

08.20 On-site inscriptions & Welcome coffee

08.50 Opening of the meeting & Presentation of the new website  
*MF Vincent (Cliniques universitaires Saint-Luc), F Eyskens (President of Metabolics.be) & N Vanhoutvin (Metabolics.be)*

## **SESSION 1 Chairs: H Stepman (UZ Gent) & D. Rymen (UZ Leuven)**

*Hof Ter Musschen main room (9.00 -10.50)*

09.00 Disorders of purine and pyrimidine metabolism  
*J Dewulf (Cliniques universitaires Saint-Luc)*

09.30 Q & A

09.40 Adenosine deaminase deficiency type 1 and type 2  
*I Meyts (UZ Leuven)*

10.10 Q & A

10.20 Targeted urine metabolomics for the diagnosis of inborn errors of metabolism  
*L Steinbusch & I. Keularts (Maastricht University Medical Center, The Netherlands)*

10.50 Q & A

11.00 Coffee break : *Luscinia & Fringilla rooms*

## **SESSION 2 Chairs: D. Roland (IPG) & F Boemer (CHU de Liege)**

*Hof Ter Musschen main room (11.30 – 12.55)*

11.30 Fluoropyridine toxicity and dihydropyrimidine dehydrogenase deficiency: clinical recommendations.  
*V Haufroid (Cliniques universitaires Saint-Luc)*

12.00 Q & A

12.10 Selected free communications: session 1

- Sebelipase alfa ERT in Wolman disease: up to ten years follow-up.  
*T Demaret (IPG)*
- Selective screening for Gaucher disease in patients presenting with splenomegaly with or without thrombocytopenia  
*F Eyskens (UZ Antwerpen)*
- Long-term, sustained efficacy and safety from a phase 1-2 clinical trial of an AAV8-mediated liver-directed gene therapy in adults with glycogen storage disease type Ia  
*TG Derks (University of Groningen, The Netherlands & Ultragenyx Pharmaceutical)*

## **SESSION 2 Joint meeting for dieticians and psychologists**

*Hirunda room (11.30 – 12.55)*

- 11.30 Therapeutic education for caregivers of a sick or a disabled child: an emerging response to their respite needs, to deal with the risk of parental burnout and its consequences on children.  
*AC Dubois (UCLouvain) & I Aujoulat (UCLouvain)*

12.55 Lunch break: Hof Ter Musschen, Luscinia & Fringilla rooms

13.45 General assembly of Metabolics.be (restricted to members)

## **SESSION 3 Chairs: L Marcelis (Centre de dépistage néonatal ULB) & F Eyskens (UZ Antwerpen)**

*Hof Ter Musschen main room (14:15 - 16:50)*

- 14.15 Selected free communications: Session 2
- Follow-up of executive functioning in patients with an inherited metabolic disease of intoxication type.  
*E Raets (UZ Antwerpen)*
  - BCKD kinase deficiency: an essential aminoacid deficiency syndrome, diagnostic and therapeutic challenges.  
*F-G Debray (CHU de Liege)*
  - A case of neonatal glutaric aciduria type 2 in a context of maternal deficiency.  
*A Empain (HUDERF)*
- 15.00 The future of Newborn Screening  
*G Gramer (University Medical Center Hamburg-Eppendorf, Germany)*
- 15.30 Q & A
- 15.40 Mitochondrial disease, mitophagy, and cellular distress in methylmalonic acidemia  
*A. Luciani (University of Zurich, Switzerland)*
- 16.10 Q & A
- 16.20 Breaking news from ICIEM 2021 and SSIEM 2021 meetings  
*MC Nassogne (Cliniques universitaires Saint-Luc)*
- 16.40 Farewell : *MF Vincent (Cliniques universitaires Saint-Luc)*