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
SS2 - PRE-CONGRESS SATELLITE SYMPOSIUM 2 - NEWBORN SCREENING

Contact Person: Gustavo Borrajol (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 Wei, Atlanta, GA, USA)
15:00 - 15:30 External Quality Assurance Program for Latin America (G Borrajol, 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 Borrajol, La Plata, Argentina)

SS3 - PRE-CONGRESS SATELLITE SYMPOSIUM 3 - HOMOCYSTINURIA

Contact Person: Tara Morrison (Baulkham Hills, NSW, Australia)
Contact Email: tara@hcunetworkaustralia.org.au

08:30 – 18:00

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 ChlC 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 chlC 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: Annemieke van Wegberg (Nijmegen, Netherlands)
   Contact E-mail: Annemieke.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 (Vitaflor)

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 Ghalioul-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 Ghalioul-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)**
<table>
<thead>
<tr>
<th>Time</th>
<th>Session Title</th>
<th>Chair/Co-Chair</th>
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<tr>
<td>07:15 – 08:45</td>
<td>SPONSORED BREAKFAST SYMPOSIA (Genzyme, Metabolon, Shire)</td>
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<td>07:15 – 08:45</td>
<td>INFORM BREAKFAST SYMPOSIUM – FATTY ACID OXIDATION DISORDERS</td>
<td>Chair: M Bennett (Philadelphia, PA, USA) Co-Chair: E Goetzman (Pittsburgh, PA, USA)</td>
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<td>07:15 – 07:45</td>
<td>Anti-oxidant therapy as an adjunct for treatment of long chain fatty acid oxidation disorders (G Leipnitz, Porto Alegre, Brazil).</td>
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<td>07:45 – 08:15</td>
<td>Novel mechanisms of pathogenesis in mitochondrial trifunctional protein deficiency: implications for clinical outcome and treatment (A El-Ghrobawy, Pittsburgh, PA, USA)</td>
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<td>08:15 – 08:45</td>
<td>Rhabdomyolysis and inflammation in metabolic muscle disease (Y Hamel, Paris, France)</td>
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<td>09:00 – 10:30</td>
<td>PARALLEL SESSIONS (PA9, PA10, PA11, PA12)</td>
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<td>09:00 – 10:30</td>
<td>PA9 – TREATMENT OF LYSOSOMAL DISEASES</td>
<td>Chair: P Hwu (Taipei, Taiwan) Co-chair: I A Barrera (Bogota, Colombia)</td>
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<td>09:00 – 09:05</td>
<td>Opening/Introduction</td>
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<td>09:05 – 09:30</td>
<td>Overview/Update: R J Desnick (New York, NY, USA)</td>
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<td>09:30 – 09:45</td>
<td>Oral presentation 43 – selected from abstracts</td>
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<td>10:15 – 10:30</td>
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<td>09:00 – 10:30</td>
<td>PA10 – MISCELLANEOUS DISORDERS (INCLUDING CITRIN METABOLISM)</td>
<td>Chair: Stephen Cederbaum (San Diego, CA, USA) Co-chair: M Vela (Mexico City, Mexico)</td>
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<td>09:00 – 09:05</td>
<td>Opening/Introduction</td>
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<td>09:05 – 09:30</td>
<td>Overview/Update: T Saheki (Kagoshima, Japan)</td>
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<td>09:00 – 10:30</td>
<td>PA11 – PSYCHOPATHOLOGY, BRAIN ALTERATIONS AND METABOLISM</td>
<td>Chair: T Kozicz (New Orleans, LA, USA) Co-chair: Andrea Schenone (Buenos Aires, Argentina)</td>
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<td>09:00 – 09:05</td>
<td>Opening/Introduction</td>
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<td>09:05 – 09:30</td>
<td>Overview/Update: C M Lourenço (Ribeirao Preto, Brazil)</td>
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<td>09:30 – 09:45</td>
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<td>09:45 – 10:00</td>
<td>Oral presentation 52 – selected from abstracts</td>
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<td>10:00 – 10:15</td>
<td>Oral presentation 53 – selected from abstracts</td>
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<td>10:15 – 10:30</td>
<td>Oral presentation 54 – selected from abstracts</td>
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<td>09:00 – 10:30</td>
<td>PA12 – VITAMIN AND MICRONUTRIENTS METABOLISM</td>
<td>Chair: A M Martins (São Paulo, Brazil) Co-chair: M Giovannini (Milan, Italy)</td>
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<tr>
<td>09:00 – 09:05</td>
<td>Opening/Introduction</td>
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<td>09:05 – 09:30</td>
<td>Overview/Update: P Clayton (London, UK)</td>
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<td>09:30 – 09:45</td>
<td>Oral presentation 55 – selected from abstracts</td>
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<td>09:45 – 10:00</td>
<td>Oral presentation 56 – selected from abstracts</td>
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<td>10:00 – 10:15</td>
<td>Oral presentation 57 – selected from abstracts</td>
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<td>10:15 – 10:30</td>
<td>Oral presentation 58 – selected from abstracts</td>
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<td>10:30 – 11:00</td>
<td>Coffee Break</td>
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11:00 – 12:30 PARALLEL SESSIONS (PA13, PA14, PA15, PA16)

11:00 – 12:30 PA13 - DISORDERS OF GLYCOSILATION 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 PL7 - 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 Chamois Lecture (Speaker indicated by SLEIMPN)
12:30 – 13:30 Next meetings presentations (SLEIMPN, SIMD, ASIEM, ISIMD, 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