

## PRELIMINARY SCIENTIFIC PROGRAM (2017-07-02b)

### SEPTEMBER 3

#### **15:30 – 21:30 SS1 - PRE-CONGRESS SATELLITE SYMPOSIUM 1 - INFORM MEETING – PART A**

**Contact Person:** *Keith McIntire (Pittsburgh, PA, USA)*

**Contact E-mail:** *keith.mcintire@chp.edu*

**Website:** *www.informnetwork.org*

#### **15:30 – 18:15 Session 1 - Welcome and Keynote Speech**

15:30 – 16:30 Registration

16:30 – 16:40 Welcome and Overview to INFORM 2017 (*J Vockley, Pittsburgh PA, USA*)

16:40 – 16:45 Keynote Speaker Introduction

16:45 – 17:45 Keynote, The Impact of NBS of FAODs (*N Longo, Salt Lake City, Utah, USA*)

17:45 – 18:15 Discussion

#### **19:00 – 21:30 Networking Reception and Opening Dinner**

### SEPTEMBER 4

#### **07:30 – 18:45 SS1 - PRE-CONGRESS SATELLITE SYMPOSIUM 1 - INFORM MEETING – PART B**

#### **07:30 – 12:30 Session 2: Basic Science**

07:30 – 08:30 Registration / Breakfast

08:30 – 09:00 Myotubes from CPT2 Deficient Patients as a Disease Model  
(*F Djouadi, Paris, France*)

09:00 – 09:30 Free Fatty Acid Receptors and the Gut Microbiome: Questions in relation to FAO Defects  
(*N Gregersen, Aarhus Denmark*)

09:30 – 09:45 Abstract Presentation 1

09:45 – 10:00 Abstract Presentation 2

#### **10:00 – 10:30 Coffee-Break**

10:30 – 11:00 Nutritional Ketosis (*K Clarke, Oxford, UK*)

11:00 – 11:30 Lessons Learned from Animal Models of VLCAD Deficiency  
(*U Spiekerkoeter, Freiburg, Germany*)

11:30 – 11:45 Abstract Presentation 3

11:45 – 12:00 Abstract Presentation 4

12:00 – 12:30 Panel Discussion

#### **12:30 - 14:00 Lunch**

#### **14:00 – 18:45 Session 3: Clinical session**

14:00 – 14:30 Ketone Body Treatment in FAODs  
(*G Visser, Amsterdam, Netherlands*)

14:30 – 15:00 Metabolism, Epigenetics, and Signaling Integrate to Control Lipid Oxidation  
(*M Hirschey, Duke University, USA*)

15:00 – 15:15 Abstract Presentation 5

15:15 – 15:30 Abstract Presentation 6

#### **15:30 – 15:45 Coffee-Break**

15:45 – 16:15 An overview of FAODs and unmet needs in Latin America  
(*I Schwartz, Porto Alegre, Brazil*)

16:15 – 16:30 Abstract Presentation 7

16:30 – 16:45 Abstract Presentation 8

16:45 – 17:15 Panel Discussion

17:15 – 17:30 Summary / Closing remarks

17:30 – 18:45 Cocktail Reception & Poster Session  
with Oral Presentations for Jr. Investigators interested in presenting

#### **19:00 INFORM Advisory Committee Meeting**

**08:30 – 18:45 SS2 - PRE-CONGRESS SATELLITE SYMPOSIUM 2 - NEWBORN SCREENING**

**Contact Person:** *Gustavo Borrajo (La Plata, Argentina)*

**Contact E-mail:** *borrajog@net-alliance.net*

**Program**

**08:30 – 10:15 Session 1: Key Considerations for the Implementation of NBS by TMS – Part 1**

08:30 - 09:00 How to decide which diseases to include (*R Howell, Miami, FL, USA*)

09:00 - 09:30 How to select an appropriate measurement method and organize the MS/MS laboratory (*Kostas Petritis, Atlanta, GA, USA*)

09:30 - 10:00 How to approach efficient and effective result interpretation and decision making (*D Matern, Rochester, MN, USA*)

10:00 - 10:15 Q & A

**10:15 - 10:45 Coffee-Break**

**10:45 – 12:30 Session 2: Key Considerations for the Implementation of NBS by TMS – Part 2**

10:45 - 11:15 How to reduce false positives using second tier testing (*V Wiley, Sydney, NSW, Australia*)

