**SS3 - PRE-CONGRESS SATELLITE SYMPOSIUM 3 - HOMOCYSTINURIA**

**Contact Person:** Tara Morrison (Sydney, Australia)

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**08:00 – 18:00**

**08:00**
Registration open

**08:45**

**JOINT STREAM**

**Chair:** I Schwartz (Porto Alegre, Brazil)

- **08:45 – 08:50**  Welcome (H Blom, Freiburg, Germany, I Schwartz, Porto Alegre, Brazil, T Morrison, Sydney, Australia)
- **08:50 – 09:00**  Introduction to Homocystinuria and EHOD (H Blom, Freiburg, Germany)
- **09:00 – 09:20**  Quality of life in Homocystinuria patients (I Schwartz, Porto Alegre, Brazil)
- **09:20 – 09:40**  Guidelines for the diagnosis and management of CBS deficiency (A Morris, Manchester, UK)
- **09:40 – 10:00**  Guidelines for diagnosis and management of the cobalamin related remethylation disorders and MTHFR deficiency (M Baugartner, Zurich, Switzerland)
- **10:00 – 10:20**  Qatar: a unique experience (T Ben-Omran, Doha, Qatar)

**10:20 – 10:40**
Coffee-Break

**10:40 – 11:00**
An overview of how our understanding has developed and current knowledge for CBS deficiency (B Wilcken, Sydney, Australia)

**11:00 – 11:20**
Looking forward: Enzyme replacement therapy for CBS deficiency (J Kraus, Denver, USA)

**11:20 – 11:40**
Strategies to repair Mutant CBS Proteins (W Kruger, Philadelphia, USA)

**11:40 – 12:00**
Research strategy, priorities and aims: a patient organisation approach (M McGlynn, Flourtown, USA)

**12:00 – 13:30**
Interactive lunch

**13:30 – 14:30**

**CBS deficiency Panel Discussion: Q & A**

**Chair:** K Bhattacharya (Sydney, Australia)

**Faculty:**
- B Wilcken (Sydney, Australia)
- I Schwartz (Porto Alegre, Brazil)
- V Kozich (Prague, Czech Republic)
- A Morris (Manchester, UK)
- Patient advocate TBC

**14:30 – 15:00**

**CblC and MTHFR Panel Discussion: Q & A**

**Chair:** K Bhattacharya (Sydney, Australia)

**Faculty:**
- M Baugartner (Zurich, Switzerland)
- C Dionisi-Vici (Rome, Italy)
- Patient Advocate TBC

**15:00 – 15:30**
Coffee-Break

**Scientific Stream**

**Chair:** H Blom (Freiburg, Germany)

**15:30 – 15:50**
Hydrogen sulfide metabolism in homocystinurias (V Kozich, Prague, Czech Republic)

**15:50 – 16:10**
Animal models of combined methylmalonic acidemia and homocystinemia cblC type: insights into disease pathophysiology and development of new therapies (J Sloan, Bethesda, USA)

**16:10 – 16:30**
Mouse models of CBS deficiency: What have we learned? (W Kruger, Philadelphia, USA)

**16:30 – 16:50**
Homocysteine and Psychiatric Disorders (V Almeida, São Paulo, Brazil)

**16:50 – 17:10**
First results CblC disease from EHOD registry (C Dionisi-Vici, Rome, Italy)

**17:10 – 17:30**
Preliminary data on CBS deficiency from EHOD registry (V Kozich, Prague, Czech Republic)

**17:30 – 17:50**
Consequences of the complex regulation of homocysteine metabolism (H Blom, Freiburg, Germany)

**17.50 -18.00**
Closing remarks
**Patient Stream**

**Chair:** Tara Morrison (*Sydney, Australia*)

15:30 – 16:10  Novel Patient Organisations: What is available and what is planned for the future
HCU Network Australia (*T Morrison, Sydney, Australia*)
HCU Network America (*M McGlynn, Pennsylvania, USA*)
Associação Brasileira de Homocistinuria (*S Aredé, Rio de Janeiro, Brazil*)
cblC Brasil (*L Brito, Salvador, Brazil*)

16:10 – 16:30  Patient care in Brazil: opportunities and challenges (*S Poloni, Porto Alegre, Brazil*)

16:30 – 16:40  Quiz (*B Santos and C Lobato, Porto Alegre, Brazil*)

16:40 – 17:00  Mental health and behaviour challenges and strategies for management (*R Massuda, Curitiba, Brazil*)

17:00 – 17:20  Dietary intervention in CBS deficiency (*B Frangipani, Sao Paulo, Brazil*)

17:20 – 17:40  Clinical management of cblC deficiency: experience from a US natural history study (*I Manoli, Bethesda, MD, USA*)

17:40 – 18:00  Importance of Natural History Studies and Registries in the homocystinurias (*H Levy, Boston, USA*)

18.00  Closing remarks