

## CDT 2025 – PRELIMINARY PROGRAM AT A GLANCE - (Lectures / Practical Sessions )

Sunday – August 3		Monday – August 4	Tuesday – August 5	Wednesday – August 6	Thursday – August 7	Friday – August 8
	8:30-10:00	<i>Inborn Errors or Metabolism: General Overview</i> <b>R Giugliani</b>  <i>Clinical approach to patients with suspected metabolic diseases</i> <b>C Fischinger</b>	<i>Treatment of Lysosomal Diseases</i> <b>F Ezgu</b>  <b>Case Report 4 – B Oliveira</b>	<b>VISIT TO CASA DOS RAROS</b>  <b>Practical Session I</b> <i>Interview with patients to discuss diagnosis and management</i> <b>C Fischinger &amp; CDT Faculty</b>	<i>Mitochondrial Diseases</i> <b>J Hennermann</b>  <i>Metabolic Diagnosis based on molecular genetics investigation.</i> <b>B Oliveira</b>	<i>Novel Therapies for the IEM</i> <b>J Hennermann</b>  <i>Gene Therapy Highlights</i> <b>F Ezgu</b>
Coffee break (10:00-10:30)						
	10:30-12:00	<i>Clinical Manifestations of Intermediate Metabolism Diseases</i> <b>C. Fischinger</b>  <i>Clinical Manifestations of Lysosomal Diseases</i> <b>F Ezgu</b>	<i>MPS and related diseases</i> <b>R Giugliani</b>  <i>Gaucher, Pompe, Fabry and ASMD: what did we learn so far?</i> <b>F Poswar</b>  <b>Case Report 5 – R Stefani</b>	<b>Practical Session II</b> <i>Interview with patients to discuss diagnosis and management</i> <b>C Fischinger &amp; CDT Faculty</b>	<i>Translational Medicine: From bench to clinics</i> <b>G Baldo</b>  <i>Genetic Counseling and Prenatal Diagnosis</i> <b>M Sanseverino</b>	<i>The IEM in the Era of Genomic Medicine</i> <b>F Vairo</b> <i>Neonatal Screening for IEM</i> <b>C Fischinger</b> <i>Evaluation &amp; Closing Remarks</i> <b>R Giugliani</b>
Lunch (12:00-14:00)						
	14:00-15:30	<i>Laboratory Diagnosis of IEM Part I – Amino acids, Organic Acidurias, FAOD</i> <b>CR Varga &amp; A.Sitta</b> <b>Case Report 1 – R Stefani</b>  <i>Laboratory Diagnosis of IEM Part II – Lysosomal Diseases</i> <b>K Michelin-Tirelli</b> <b>Case Report 2 – F Poswar</b>	<i>Peroxisomal diseases and White Matter Diseases</i> <b>F Kok</b>  <i>Neuroimaging in selected IEMs.</i> <b>J Duarte (TBC)</b>  <i>Seizures and Movement Disorders in IEM</i> <b>F Kok</b>	<b>Practical Session III</b> <i>Laboratory Diagnosis of Lysosomal Disorders</i> <b>M Riegel &amp; CDT Faculty</b>	<b>Case Reports presented by students</b>	<b>Departures</b>
Coffee break (15:30-16:00)						
Arrivals	16:00-17:30	<i>Hypoglicemia and IEMs</i> <b>C Fischinger</b>  <b>Case Report 3 – C Fischinger</b>   <i>Congenital Disorders of Glycosylation</i> <b>C Lourenço</b>	<i>Metabolic Dementias and other selected neurodegenerative diseases</i> <b>C Lourenço</b>  <i>Treatment of Intermediate Metabolism Disorders</i> <b>J Hennermann</b>	<b>Practical Session III</b> <i>Laboratory Diagnosis of Intermediate Metabolism Disorders</i> <b>M Riegel &amp; CDT Faculty</b>	<b>Case Reports presented by students (cont.)</b>	
<b>18:30-22:00</b> Introduction Opening remarks from the speakers Welcome Dinner		Free Evening	Free evening	Free Evening	Official Dinner	