ICIEM 2013 Barcelona - Scientific Programme

TUESDAY, SEPTEMBER 3rd

15:00 - 16:30 **AUDITORIUM**	**Plenary Session 1** **GLYCOSYLATION DEFECTS** **Chairs:** Jaak Jaeken, *University Hospital Gasthuisberg, Leuven, Belgium* Paz Briones, *Hospital Clínic and CSIC, Barcelona, Spain* 15:00 **Type 2 Golgi - biochemical update** Hudson H. Freeze, *Sanford-Burnham Medical Research Institute, La Jolla CA, United States* 15:30 **Clinical phenotype update diagnostic algorithms** Eva Morava, *Tulane University Medical School , New Orleans LA, United States* 16:00 **Animal and cellular models** Christian Koerner, *Center for Child and Adolescent Medicine Heidelberg, Heidelberg, Germany*
17:00 - 19:00 **AUDITORIUM**	**Plenary Session 2** **MOVEMENT DISORDERS AND INBORN ERRORS OF METABOLISM** **Chairs:** Emilio Fernández, *Hospital Sant Joan de Deu, Barcelona, Spain* Michael Gibson, *Washington State University, Spolcane, United States* 17:00 **Clinical approach to metabolic dystonias** Jean Pierre Lin, *Guy's and St Thomas' NHS Foundation Trust, London, United Kingdom* 17:30 **Rigid akinetic syndrome in inborn errors of metabolism** Angels García Cazorla, *Hospital Sant Joan de Déu , Barcelona, Spain* 18:00 **The link between the GBA gene and parkinsonism**

	Kathrin Brockmann, *Zentrum fur Neurologie, Abteilung Neurodegeneration, Tubingen, Germany* 18:30 **Neurodegeneration with brain iron accumulation (NBIA)** Susan Hayflick, *Oregon Health and Science University, Portland, United States*

WEDNESDAY, SEPTEMBER 4th

08:30 - 10:30 **AUDITORIUM**	**Plenary Session 3** **NOVEL TREATMENT STRATEGIES IN INBORN ERRORS OF METABOLISM** **Chairs:** Guillem Pintos, *Hospital Germans Trias i Pujol, Badalona, Spain* Janice Fletcher, *Women's and Children's Hospital, North Adelaide, Australia* 08:30 **Strategies for gene therapy** Fátima Bosch, *Universidad Autónoma de Barcelona, Barcelona, Spain* 09:00 **Adenosine deaminase gene therapy** Alessandro Aiuti, *Ospedale San Raffaele, Milano, Italy* 09:30 **Chaperone therapies. Functional principles** Ania Carolina Muntau, *Dr. von Hauner Children's Hospital, Munich , Germany* 10:00 **Liver stem cells transplantation** Etienne Sokal, *Cliniques Universitaires St Luc, Leuven, Belgium*
11:00 - 12:30	**PARALLEL WORKSHOPS** ROOM H1+H2 **Workshop 1: NEUROTRANSMITTER DEFECTS** **Chairs:** Nenad Blau, *University Children's Hospital, Heidelberg , Germany* Simon Heales, *Great Ormond Street Hospital, London, United Kingdom*

	ROOM H3+J **Workshop 2: PHENYLKETONURIA. RECENT TRENDS IN THERAPY** **Chairs:** Verónica Cornejo, *University of Chile, Santiago, Chile* Francjan van Spronsen, *University Medical Centre Groningen, Groningen, The Netherlands* AUDITORIUM **Workshop 3: LYSOSOMAL DISEASES** **Chairs:** Roberto Giugliani, *Hospital de Clinicas de Porto Alegre, Porto Alegre, Brazil* Hiroyuki Ida, *Jikei University School of Medicine, Nishi-Shimbashi, Minato-ku, Tokyo, Japan* ROOM F **Workshop 4: FATTY ACID OXIDATION DISORDERS AND METABOLISM OF KETONE BODIES** **Chairs:** Jörn Oliver Sass , *University of Zürich Children's Hospital, Zürich, Switzerland* Grant Mitchell, *Medical Genetics, CHU Sainte-Justine, Montreal, Canada*
15:00 - 16:30	**PARALLEL ORAL PRESENTATIONS** ROOM F **Oral presentations 1: NUTRITION AND DIETETICS** **Chairs:** Marjorie Dixon, *Great Ormond Street Hospital, London, United Kingdom* Mª Luz Couce, *Hospital Universitario de Santiago, Santiago de Compostela, Spain* ROOM H3+J

