PARALLEL SESSIONS

Wednesday 7th September 08:30 – 10:00

Parallel Session 1

Parallel session 1A: Mitochondrial and fatty acid oxidation disorders

Auditorium
T. Derks, The Netherlands; U. Caruso, Italy

1) **O-041**: Cytosolic phosphoenolpyruvate carboxykinase deficiency presenting with acute liver failure following gastroenteritis.
   S. Santra, UK

2) **O-035**: Decreased stability of the OCTN2 carnitine transporter in patients with primary carnitine deficiency.
   M. Frigeni, USA

3) **O-036**: Cellular models for medium-chain acyl-CoA dehydrogenase deficiency based on induced pluripotent stem cell technology.
   P. Bross, Denmark

4) **O-040**: Clinical exome sequencing in 900 index cases: diagnostic rate and new disease genes.
   T.B. Haack, Germany

5) **O-033**: Characterizing the molecular architecture of mitochondrial energy metabolism apparatus.
   J. Vockley, USA

6) **O-042**: Urinary organic acids in paediatric single mitochondrial DNA deletion disorders.
   S. Boenzi, Italy
Parallel session 1B: Glycosylation and carbohydrate disorders

Blue Room
R. Santer, Germany; P. Strisciuglio, Italy

1) **O-030**: The development and validation of a semi-automated enzyme panel for muscle glycolytic disorders.
   *R.G. Wigley, UK*

2) **O-031**: A conserved phosphatase destroys toxic glycolytic side-products in mammals and yeasts.
   *F. Baldin, Belgium*

3) **O-032**: Is G6PC3, the enzyme deficient in severe congenital neutropenia type 4, really a glucose-6-phosphatase?
   *M. Veiga-da-Cunha, Belgium*

4) **O-056**: A novel sugar metabolic pathway in human: ISPD synthesises CDP-ribitol.
   *D.J. Lefeber, The Netherlands*

5) **O-057**: ISPD produces CDP-ribitol used by FKTN and FKRP to transfer ribitol-phosphate onto α-dystroglycan.
   *G.T. Bommer, Belgium*

6) **O-060**: NANS-mediated synthesis of sialic acid is required for brain and Skeletal development
   *C. van Karnebeek, Canada*

Parallel session 1C: Miscellaneous disorders

Red Room
V. Kozich, Czech Republic; D. Martinelli, Italy

1) **O-003**: Multi-omics tools for the diagnosis and treatment of rare neurological disease.
   *T.L. Simmons, CH*

2) **O-004**: Investigating applications of next generation sequencing in newborn screening.
   *C.M. Gladding, UK*

3) **O-005**: A newborn screening method for Cerebrotendinous Xanthomatosis: data from a pilot validation study.
   *F.M. Vaz, The Netherlands*

4) **O-062**: Progressive cognitive deterioration and pathological hallmarks in murine model creatine transporter deficiency.
L. Baroncelli, Italy

5) **O-065**: GNAO1 mutation: a new cause of transmembrane signalling derangement causing early onset movement disorder.
   F.R. Danti, Italy

6) **O-068**: Developments in the diagnosis and treatment of PNPO deficiency.
   M.P. Wilson, UK

### Parallel session 1D: Nutrition and dietetics & miscellaneous

**Salone della Cultura**

*V. Cornejo, Chile; S. Paci, Italy*

1) **O-002**: A double-blind placebo-controlled trial of triheptanoin in adult polyglucosan body disease.
   R. Schiffmann, USA

2) **O-007**: Longitudinal study examining nutritional status in children with organic acidaemias on a modular feed using a protein free module especially developed for children with IMD.
   A. Daly, UK

3) **O-008**: Dietary treatment of 49 MSUD Italian patient.
   S. Salera, Italy

4) **O-009**: Protein intake and physical activity are associated with body composition in patients with phenylalanine hydroxylase (PAH) deficiency.
   R.H. Singh, USA

5) **O-014**: Large neutral amino acid supplementation as a possible alternative treatment for adult PKU patients: evidence in PKU mice.
   D. Van Vliet, The Netherlands

6) **O-034**: Mitochondrial fatty acid biosynthesis (mFASII) mediates the substrate switch in white skeletal muscle of very-long-chain acyl-CoA dehydrogenase- (VLCAD-/-)- deficient mice.
   S. Tucci, Germany
Wednesday 7th September  14:15 – 15:45

Parallel Session 2

Parallel session 2A: Aminoacid and urea cycle disorders

Blue Room

J. Häberle, CH; M. Spada, Italy

Update lecture: Inherited disorders of proline metabolism

V. Rubio, Spain


R. Yahyaoui, Spain

2) O-018: Minimal NTBC concentrations necessary to prevent formation of succinylacetone in Tyrosinemia type 1 patients.