11:15 - 11:45 Establishing the confirmatory laboratory (*C Rizzo, Rome, Italy*)

11:45 - 12:15 How to organize the treatment and follow-up of the detected newborns (*J Abdenur, Buenos Aires, Argentina/Orange County, CA, USA*)

12:15 - 12:30 Q & A

**12:30 - 14:00 Lunch-Break**

**14:00 – 15:45 Session 3: Accreditation and Quality of Results**

14:00 - 14:30 Measurement of uncertainty in NBS tests (*J Bonham, Sheffield, UK*)

14:30 - 15:00 CDC Involvement in Ensuring Quality (*J Mei, Atlanta, GA, USA*)

15:00 - 15:30 External Quality Assurance Program for Latin America (*G Borrajo, La Plata, Argentina*)

15:30 - 15:45 Q & A

**15:45 - 16:15 Coffee-Break**

**16:15 – 17:25 Session 4: Targeted Genomics in NBS**

16:15 - 16:45 The Role of Whole Genome Sequencing in Screening Neonates Today (*D Dimmock, San Diego, CA, USA*)

16:45 - 17:15 Next Generation Sequencing in Newborn Screening: Cystic Fibrosis and Beyond (*M Baker, Madison, WI, USA*)

17:15 - 17:25 Q & A

**17:25 – 18:35 Session 5: New Diseases – Further Expansion**

17:25 - 17:55 NBS for lysosomal disorders (*J Fletcher, Adelaide, SA, Australia*)

17:55 - 18:25 NBS for Duchenne Muscular Dystrophy (*R Howell, Miami, FL, USA*)

18:25 - 18:35 Q & A

**18:35 – 18:45 Closure** (*G Borrajo, La Plata, Argentina*)

**08:30 – 18:00 SS3 - PRE-CONGRESS SATELLITE SYMPOSIUM 3 - HOMOCYSTINURIA**

**Contact Person:** *Tara Morrison (Baulkham Hills, NSW, Australia)*

**Contact Email:** *tara@hcunetworkaustralia.org.au*

**08:30 Registration open**

**09:00 JOINT STREAM**

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

08:45 – 09:00 Welcome / Introduction to Homocystinuria and EOHD (*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, UK*)

09:40 – 10:00 Guidelines for diagnosis and management of the cobalamin related remethylation disorders and MTHFR deficiency (*M Baungartner, Zurich, Switzerland*)

10:00 – 10:20 Qatar: a unique experience (*Tawfeg Ben Omram, 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, USA*)
- 11:20 – 11:40 Strategies to repair Mutant CBS Proteins (*W Kruger, USA*)
- 12:00 – 12:20 Research strategy, priorities and aims: a patient organisation approach (*M McGlynn, USA*)

**12:20 – 13:50 Interactive lunch**

**13:50 – 15:10 CBS deficiency Panel Discussion: Q & A**

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

**Faculty:** *B Wilcken (Sydney, Australia)*

*Ida Schwartz (Porto Alegre, Brazil)*

*Victor Kozick (Prague, Czech Republic)*

*Andrew Morris (UK)*

*Patient advocate TBC*

**13:50 – 15:10 CBS deficiency Panel Discussion: Q & A**

**Chair:** *C Dionisi-Vici (Rome, Italy)*

**Faculty:** *M Baugartner (Zurich, Switzerland)*

*Patient Advocate TBC*

**15:10 – 15:30 Coffee-Break**

**Scientific Stream**

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

- 15:30 – 15:50 Newborn screening for homocystinuria and methylation disorders  
(*V Kozich, Prague, Czech Republic*)

- 16:10 – 16:30 Mouse models of CBS deficiency: What we have learned (*W Kruger, 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 H2S (*Victor Kozick, Prague, Czech Republic*)

- 17:30 – 17:50 TBA (*H Blom, Freiburg, Germany*)

**Patient Stream**

**Chair:** *Tara Morrison (Baulkham Hills, NSW, Australia)*

- 15:30 – 15:50 Novel Patient Organisations: What is available and what is planned for the future  
HCU Network Australia (*T Morrison, Baulkham Hills, NSW, Australia*)  
HCU Network America (*M McGlynn, USA*)  
Brazilian Organisation (*S Poloni, Brazil*)

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

- 16:30 – 16:50 Mental health and behaviour challenges and strategies for management (*R Massuda*)

- 16:50 – 17:10 Importance of Natural History Studies and Registries in the homocystinurias (*H Levy, USA*)

