz (TBC)</p> |
| Coffee break (10:00-10:30) | | | | | |
| | 10:30-12:00 | <p><i>Clinical Approach to the Diagnosis Intermediate Metabolism Diseases</i> C Fischinger</p> <p><i>Clinical Approach to the Diagnosis of Lysosomal Diseases</i> F Lagler</p> | <p><i>Treatment of Lysosomal Storage Diseases - Overview</i> F Poswar</p> <p>Case Report 4 - F Poswar & CDT team</p> | <p>Practical Session II – part A <i>Laboratory Diagnosis of Lysosomal Disorders</i></p> <p>Practical Session II – part B <i>Laboratory Diagnosis of Intermediate Metabolism Disorders</i></p> | <p><i>Novel Therapies for the IEM</i> R Giugliani</p> <p><i>Gene Therapy Highlights</i> D Weinstein</p> <p><i>Evaluation & Closing Remarks</i> R Giugliani</p> |
| Lunch (12:00-14:00) | | | | | |
| | 14:00-15:30 | <p>Case Report 1 – Presentation <i>Laboratory Diagnosis of IEM Part I – Amino acids, Organic Acidurias, FAOD</i> A Sitta & C Vargas (TBC)</p> <p>Case Report 1 – Discussion</p> <p>Case Report 2 – Presentation <i>Laboratory Diagnosis of IEM Part II – Lysosomal Diseases</i> K Michelin-Tirelli & L Faqueti (TBC)</p> <p>Case Report 2 – Discussion</p> | <p><i>Hypoglycemia and IEMs</i> D Weinstein</p> <p><i>Mitochondrial Diseases</i> F Lagler</p> | <p><i>Metabolic Emergencies</i> F Lagler</p> <p>Case Report 6 – F Poswar & CDT team</p> <p><i>Seizures and Movement Disorders in IEM</i> F Kok</p> | |
| Coffee break (15:30-16:00) | | | | | |
| Arrivals | 16:00-17:30 | <p><i>Mucopolysaccharidoses, Mucopolidoses and Glycoproteinoses</i> R Giugliani</p> <p><i>Porphyrias</i> T Oliveira Silva</p> <p><i>Congenital Disorders of Glycosilation</i> C Lourenço</p> | <p><i>Gaucher, Pompe, Fabry and ASMD: what did we learn so far?</i> C Fischinger & F Poswar</p> <p>Case Report 5 – F Poswar & CDT team</p> <p><i>Genetic Counseling and Prenatal Diagnosis</i> M Sanseverino (TBC)</p> | <p><i>Peroxisomal disorders and White Matter Diseases</i> F Kok</p> <p><i>Neuroimaging in selected IEMs.</i> J Duarte (TBC)</p> <p><i>CLNs, NPC and other selected neurodegenerative metabolic diseases</i> C Lourenço</p> | |
| 18:30-22:00 Introduction R Giugliani Opening remarks from the international invited speakers Welcome Dinner | | Free Evening | Free Evening | Official Dinner | |