H.E. Van Reemst, The Netherlands

3) O-019: Genetic cause and prevalence of hydroxyprolinemia.

C. Staufner, Germany

4) O-020: Gain of function mutation in GLS1 causes infantile onset cataract and profound cognitive impairment.

L. Rumping, The Netherlands

Parallel session 2B: Mitochondrial disorders

Salone della Cultura

R. Van Coster, Belgium; E. Bertini, Italy

Update lecture: New treatments in mitochondrial disorders

H. R. Horvath, UK


A. Pop, The Netherlands
6) **O-037**: TANGO2 deficiency, a novel neurometabolic disorder with recurrent encephalo-cardio-myopathic crises.
   C. Muhlhausen, Germany

7) **O-038**: Decanoic acid treatment of fibroblasts from patients with nuclear-encoded complex I deficient Leigh syndrome: a step towards personalised medicine?
   S. Rahman, UK

8) **O-039**: A novel causative gene of mitochondrial respiratory chain disorders in an apparent life-threating event (ALTE).
   A. Matsunaga, Japan

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**Parallel session 2C: Laboratory diagnosis of lysosomal disorders**

**Auditorium**

*H. Michelakakis, Greece; A. Dardis, Italy*

**Update lecture: Contribution of tandem mass spectrometry to the diagnosis of lysosomal storage disorders**

*M. Piraud, France*

1) **O-043**: A reliable multiplex mass spectrometry analysis of glycosaminoglycans for mucopolysaccharidoses.
   C. Auray-Blais, Canada

2) **O-045**: Increased collagen glycosylated hydroxylysine in the urine of MPS I, II and VI patients.
   W.E. Heywood, UK

3) **O-048**: Identification of a new biomarker in Fabry disease by plasma proteomic analysis.
   B.H. Lee, Korea

4) **O-050**: Low reliability of functional (enzymatic) diagnostics of lysosomal storage disorders in Dry Blood Spots (DBS) compared to fibroblasts.
   K. Schoonderwoerd, The Netherlands
Parallel session 2D: New therapies

Red Room
B. Plecko, CH; N. Longo, US

Update lecture: Gene therapy in hyperoxaluria
E. Salido, Spain

1) **O-001**: Interim data from a randomized, placebo controlled, phase 1 study of ALN-AS1, an investigational RNAi therapeutic for the treatment of acute hepatic porphyria.
   *E. Sardh, Sweden*

2) **O-016**: Effect of enzyme replacement therapy on osteoporosis in several CBS-deficient homocystinuric mouse models
   *T. Majtan, USA*

3) **O-028**: The utility of patient-derived hepatocytes for developing liver-directed therapies in propionic acidemia.
   *B.R. Wamhoff, USA*

4) **O-063**: Aminoadipate semialdehyde synthase (AASS) as a therapeutic target for pyridoxine dependent epilepsy by substrate reduction.
   *W.W. Yue, UK*
Thursday 8\textsuperscript{th} September 10:30 – 12:00

Parallel Session 3

Parallel session 3A: Methylation disorders

Blue Room

I. Tavares de Almeida, Portugal; G. La Marca, Italy

Update lecture: What is new in methylation disorders
I. Baric, Croatia

1) **O-017**: S-Adenosylhomocysteine alters methylation of cellular RNA.
   M. Barroso, USA

2) **O-066**: Interaction and characterization of the cblF (LMBD1) and cblU (ABCD4) membrane proteins.
   S. Froese, CH

3) **O-067**: Disturbed regulation of methylenetetrahydrofolate reductase by S-adenosylmethionine.
   D.E. Smith, The Netherlands

4) **O-069**: The low-density lipoprotein (LDL) receptor-related protein 2 is essential for the exosome-dependent cerebral folate transport.
   R. Steinfeld, Germany

Parallel session 3B: New therapies in lysosomal disorders

Auditorium

M. L. Couce, Spain; A. Fiumara, Italy

Update lecture: Gene therapy in lysosomal storage diseases
A. Aiuti, Italy

1) **O-044**: Long-term Outcomes with rhGUS in a Phase I/II Clinical Trial in MPS VII.
   A.S. Jones, UK