- 17:10 – 17:30 Dietary intervention in CBS deficiency (*B Frangipani, Brazil*)

- 17:30 – 17:50 Clinical management of cblC deficiency: experience from a US natural history study  
(*I Manoli, Bethesda, MD, USA*)

**14:00 – 17:30 SS4 - PRE-CONGRESS SATELLITE SYMPOSIUM 4 - NUTRITION & DIETETICS**

**Contact Person:** *Annemiek van Wegberg (Nijmegen, Netherlands)*

**Contact E-mail:** *Annemiek.vanWegberg@radboudumc.nl*

**14:00 – 15:30 - PARALLEL WORKSHOPS 1, 2, 3 and 4 – ROUND 1**

**WORKSHOP 1 – Dietary treatment in MSUD, OA, UCD**

*J Gribben (London, UK)*

**WORKSHOP 2 – Tools and Apps: What is out there and how does it affect our patients?**

*D Hook (Long Beach, CA, USA)*

**WORKSHOP 3 – Criteria to keep patients under sapropterin**

*J Rocha (Porto, Portugal)*

**WORKSHOP 4 – Complexity versus precision in dietary treatment**

*A MacDonald (Birmingham, UK) and A Daly (Birmingham, UK)*

**15:30 – 16:00 – Coffee-break**

**16:00 – 17:30 – PARALLEL WORKSHOPS 1, 2, 3 and 4 – ROUND 2**

**WORKSHOP 1 – Dietary treatment in MSUD, OA, UCD**

*J Gribben (London, UK)*

**WORKSHOP 2 – Tools and Apps: What is out there and how does it affect our patients?**

*D Hook (Long Beach, CA, USA)*

**WORKSHOP 3 – Criteria to keep patients under sapropterin**

*J Rocha (Porto, Portugal)*

**WORKSHOP 4 – Complexity versus precision in dietary treatment**

*A MacDonald (Birmingham, UK) and A Daly (Birmingham, UK)*

## **SEPTEMBER 5**

**07:15 – 08:45 SPONSORED BREAKFAST SYMPOSIA (Vitaflo)**

**09:00 – 10:30 PRE-CONGRESS PARALLEL SESSIONS (PC 1, PC2, PC3, PC4a, PC4b)**

**09:00 – 12:30 PC1 - NUTRITION AND DIETETICS: EMERGING THERAPIES AND TECHNOLOGIES**

**Chair:** *A van Wegberg (Nijmegen, Netherlands)*

**Co-chair:** *V Cornejo (Santiago, Chile)*

09:00 – 09:10 Introduction (*A van Wegberg, Nijmegen, Netherlands*)

09:10 – 09:45 From newborn screening to genomics. Changing style of dietary management  
(*R Singh, Atlanta, GA, USA*)

09:45 – 10:15 Arginine supplementation in GA type 1 - particularly emergency management  
(*S Kölker, Heidelberg, Germany*)

10:15 – 10:45 Understanding gut flora and impact in IMD conditions (*I Schwartz, Porto Alegre, Brazil*)

**10:45 – 11:15 Coffee Break**

11:15 – 11:45 Genetically engineered probiotics for the treatment of PKU

(*K Durrer-Deming, Fort Worth, TX, USA*)

11:45 – 12:20 Amino-acid supplementation in MMA/PA: more harm than good? Effects of medical food versus intact protein on branched-chain AA ratio (*I Manoli, Bethesda, MD, USA*)

12:20 – 12:30 Wrap-up (*V Cornejo, Santiago, Chile*)

**09:00 – 12:30 PC2 - METABOLIC DISEASES IN THE PLAIN PEOPLE: THE PATH FROM GENOMIC DISCOVERY TO THERAPY**

**Chair:** *J Vockley (Pittsburgh, PA, USA)*

**Co-Chair:** *L Ghaloul-Gonzalez (Pittsburgh, PA, USA)*

09:00 – 09:10 Introduction (*J Vockley, Pittsburgh, PA, USA*)

09:10 – 09:50 Genetic discovery among the Plain populations x precision medicine (*K Strauss, Strasburg, PA, USA*)

09:50 – 10:30 Determining the Best Liver Transplant Options in IEMs (*R Sindhi, Pittsburgh, PA, USA*)

**10:30 – 11:00 Coffee Break**

11:00 – 11:40 Reduced Intensity Cord Blood or BMT for IEMs (*P Szabolcs, Pittsburgh, PA USA*)