	Oral presentations 2: ORGANIC ACIDS AND CREATINE DISORDERS **Chairs:** Gajja Salomons, *VU University Medical Center, Amsterdam, The Netherlands* Belén Pérez Dueñas, *Hospital Sant Joan de Déu, Barcelona, Spain* AUDITORIUM **Oral presentations 3: MITOCHONDRIAL DISORDERS I** **Chairs:** Ute Spiekerkoetter, *University Children's Hospital, Freiburg, Germany* Mª Teresa García-Silva, *Hospital 12 de Octubre, Madrid, Spain* ROOM H1+H2 **Oral presentations 4: MISCELLANEOUS** **Chairs:** Philip Mayne, *Children's University Hospital, Dublin, Ireland* Mª Ángeles Ruiz, *Hospital Son Espases, Palma de Mallorca, Spain*
17:00 - 18:30	**PARALLEL ORAL PRESENTATIONS** AUDITORIUM **Oral presentations 5: MITOCHONDRIAL DISORDERS II** **Chairs:** Shamima Rahman, *Institute of Child Health, London, United Kingdom* Frederic Tort, *Hospital Clínic-CIBERER, Barcelona, Spain* ROOM H1+H2 **Oral presentations 6: AMINO ACID DISORDERS** **Chairs:** Victor Kozich, *Charles University In Prague, First Faculty of Medicine, Prague, Czech Republic* Mireia del Toro, *Hospital Universitari Vall d'Hebron, Barcelona, Spain* ROOM F

Oral presentations 7: METHODS AND TREATMENT STRATEGIES

Chairs:

Philippa Mills, *Institute of Child Health, London, United Kingdom*

Mª José Coll, *Hospital Clínic, Barcelona, Spain*

ROOM H3+J

Oral presentations 8: CDG, LYSOSOMAL AND PEROXISOMAL DISORDERS

Chairs:

Fanny Mochel, *Hospital Pitié-Salpêtrière, Paris, France*

Marisa Girós, *Hospital Clinic, Barcelona, Spain*

THURSDAY, SEPTEMBER 5th

08:30 - 10:30

AUDITORIUM

Plenary Session 4

THE CHANGING FACE OF LABORATORY DIAGNOSTICS OF INBORN ERRORS OF METABOLISM

Chairs:

Christine Vianey-Saban, *Centre de Biologie et de Pathologie Est (CBPE) CHU de Lyon, Lyon, France*

Johannes Zschocke, *Medical University Innsbruck, Innsbruck, Austria*

08:30 **Targeted metabolomic approach using Tandem Mass Spectrometry**

Neil Dalton, *King's College London, Evelina Children's Hospital, London, United Kingdom*

09:00 **Exome function and content, perspectives for diagnosis and treatment**

Bas van Balkom, *University Medical Certer Utrecht, Utrecht, The Netherlands*

09:30 **Exome and Genome sequencing. Bioinformatics**

Gert Matthijs, *University of Leuven, Leuven, Belgium*

	10:00 **The role of targeted DNA enrichment panels in the diagnosis of inborn errors of metabolism** Xavier Estivill, *Center for Genomic Regulation, Barcelona , Spain*
11:00 - 12:30	**PARALLEL WORKSHOPS** AUDITORIUM **Workshop 5: OUTCOME OF PATIENTS DETECTED THROUGH EXPANDED NEWBORN SCREENING** **Chairs:** Georg Hoffmann, *University Children's Hospital, Heidelberg, Germany* Avihu Boneh, *Royal Children's Hospital, Melbourne, Australia* ROOM F **Workshop 6: THE POTENTIAL OF MODEL SYSTEMS TO UNDERSTAND INBORN ERRORS OF METABOLISM** **Chairs:** Ubaldo Caruso, *Gaslini Institute- University , LABSIEM , Genova, Italy* Fumio Endo, *Kumamoto University, Kurokami Kumamoto, Japan* ROOM H1+H2 **Workshop 7: METABOLIC LEUKODYSTROPHIES** **Chairs:** Frederic Sedel, *Pitie-Salpêtrière Hospital, Paris, France* Barbara Plecko, *Kinderspital Zurich, Zurich, Switzerland* ROOM H3+J **Workshop 8: NUCLEAR ENCODED MITOCHONDRIAL DISEASES** **Chairs:** Ramon Martí, *Vall d'Hebron Institut de Recerca, Barcelona, Spain* Wolfgang Sperl, *Paracelsus Medical Unicersity- Landeskliniken, Salzburg, Austria*