2) **O-047**: ZFN-mediated correction of murine MPS I and MPS II models by expression of the human alpha-L-iduronidase and iduronate-2-sulfatase cDNAs from the albumin locus.
C.B. Chester, USA
3) O-052: Intracerebroventricular cerliponase alfa (BMN 190) in children with CLN2
disease: Results from a Phase 1/2, open-label, dose-escalation study.
   A. Schulz, Germany

4) O-053: Novel treatment for Fabry disease - IV administration of plant derived alpha-gal-
a enzyme safety and efficacy, 1 year experience.
   R. Schiffmann, USA

Parallel session 3C: Glycosylation and cellular network disorders

Salone della cultura
   S. Grunewald, UK; R. Barone, Italy

Update lecture: What is new in CDGs
   J. Jaeken, Belgium

1) O-055: A novel group of metabolic disorders due to tissue-specific defects in V-ATPase
   assembly.
   D.J. Lefeber, The Netherlands

2) O-058: Toward a folding therapy for PMM2-CDG.
   P. Yuste-Checa, Spain

3) O-059: MAGT1-deficiency: new insights into a controversial protein with a key role in N-
glycosylation.
   R. Peanne, Belgium

4) O-061: SLC39A8 deficiency is a novel treatable disorder of manganese metabolism and
glycosylation.
   J.H. Park, Germany
Thursday 8th September  13:15 – 14:45

Parallel Session 4

Parallel session 4A: Organic acidurias

Blue Room
D. Karall, Austria; F. Porta, Italy

1) **O-022**: A fish model for propionic acidemia: increased survival and improvement of neurological phenotype by anaplerotic diet.
   V. Ginocchio, Italy

2) **O-023**: Medium term outcome of liver transplantation for children with propionic acidaemia.
   R. Vara, UK

3) **O-024**: Propionate anions, accumulated in Propionic Acidemia, affect cardiac excitation-contraction coupling, gene regulation and cellular growth, which may contribute to heart dysfunction.
   K. Ford, UK

4) **O-025**: Axonal peripheral neuropathy in propionic acidaemia: a severe side effect of long-term metronidazole treatment.
   D. Diodato, Italy

5) **O-026**: Stable isotope breath testing to assess in vivo metabolite flux in methylmalonic acidemia (MMA). From mouse models to patients.
   I. Manoli, USA

6) **O-027**: Insights into disease mechanisms of cblA-type methylmalonic aciduria from 67 new patients and functional MMAA missense mutation characterization.
   T. Plessl, CH

Parallel session 4B: Phenylketonuria & neurotransmitters disorders

Salone della Cultura
N. Blau, CH; V. Leuzzi, Italy

1) **O-010**: Secondary pterins alteration in patients with Phenylalanine Hydroxylase deficiency.
   F. Nardecchia, Italy
2) **O-011**: Phenylalanine hydroxylase N-terminal domain is an allosteric binding site and can be target for pharmacological chaperone design.
   *W. W. Yue, UK*

3) **O-012**: The first structure of full-length phenylalanine hydroxylase has finally been determined.
   *E. K. Jaffe, USA*

   *N. Longo, USA*

5) **O-015**: New generation of chemical scaffolds able to bind to human phenylalanine hydroxylase.
   *P. Leandro, Portugal*

6) **O-064**: Clinical, biochemical and genetic approaches to improve the diagnosis of neurodevelopmental diseases related to neurotransmitter metabolism.
   *L. Christa, France*

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**Parallel session 4C: Lysosomal & autophagy disorders**

**Auditorium**
*S. A. Jones, UK; B. Bembi, Italy*

1) **O-021**: Ammonia activates hepatic autophagy in vivo and its enhancement protects against acute and chronic hyperammonemia.
   *L. Soria, Italy*

2) **O-046**: Initial, 24 week results of heparan sulfate (HS) levels in cerebrospinal fluid (CSF), brain structural MRI and neurocognitive evaluations in an open label, phase I/II, first-in-human clinical trial of intravenous SBC-103 in mucopolysaccharidosis IIB.
   *C. B. Whitley, USA*

3) **O-049**: Farber disease: acid ceramidase deficiency is more common than previously thought and slowly progressive disease may only be diagnosed in adulthood.
   *A. Solyom, USA*

4) **O-051**: TAR-DNA binding protein 43 (TDP-43) pathology in Niemann Pick type C disease.
   *A. Dardis, Italy*

5) **O-054**: The emerging neurocognitive profile of classic infantile Pompe disease.
   *J. M. P. Van den Hout, The Netherlands*

6) **O-070**: The immunological basis of Vici syndrome.
   *E. Piano Mortari, Italy*