11:40 – 12:20 Utilizing Nat Hist to Assess Outcomes for Novel Therapies (*M Escolar, Pittsburgh, PA, USA*)

12:20 – 12:30 Wrap-up (*L Ghaloul-Gonzalez, Pittsburgh, PA, USA*)

**09:00 – 12:30 PC3 - DISORDERS OF PHOSPHOLIPID METABOLISM**

**Chair:** *R Wanders (Amsterdam, Netherlands)*

**Co-chair:** TBC

09:00 – 09:10 Introduction (*R Wanders, Amsterdam, Netherlands*)

09:10 – 09:50 Cardiolipin and the changing face of Barth Syndrome (*M Schlame, New York, USA*)

09:50 – 10:30 Etherphospholipids and inborn errors of metabolism (*P Brites, Porto, Portugal*)

**10:30 – 11:00 Coffee Break**

11:00 – 11:40 Defects in fatty acid chain elongation (*F Mochel, Paris, France*)

11:40 – 12:20 Lipidomics as a new tool to identify unrecognized defects in fatty acid homeostasis  
(*Benoit Colsch, Paris, France*)

12:20 – 12:30 Wrap-up (TBC)

**09:00 – 10:30 PC4 - ADULT IEM Meeting**

**Co-Chairs:** *F Mochel (Paris, France) and M Tchan (Sydney, Australia)*

- 09:00 – 10:30 Program: 6 presentations, selected from the abstracts submitted  
09:00 – 09:15 Oral Presentation 01 – selected from Adult IEM abstracts  
09:15 – 09:30 Oral Presentation 02 – selected from Adult IEM abstracts  
09:30 – 09:45 Oral Presentation 03 – selected from Adult IEM abstracts  
09:45 – 10:00 Oral Presentation 04 – selected from Adult IEM abstracts  
10:00 – 10:15 Oral presentation 05 – selected from Adult IEM abstracts  
10:15 – 10:30 Oral presentation 06 – selected from Adult IEM abstracts

**10:30 – 11:00 Coffee Break**

**11:00 – 12:20 PC5 - ERNDIM Meeting**

**Chair:** *C Vianey-Saban (Lyon, France)*

- 11:00 – 11:20 What is ERNDIM (*B Fowler, Basel, Switzerland*)  
11:20 – 11:50 Practical Relevance of Measurement Uncertainty in IEM (*J Bonham, Sheffield, UK*)  
11:50 – 12:05 The role of ERNDIM in the MetaBERN (*V Kozich, Prague, Czech Republic*)  
12:05 – 12:30 Targeted NGS for NBS of IEM in UK: a pilot experience (*J Bonham, Sheffield, UK*)

**12:45 – 13:45 SPONSORED LUNCH SYMPOSIA (Amicus, BioMarin, Shire, Sobi)**

**14:00 – 16:00 PL1 - PLENARY SESSION 1 – OPENING SESSION**

**Chair:** *R Giugliani (Porto Alegre, Brazil)*

**Co-chairs:** *J Campistol (Barcelona, Spain), A Ribes (Barcelona, Spain) & A Inwood (Brisbane, QLD, Australia)*

- 14:00 – 14:30 Opening Ceremony  
14:30 – 15:00 IEM in the Era of Genomic Medicine (*J Christodoulou, Melbourne, VIC, Australia*)  
15:00 – 15:30 The link between rare and common diseases (*S Pena, Belo Horizonte, Brazil*)  
15:30 – 16:00 Rare Diseases and Challenges to Health Care (*JF Cabello, Santiago, Chile*)

**16:00 – 16:30 Coffee-break**

**16:30 – 18:00 PL2 - PLENARY SESSION 2 – IEM DIVERSITY**

**Chair:** *M Summar (Washington, DC, USA)*

**Co-chairs:** *B Fowler (Basel, Switzerland) & H Ida (Tokyo, Japan)*

- 16:30 – 17:00 Bioinformatic tools in IEM: genotype-phenotype correlation and prediction (*C van Karnebeek, Vancouver, Canada*)  
17:00 – 17:30 One gene, many faces - the many phenotypes of POLG disease (*R Naviaux, San Diego, CA, USA*)  
17:30 – 18:00 One phenotype, several diseases – The SMA case (*E Schuchman, New York, NY, USA*)

