

PRELIMINARY SCIENTIFIC PROGRAM (2017-06-15)

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 SCIENTIFIC STREAM

Chair: *H Blom (Freiburg, Germany)*

09:00 Newborn Screening for Homocystinurias and Methylation Disorders

09:30 CblC Proteomics

10:00 As above options

10:30 Coffee-Break

11:00 First results CblC disease from EHOD registry

11:30 H2S

12:00 Late-breaking News / or CBS history

- 09:00 PATIENT STREAM**
Chair: *T Morrison (Baulkham Hills, NSW, Australia)*
 09:00 Welcome (*T Morrison, Baulkham Hills, NSW, Australia*)
 09:10 Novel patient organisations: what is planned and what is available for the future
 HCU Network Australia
 HCU Network America
 Brazilian Organization
 09:40 Patient Care in Brazil
 10:00 Round table discussion on what the key patient needs are and most important priorities.
 Include patient personal story
10:30 Coffee-Break
 11:00 Dietary intervention overview
 11:20 Patient perspective on best practices on dietary management
 11:40 Mental health, behaviour challenges and strategies for management
 12:00 Overview of how our understanding has developed over the years and Q&A
- 12:30 INTERACTIVE LUNCH – MEET THE EXPERTS**
- 13:50 JOINT STREAM**
Chair: *I Schwartz (Porto Alegre, Brazil)*
 13:50 Welcome – Introduction to homocystinurias and EHOD (*H Blom, Freiburg, Germany*)
 14:00 CBS Guidelines
 14:20 Overview of research strategy and aims of organisations: collaborations between HCU
 Network Australia and HCU Network America – Discussion on research priorities
 14:40 QoL in Homocystinuria patients
 15:00 ERT for CBS deficiency
 15:20 The unique Qatar experience
 15:40 CblC research
16:00 Coffee-Break
 16:30 CBS deficiency Panel Discussion including guidelines:
 How do we get greater dissemination and adoption of the guidelines & how do we achieve
 timely diagnosis and the role of each stakeholder
 16:30 CblC and MTHFR Panel Discussion including guidelines:
 How do we get greater dissemination and adoption of the guidelines & how do we achieve
 timely diagnosis and the role of each stakeholder
- 18:00 Close**
- 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 on this topic

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

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 01 – selected from abstracts

11:45 – 12:00 Oral presentation 02 – selected from abstracts

12:00 – 12:15 Oral presentation 03 – selected from abstracts

12:15 – 12:30 Oral presentation 04 – selected from abstracts

11:00 – 12:30 PA2 – ORPHAN DRUG DEVELOPMENT: LESSONS LEARNED

Chair: *TBC*

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

11:00 – 11:05 Opening/Introduction

11:05 – 11:30 Overview/Update: *M Ries (Heidelberg, Germany)*

11:30 – 11:45 Oral presentation 05 – selected from abstracts

11:45 – 12:00 Oral presentation 06 – selected from abstracts

12:00 – 12:15 Oral presentation 07 – selected from abstracts

12:15 – 12:30 Oral presentation 08 – 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 09 – selected from abstracts

11:45 – 12:00 Oral presentation 10 – selected from abstracts

12:00 – 12:15 Oral presentation 11 – selected from abstracts

12:15 – 12:30 Oral presentation 12 – 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 13 – selected from abstracts

11:45 – 12:00 Oral presentation 14 – selected from abstracts

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

12:15 – 12:30 Oral presentation 16 – selected from abstracts

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

14:00 – 15:30 PARALLEL SESSIONS (PA5, PA6, PA7, PA8)

14:00 – 15:30 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*

14:30 – 14:45 Oral presentation 17 – selected from abstracts

14:45 – 15:00 Oral presentation 18 – selected from abstracts

15:00 – 15:15 Oral presentation 19 – selected from abstracts

15:15 – 15:30 Oral presentation 20 – selected from abstracts

14:00 – 15:30 PA6 – FATTY ACID OXIDATION 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 21 – selected from abstracts