FRIDAY, SEPTEMBER 6th

08:30 - 10:30 **AUDITORIUM**	**Plenary Session 5** **DISEASES RELATED TO MITOCHONDRIAL COFACTOR METABOLISM** **Chairs:** Carlos Santos-Ocaña, *Universidad Pablo de Olavide, Sevilla, Spain* Holger Prokisch, *Klinikum Rechts der Isar, Munich, Germany* 08:30 **CoQ10 deficiencies** Michio Hirano, *Columbia University Medical Center, New York, United States* 09:00 **Defects of thiamine transport and metabolism** Garry Brown, *University of Oxford, Oxford, United Kingdom* 09:30 **Lipoic acid biosynthesis defects** Johannes Mayr, *Paracelsus Medical University, Salzburg, Austria* 10:00 **Diseases related to iron sulphur cluster metabolism** Roland Lill, *Philipps-Universität Marburg, Marburg, Germany*
11:00 - 12:30	**PARALLEL WORKSHOPS** ROOM H1+H2 **Workshop 9: TRANSPORT DEFECTS** Chairs: Graziela Uziel, *Instituto Neurologico C. Besta, Milano, Italy* Lourdes Desviat, *Universidad Autónoma de Madrid, Madrid, Spain* AUDITORIUM **Workshop 10: PURINE/PYRIMIDINE AND UREA CYCLE DISORDERS** Chairs: José Antonio Arranz, *Hospital Universitari Vall d'Hebron, Barcelona, Spain* Jorgen Bierau, *Maastricht University Medical Centre, Maastricht , The*

	Netherlands ROOM F **Workshop 11: COUNTRIES WITH EMERGING INBORN ERRORS OF METABOLISM SERVICES** Chairs: Mubeccel Demirkol, *Children's Hospital, 1st Med Faculty, Istanbul, Turkey* Anil Jalan, *Navi Mumbai Institute of Research, Navi Mumbai, India* ROOM H3+J **Workshop 12: DISORDERS OF COMPLEX LIPIDS** Chairs: Ronald Wanders, *Academic Medical Center, Amsterdam , The Netherlands* Barbara Burton, *Ann and Robert H Lurie Children's Hospital, Chicago, United States*
13:30 - 16:45 **AUDITORIUM**	**Chairs:** Cornelis Jakobs, *VU University Medical Center, Amsterdam, The Netherlands* Peter Clayton, *Institute of Child Health, London, United Kingdom* **HOT TOPICS AND LATE BREAKING NEWS** 13:30 **LBN-01: A new congenital disorder of glycosylation (CDG) discovery of signal sequence receptor SSR3 to be essential for efficient protein-N-glycosylation at distinct sites** **Rust S[1], Moormann S, Reunert J, Wada Y, Marquardt T** *[1]Leibniz-Inst for Arteriosclerosis Res, Münster, Germany* 13:45 **LBN-02: identification of mutations in LIPT1 as a cause of an early onset fatal disease associated with a specific lipoylation defect of the 2-oxoacid dehydrogenase complexes**

	Tort F[1], Garcia-Cazorla A, Ferrer-Cortès X, Thió M, Quintana E, Bujan N, Navarro-Sastre A, Matalonga L, Artuch R, Acquaviva C, Vianey-Saban C, Briones P, Ribes A *[1]Hospital Clínic, IDIBAPS,U737-CIBERER, Barcelona, Spain* 14:00 **LBN-03: Efficacious and sustained gene-therapy of canavan disease by singl intravenous doses of novel rAAVs** Gao G[1], Ahmed S, Matalon R *[1]University of Massachusetts Medical School, Worcester, USA* 14:15 **LBN-04: Mutations in NALCN cause an autosomal recessive syndrome with mild facial dysmorphism variable hypotonia, speech delay, chronic constipation and intellectual disability** Al-Sayed M[1], Al-Zaidan H, Albakheet A, Hakami H, Kenana R, Al-Yafee Y, Al-Dosary M, Qari A, Al-Sheddi T, Al-Muheiza M, Al-Qubbaj W, Lakmache Y, Al-Hindi H, Ghaziuddin M, Colak D, Kaya N *[1]King Faisal Specialist Hospital and Research Center, Riyadh, Saudi Arabia*
14:30	**KOMROWER LECTURE: The daily challenge of understanding and treating metabolic diseases in children** Carlo Dionisi-Vici, *Children's Research Hospital Bambino Gesù, Rome, Italy*
15:15	**EMMANUEL SHAPIRA AWARD** (To the best paper of MGM) **Restoration of impaired nitric oxide production in MELAS syndrome with citrulline and arginine supplementation.** Molecular Genetics and Metabolism Volume 105, Issue 4, (April 2012) pg 607-614. Ayman El-Hattab, *Children's Hospital King Fahad Medical City Riyadh, Saudia Arabia*

15:45	**ARCHIBALD GARROD AWARD** (To the best paper of JIMD) **The Adult Galactosemic Phenotype** Journal of Inherited Metabolic Diseases, 2012, vol 35, pages 279-286 *Susan E. Waisbren, Children's Hospital Boston, Boston, MA, USA*