**18:00 – 20:00 WELCOME COCKTAIL**

**SEPTEMBER 6**

**07:15 – 08:45 SPONSORED BREAKFAST SYMPOSIA (BioMarin, Centogene, Genzyme, Nutricia)**

**09:00 – 10:30 PL3 - PLENARY SESSION 3 – ALTERNATIVE APPROACHES TO TREAT THE CNS IN IEM**

**Chair:** *G Salomons (Amsterdam, Netherlands)*

**Co-chairs:** *David Begley (London, UK) & JF Cabello (Santiago, Chile)*

- 09:00 – 09:30 Unanticipated links between rare and common diseases: insights from Niemann-Pick type C disease (*F Platt, Oxford UK*)  
09:30 – 10:00 Soluble Carrier Transporters (*G Salomons, Amsterdam, Netherlands*)  
10:00 – 10:30 Gene Therapy in IEM (*C P Venditti, Washington, DC, USA*)

**10:30 – 11:00 Coffee-Break**

**11:00 – 12:30 PARALLEL SESSIONS (PA1, PA2, PA3, PA4)**

**11:00 – 12:30 PA1 - PEROXISOMAL DISORDERS / THERAPIES FOR THE CNS**

**Chair:** *TBC*

**Co-chair:** *H Amartino (Buenos Aires, Argentina)*

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *Stephen Kemp (Amsterdam, Netherlands)*

11:30 – 11:45 Oral presentation 07 – selected from abstracts

11:45 – 12:00 Oral presentation 08 – selected from abstracts

12:00 – 12:15 Oral presentation 09 – selected from abstracts

12:15 – 12:30 Oral presentation 10 – selected from abstracts

**11:00 – 12:30 PA2 – IEM IN DEVELOPING REGIONS (OVERVIEW) / BIOMARKERS**

**Chair:** *Pascale Karam (Beirut, Lebanon)*

**Co-chair:** *Silvana Zanlungo (Santiago, Chile)*

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *M AlSayed (Riyhad, Saudi Arabia)*

11:30 – 11:45 Oral presentation 11 – selected from abstracts

11:45 – 12:00 Oral presentation 12 – selected from abstracts

12:00 – 12:15 Oral presentation 13 – selected from abstracts

12:15 – 12:30 Oral presentation 14 – selected from abstracts

**11:00 – 12:30 PA3 – POPULATION MEDICAL GENETICS AND IEM**

**Chair:** *Michael Beck (Mainz, Germany)*

**Co-chair:** *Angelina Acosta (Salvador, Brazil)*

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *F Ezgu (Ankara, Turkey)*

11:30 – 11:45 Oral presentation 15 – selected from abstracts

11:45 – 12:00 Oral presentation 16 – selected from abstracts

12:00 – 12:15 Oral presentation 17 – selected from abstracts

12:15 – 12:30 Oral presentation 18 – selected from abstracts

**11:00 – 12:30 PA4 – DIETARY TREATMENT OF IEM**

**Chair:** *A van Wegberg (Nijmegen, Netherlands)*

**Co-chair:** *A Chiesa (Buenos Aires, Argentina)*

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *V Cornejo (Santiago, Chile)*

