

# INBORN ERRORS OF METABOLISM: A PERMANENT CHALLENGE

## 24 November 2006

- 14:15 **Opening**  
Chairpersons: I. Tavares de Almeida (CPM / FF-UL, Lisboa),  
F Eusebio (HSM, Lisboa)
- IEM from Suspicion to the Diagnosis**  
A Cabral (Chairman of SPDM)
- 14:30 **The Suspicion of IEM in the neonate**  
A Cabral (SPDM, Lisboa)
- 14:55 **Late-onset forms of IEM**  
H Ogier de Baulny (Hop Robert Debre, Paris, France)
- 15:20 **Biomarkers: from the detection to the diagnosis**  
M Duran (AMC; Univ. Amsterdam, The Netherlands)
- 15:45 **The role of molecular genetics in the diagnosis**  
L Rivera (CPM / FFUL, Lisboa)
- 16:05 **Discussion - Painel**
- 16:25 **Coffee Break and Poster Viewing**
- 17:00 **Free Communications**  
Chairpersons: S. Sequeira (HDE, Lisboa), P Garcia (I-IP, Cdmbr)
- 18:30 **End of the Session**
- 19:45 **Dinner**

## 25 November 2010

- IEM Treatment**  
Chairpersons: L Diogo (HP, Coimbra), A Cabral (SPDM, Lisboa)
- 09:00 **Emergency therapeutic approaches**  
S. Sequeira (HDE, Usboa)
- 09:20 **Supportive treatment: energy metabolism disorders**  
E Marlins (HMPia, Porto)
- 09:40 **Supportive treatment: intoxication type disorders**  
F Eusebio (HSM, Lisboa)
- 10:00 **Discussion - Painel**
- 10:20 **Coffee Break and Poster Viewing**
- Lysosomal Storage Disorders**  
Chairpersons: MC Sá Miranda (IBMC, Poilo), P Garcia (HP, Coimbra)
- 11:00 **Clinical, biochemical, genetic and therapeutic aspects: an overview**  
M Patterson (Columbia Univ. Medical Center, NY, USA)
- 11:30 **Enzyme replacement therapy: advantages and pitfalls**  
E Leao Teles (HSJoao, Porto)
- 11:50 **Gaucher Disease: substrate reduction therapy**  
P Giraldo (Hosp Univ Miguel Servet, Zaragoza, Spain)
- 12:10 **Experience on therapy of Niemann-Pick type C with Miglustat**  
M Pineda (Hop Sant Joan de Deu, Barcelona, Spain)
- 12:30 **Early diagnosis: new perspectives**  
M C Sa Miranda (IBMC, Porto)
- 12:50 **Discussion-Painel**
- 13:15 **Lunch**
- Other IEM**  
Chairpersons: M Duran (AMC, Amsterdam), H Araujo (UMa, Madeira)
- 14:45 **Dyslipidemias**  
A Gaspar (HSM, Lisboa)
- 15:05 **Homocystine: a risk factor?**  
I Tavares de Almeida (CPM / FFUL, Lisboa)
- 15:25 **Creatine deficiency disorders**  
C Jacobs (VU, Amsterdam, The Netherlands)
- 15:50 **Discussion - Painel**
- 16:10 **Coffee Break**
- 16:30 **Free Communications**  
Chairpersons: E Leao Teles (HSJoao, Porto), I Rivera (CPM, FFUL)
- 18:00 **Free Communications and Poster Awards**  
Closing Remarks
- 18:15 **End of the Symposium**