14:45 – 15:00 Oral presentation 22 – selected from abstracts

15:00 – 15:15 Oral presentation 23 – selected from abstracts

15:15 – 15:30 Oral presentation 24 – selected from abstracts

14:00 – 15:30 PA7 – IEM IN DEVELOPING REGIONS

Chair: *Pascale Karam (Beirut, Lebanon)*

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

14:00 – 14:05 Opening/Introduction

14:05 – 14:30 Overview/Update: *M AlSayed (Riyhad, Saudi Arabia)*

14:30 – 14:45 Oral presentation 25 – selected from abstracts

14:45 – 15:00 Oral presentation 26 – selected from abstracts

15:00 – 15:15 Oral presentation 27 – selected from abstracts

15:15 – 15:30 Oral presentation 28 – selected from abstracts

14:00 – 15:30 PA8 – NEWBORN SCREENING AROUND THE WORLD

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 29 – selected from abstracts

14:45 – 15:00 Oral presentation 30 – selected from abstracts

15:00 – 15:15 Oral presentation 31 – selected from abstracts

15:15 – 15:30 Oral presentation 32 – selected from abstracts

15:30 – 16:00 Coffee-Break

16:00 – 17:30 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*)

17:30 – 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 - 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 33 – selected from abstracts

09:45 – 10:00 Oral presentation 34 – selected from abstracts

10:00 – 10:15 Oral presentation 35 – selected from abstracts

10:15 – 10:30 Oral presentation 36 – selected from abstracts

09:00 – 10:30 PA10 – CITRIN METABOLISM & MISCELLANEOUS DISORDERS

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 37 – selected from abstracts

09:45 – 10:00 Oral presentation 38 – selected from abstracts

10:00 – 10:15 Oral presentation 39 – selected from abstracts

10:15 – 10:30 Oral presentation 40 – selected from abstracts

09:00 – 10:30 PA11 – PSYCHOPATHOLOGY 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 41 – selected from abstracts

09:45 – 10:00 Oral presentation 42 – selected from abstracts

10:00 – 10:15 Oral presentation 43 – selected from abstracts

10:15 – 10:30 Oral presentation 44 – 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 45 – selected from abstracts

09:45 – 10:00 Oral presentation 46 – selected from abstracts

10:00 – 10:15 Oral presentation 47 – selected from abstracts

10:15 – 10:30 Oral presentation 48 – selected from abstracts

10:30 – 11:00 Coffee Break

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

11:00 – 12:30 PA13 - CONGENITAL DISORDERS OF GLYCOSYLATION

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 49 – selected from abstracts

11:45 – 12:00 Oral presentation 50 – selected from abstracts

12:00 – 12:15 Oral presentation 51 – selected from abstracts

12:15 – 12:30 Oral presentation 52 – selected from abstracts

11:00 – 12:30 PA14 – KETONE BODIES: ENERGY SUPPLY 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 53 – selected from abstracts

11:45 – 12:00 Oral presentation 54 – selected from abstracts

12:00 – 12:15 Oral presentation 55 – selected from abstracts

12:15 – 12:30 Oral presentation 56 – selected from abstracts

11:00 – 12:30 PA15 – NETWORKING EXPERIENCES – REMOTE DIAGNOSIS/FOLLOW-UP OF IEM

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 57 – selected from abstracts

11:45 – 12:00 Oral presentation 58 – selected from abstracts

12:00 – 12:15 Oral presentation 59 – selected from abstracts

12:15 – 12:30 Oral presentation 60 – 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 61 – selected from abstracts

11:45 – 12:00 Oral presentation 62 – selected from abstracts

12:00 – 12:15 Oral presentation 63 – selected from abstracts

12:15 – 12:30 Oral presentation 64 – 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 (*SLEIMP*N), Garrod Award (*SSIEM*)

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

12:30 – 13:30 Next meetings presentations (*SLEIMP*N, *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 – SLEIMP

12:45 – 13:45 – SIMD Business Meeting

12:45 – 13:45 – ASIEM Business Meeting

SEP 7

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