11:30 – 11:45 Oral presentation 19 – selected from abstracts

11:45 – 12:00 Oral presentation 20 – selected from abstracts

12:00 – 12:15 Oral presentation 21 – selected from abstracts

12:15 – 12:30 Oral presentation 22 – selected from abstracts

**12:45 – 13:45 IOC SOCIETIES BUSINESS MEETINGS (SSIEM, SIMD, ASIEM, SLEIMPN)**

**14:00 – 15:45 PARALLEL SESSIONS (PA5, PA6, PA7, PA8)**

**14:00 – 15:45 PA5 - MITOCHONDRIAL DISORDERS**

**Chair:** *N Griegersen (Copenhagen, Denmark)*

**Co-chair:** *TBC*

14:00 – 14:05 Opening/Introduction

14:05 – 14:30 Overview/Update: *Akira Ohtake (Saitama, Japan)*

14:30 – 14:45 Oral presentation 23 – selected from abstracts

14:45 – 15:00 Oral presentation 24 – selected from abstracts

15:00 – 15:15 Oral presentation 25 – selected from abstracts

15:15 – 15:30 Oral presentation 26 – selected from abstracts

15:30 – 15:45 Oral presentation 27 – selected from abstracts

**14:00 – 15:45 PA6 – FATTY ACID OXIDATION AND BRANCHED-CHAIN AMINO ACID DISORDERS**

**Chair:** *M Wajner (Porto Alegre, Brazil)*

**Co-chair:** *N Longo (Salt Lake City, UT, USA)*

- 14:00 – 14:05 Opening/Introduction
- 14:05 – 14:30 Overview/Update: *J Vockley (Pittsburgh, PA, USA)*
- 14:30 – 14:45 Oral presentation 28 – selected from abstracts
- 14:45 – 15:00 Oral presentation 29 – selected from abstracts
- 15:00 – 15:15 Oral presentation 30 – selected from abstracts
- 15:15 – 15:30 Oral presentation 31 – selected from abstracts
- 15:30 – 15:45 Oral presentation 32 – selected from abstracts

**14:00 – 15:45 PA7 – ORPHAN DRUG DEVELOPMENT: LESSONS LEARNED**

**Chair:** *TBC*

**Co-chair:** *Dafne Horovitz (Rio de Janeiro, Brazil)*

- 14:00 – 14:05 Opening/Introduction
- 14:05 – 14:30 Overview/Update: *M Ries (Heidelberg, Germany)*
- 14:30 – 14:45 Oral presentation 33 – selected from abstracts
- 14:45 – 15:00 Oral presentation 34 – selected from abstracts
- 15:00 – 15:15 Oral presentation 35 – selected from abstracts
- 15:15 – 15:30 Oral presentation 35 – selected from abstracts
- 15:30 – 15:45 Oral presentation 37 – selected from abstracts

**14:00 – 15:45 PA8 – NEWBORN SCREENING / SCREENING TECHNOLOGIES**

**Chair:** *D Webster (Auckland, New Zealand)*

**Co-chair:** *A Fonseca (Rio de Janeiro, Brasil)*

- 14:00 – 14:05 Opening/Introduction
- 14:05 – 14:30 Overview/Update: *G Borrajo (La Plata, Argentina)*
- 14:30 – 14:45 Oral presentation 38 – selected from abstracts
- 14:45 – 15:00 Oral presentation 39 – selected from abstracts
- 15:00 – 15:15 Oral presentation 40 – selected from abstracts
- 15:15 – 15:30 Oral presentation 41 – selected from abstracts
- 15:30 – 15:45 Oral presentation 42 – selected from abstracts

**15:45 – 16:15 Coffee-Break**

**16:15 – 17:45 PL4 - PLENARY SESSION 4 – TRENDS IN NEWBORN SCREENING**

**Chair:** *N Blau (Heidelberg, Germany)*

**Co-chairs:** *B Wilcken (Sydney, NSW, Australia), M Saborio (San Jose, Costa Rica)*

- 16:00 – 16:30 Global overview of NBS (*V Wiley, Sydney, NSW, Australia*)
- 16:30 – 17:00 Is there a limit for the expansion of NBS? (*S Berry*)
- 17:00 – 17:30 Whole Genome Sequencing as a Tool for NBS (*D Dimmock, San Diego, CA, USA*)

**18:00 – 20:00 ATTENDED POSTER SESSION (with drinks and appetizers)**

**20:00 – 22:00 FACULTY MEMBERS DINNER (by invitation only)**

## **SEPTEMBER 7**

**07:15 – 08:45 SPONSORED BREAKFAST SYMPOSIA (Genzyme, Metabolon, Shire)**

**07:15 – 08:45 INFORM BREAKFAST SYMPOSIUM – FATTY ACID OXIDATION DISORDERS**

**Chair:** *M Bennett (Philadelphia, PA, USA)*

**Co-Chair:** *E Goetzman (Pittsburgh, PA, USA)*

07:15 – 07:45 Anti-oxidant therapy as an adjunct for treatment of long chain fatty acid oxidation disorders (*G Leipnitz, Porto Alegre, Brazil*).

07:45 – 08:15 Novel mechanisms of pathogenesis in mitochondrial trifunctional protein deficiency: implications for clinical outcome and treatment (*A El-Gharbawy, Pittsburgh, PA, USA*)

08:15 – 08:45 Rhabdomyolysis and inflammation in metabolic muscle disease (*Y Hamel, Paris, France*)

**09:00 – 10:30 PARALLEL SESSIONS (PA9, PA10, PA11, PA12)**

**09:00 – 10:30 PA9 – TREATMENT OF LYSOSOMAL DISEASES**

**Chair:** *P Hwu (Taipei, Taiwan)*

**Co-chair:** *L A Barrera (Bogota, Colombia)*

09:00 – 09:05 Opening/Introduction

09:05 – 09:30 Overview/Update: *R J Desnick (New York, NY, USA)*

09:30 – 09:45 Oral presentation 43 – selected from abstracts

09:45 – 10:00 Oral presentation 44 – selected from abstracts

10:00 – 10:15 Oral presentation 45 – selected from abstracts

10:15 – 10:30 Oral presentation 46 – selected from abstracts

**09:00 – 10:30 PA10 – MISCELANEOUS DISORDERS (INCLUDING CITRIN METABOLISM)**

**Chair:** *Stephen Cederbaum (San Diego, CA, USA)*

**Co-chair:** *M Vela (Mexico City, Mexico)*

09:00 – 09:05 Opening/Introduction

09:05 – 09:30 Overview/Update: *T Saheki (Kagoshima, Japan)*

09:30 – 09:45 Oral presentation 47 – selected from abstracts

09:45 – 10:00 Oral presentation 48 – selected from abstracts

10:00 – 10:15 Oral presentation 49 – selected from abstracts

10:15 – 10:30 Oral presentation 50 – selected from abstracts

**09:00 – 10:30 PA11 – PSYCHOPATHOLOGY, BRAIN ALTERATIONS AND METABOLISM**

**Chair:** *T Kozicz (New Orleans, LA, USA)*

**Co-chair:** *Andrea Schenone (Buenos Aires, Argentina)*

09:00 – 09:05 Opening/Introduction

09:05 – 09:30 Overview/Update: *C M Lourenço (Ribeirao Preto, Brazil)*

09:30 – 09:45 Oral presentation 51 – selected from abstracts

09:45 – 10:00 Oral presentation 52 – selected from abstracts

10:00 – 10:15 Oral presentation 53 – selected from abstracts

10:15 – 10:30 Oral presentation 54 – selected from abstracts

**09:00 – 10:30 PA12 – VITAMIN AND MICRONUTRIENTS METABOLISM**

**Chair:** *A M Martins (São Paulo, Brazil)*

**Co-chair:** *M Giovannini (Milan, Italy)*

09:00 – 09:05 Opening/Introduction

09:05 – 09:30 Overview/Update: *P Clayton (London, UK)*

09:30 – 09:45 Oral presentation 55 – selected from abstracts

09:45 – 10:00 Oral presentation 56 – selected from abstracts

10:00 – 10:15 Oral presentation 57 – selected from abstracts

10:15 – 10:30 Oral presentation 58 – selected from abstracts

**10:30 – 11:00 Coffee Break**



**11:00 – 12:30 PARALLEL SESSIONS (PA13, PA14, PA15, PA16)**

**11:00 – 12:30 PA13 - DISORDERS OF GLYCOSYLATION AND PROTEIN MODIFICATION**

**Chair:** *E Morava (New Orleans, LA, USA)*

**Co-chair:** *K Raymond (Rochester, MN, USA)*

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *C Astegiano (Cordoba, Argentina)*

11:30 – 11:45 Oral presentation 59 – selected from abstracts

11:45 – 12:00 Oral presentation 60 – selected from abstracts

12:00 – 12:15 Oral presentation 61 – selected from abstracts

12:15 – 12:30 Oral presentation 62 – selected from abstracts

**11:00 – 12:30 PA14 – KETONE BODIES, UREA CYCLE DISORDERS, AND EPIGENETICS**

**Chair:** *T Fukao (Gifu, Japan)*

**Co-chair:** *G Mitchell (Montreal, Canada)*

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *JO Sass (Rheinbach, Germany)*

11:30 – 11:45 Oral presentation 63 – selected from abstracts

11:45 – 12:00 Oral presentation 64 – selected from abstracts

12:00 – 12:15 Oral presentation 65 – selected from abstracts

12:15 – 12:30 Oral presentation 66 – selected from abstracts

**11:00 – 12:30 PA15 – NETWORKING EXPERIENCES/WEB PLATFORMS/REMOTE DIAGNOSIS**

**Chair:** *T Derks (Groningen, Netherlands)*

**Co-chair:** *M Scarpa (Wiesbaden, Germany)*

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *I Schwartz (Porto Alegre, Brazil)*

11:30 – 11:45 Oral presentation 67 – selected from abstracts

11:45 – 12:00 Oral presentation 68 – selected from abstracts

12:00 – 12:15 Oral presentation 69 – selected from abstracts

12:15 – 12:30 Oral presentation 70 – selected from abstracts

**11:00 – 12:30 PA16 – GUT MICROBIOTA AND HUMAN METABOLISM**

**Chair:** *TBC*

**Co-chair:** *Isabel Ibarra (Mexico City, Mexico)*

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *TBC*

11:30 – 11:45 Oral presentation 71 – selected from abstracts

11:45 – 12:00 Oral presentation 72 – selected from abstracts

12:00 – 12:15 Oral presentation 73 – selected from abstracts

12:15 – 12:30 Oral presentation 74 – selected from abstracts

**12:45 – 13:45 SPONSORED LUNCH SYMPOSIA (BioMarin, Genzyme, Orphan/Recordati, Shire)**

**14:00 – 15:30 PL5 - PLENARY SESSION 5 – THE ROLE OF MASSIVE PARALLEL SEQUENCING IN IEM DIAGNOSIS**

**Chair:** *C Dionisi-Vici (Rome, Italy)*

**Co-chairs:** *S McCandless (Cleveland, OH, USA) & F Endo (Kumamoto, Japan)*

14:00 – 14:30 Massive Parallel Sequencing as a first line test for IEM diagnosis (*WW Grody, Los Angeles, CA, USA*)

14:30 – 15:00 IEM as an incidental diagnosis in the investigation with NGS (*Fernando Kok, Sao Paulo, Brazil*)

14:00 – 15:30 Pathogenic genotypes in healthy subjects (*Andres Klein, Santiago, Chile*)

**15:30 – 16:00 Coffee-Break**

**16:00 – 18:00 Poster Viewing**

**20:00 – 24:00 NETWORKING EVENING**

## **SEPTEMBER 8**

### **09:00 – 10:30 PL6 - PLENARY SESSION 6 – TRENDS IN ORGANELLE DISEASES**

**Chair:** *Y Eto (Tokyo, Japan)*

**Co-chairs:** *J Fletcher (Adelaide, SA, Australia) / S Tomatsu (Wilmington, DE, USA)*

09:00 – 09:30 Contemporary Mitochondrial Medicine (*Michio Hirano, New York, NY, USA*)

09:00 – 10:00 Peroxisomal Disorders (*Ronald J Wanders, Amsterdam, Netherlands*)

10:00 – 10:30 Overview on the development of therapies for LSDs (*Lorne Clarke, Vancouver, Canada*)

### **10:30 – 11:00 Coffee-Break**

### **11:00 – 13:00 PL 7 - PLENARY SESSION 7 – CLOSING SESSION**

**Chair:** *J Abdenur (Buenos Aires, Argentina/Orange County, CA, USA)*

**Co-chair:** *K Bhattacharya (Sydney, NSW, Australia) / H Michelakalis (Athens, Greece)*

11:00 – 11:30 Late Breaking News (*3 presenters to be selected from the LBN abstracts*)

11:30 – 12:00 SIMD Awards (*SIMD*), JIEMS Awards (*SLEIMPN*), Garrod Award (*SSIEM*)

12:00 – 12:30 Chamoles Lecture (*Speaker indicated by SLEIMPN*)

12:30 – 13:30 Next meetings presentations (*SLEIMPN, SIMD, ASIEM, JSIMD, SSIEM, ICIEM*)

**13:00 Closure**

### **13:00 – 13:30 Lunch Break**

### **13:30 – 17:30 Networking Afternoon Excursions**

## **BUSINESS MEETINGS**

### **SEP 4**

16:00 – 18:00 – SSIEM Council Meeting

### **SEP 5**

09:00 – 13:00 – JIMD Editors Meeting

09:30 – 12:30 – MetabERN Meeting

11:30 – 13:45 – SIMD Board of Directors Meeting

20:00 – 22:00 – JIMD Communicating Editors Dinner

### **SEP 6**

09:00 – 10:30 – SSIEM–DG Meeting

12:45 – 13:45 – SSIEM Business Meeting

12:45 – 13:45 – SLEIMPN Business Meeting

12:45 – 13:45 – SIMD Business Meeting

12:45 – 13:45 – ASIEM Business Meeting

### **SEP 7**

12:30 – 14:00 – JIMD Communicating Editors Meeting