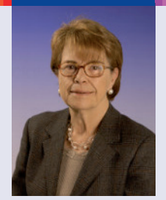
III POST SSIEM Roma Madrid 2016

5 NOVIEMBRE
2016



Hora: 09:00h.
Lugar: Casino de Madrid
C/ Alcalá, 15 | Madrid
Sala: Salón Real

Apreciados amigos!

Es un placer para mí, una vez más, poder coordinar la **III reunión científica post SSIEM 2016, que se celebrará en Madrid el próximo día 5 de Noviembre y que nos ofrece la oportunidad de, como en años anteriores, poder resumir el congreso SSIEM que tuvo lugar en Roma**, a todos aquellos profesionales interesados o que no pudieron asistir.

Los resúmenes que se os presentarán están divididos por los diversos errores congénitos del Metabolismo, dando a cada profesional una o varias vías metabólicas de las cuales considero que son expertos.

Cada uno de los ponentes va a exponer lo que ha considerado más novedoso y significativo de las diferentes charlas que tuvieron lugar durante el congreso, tanto Sesiones Plenarias, como Paralelas o Simposios.

Estoy segura de que también este año será todo un éxito y de gran interés y provecho para cada uno de nosotros.

Nos vemos en Madrid!!

Un saludo,
M. Pineda

III POST **AVALADO POR:**

SSIEM SOCIETY FOR THE STUDY OF INBORN ERRORS OF METABOLISM
AECOM
SEMI SOCIEDAD ESPAÑOLA DE MEDICINA INTERNA
Sociedad Española de Errores Innatos del Metabolismo

Solicitada Acreditación a la Comisión de Formación Continuada:

BIOMARIN ESPAÑA

Miniparc II . C/ Caléndula, 95 - Edf. O - 1ª Plta.
Of. 9-10 . 28109 Alcobendas (Madrid) **España**
T.: +34 91 827 81 63 **Fax**: +34 91 827 82 11
www.bmrn.com

BIO/EU/XX/XXX

Sponsored by:

BIOMARIN
BioMarin Europe Ltd.

AGENDA
5 NOVIEMBRE
2016 | MAD

III POST SSIEM Roma Madrid 2016

09:00 09:15 **PRESENTACIÓN**

Dr. David Gil Presidente AECOM
Dra. Antonia Ribes Council Member SSIEM
Dra. Mercé Pineda Coordinadora Post-SSIEM

09:00 09:15 **Sesión I** — *Dr. M. Lopez*, Hospital de la Cruz Roja (Madrid)

Adult Group Session
- *When shall we treat adult patients with cerebral ALD?*
- *When shall we treat adult patients with Krabbe disease and metachromatic leukodystrophy?*
- *Small molecules in neurometabolic diseases*

Free Communiations:
- *Nitisinone in alkaptonuria - quantifying the pigmentary pathway for the first time*
- *Challenges in the application of medical assistance in dying to patients with rare genetic diseases*
- *Olipudase alfa for the treatment of acid sphingomyelinase deficiency (ASMD): 18-month safety and efficacy data*
- *Late-onset Multiple Acyl-CoA Dehydrogenase Deficiency - a life threatening disease*
- *Two sibling pairs with metachromatic leukodytrophy*
- *Adult Niemann-Pick Type C mimicking Wilson Disease: the importance of oxysterols and lysosphingolipids pattern*

Industry Sponsored Symposia:
Shire *- Our Fabry Family Tree*

09:30 09:45 **Sesión II** — *Dr. P. Sanjurjo*, Hospital de Cruces (Bilbao)

Industry Sponsored Symposia:
Sigma-Tau *- Carnitine Deficiencies and Fatty Acid Oxidation*

Nutrition & Dietetics Meeting
- *Dietary Restriction in non-classical inborn errors of metabolism*
- *Side effects of dietary treatment in PKU, glycogen storeage disease and other hypoglycemias*
- *Diet after liver transplantation. Comparison of experience*
- *Lysine restricted diet and arginige supplementation in pyridoxine dependent epilepsy*
- *Mathematic modeling; what does it mean for the diet?*

09:45 10:00 **Sesión III** — *Dra. A. Belanger*, Hospital Ramón y Cajal (Madrid)

Parallel Session 1D: Nutricion and dietetics & miscellaneous
- *A double-blind placebo-controlled trial of triheptanoin in adult polyglucosan body disease*
- *Longitudinal study examining nutriotional status in children with organic acidaemias on a modular feed using a protein free module espcially developed for children with IMD*
- *Dietary treatment of 49 MSUD Italian patient*
- *Protein intake and physical activity are associated with body composition in patients with phenylalanine hydroxylase (PAH) deficiency*
- *Large neutral amino acid supplementation as a possible alternative treatment for adult PKU patients: evidence in PKU mice*
- *Mitochondrial fatty acid biosynthesis (mFASII) mdiates the substrate swithc in white skeletal muscle of very-long-chain acyl-CoA dehydrogenase-(VLCAD-/-)-deficient mices*

Industry Sponsored Symposia:
Orphan Europe *- Expert Viewpoints – Effective management of hyperammonaemia in organic acidaemias*

Parallel Session 4B: Phelylketonuria & neurotransmitters disorders
- *Secondary pterins alteration in patients with Phenylalanine Hydroxylase deficiency.*
- *Phenylalanine hydroxylase N-terminal domain is an allosteric binding site and can be target for pharmacological chaperone design*
- *The first structure of full-length phenylalanine hydroxylase has finally been determined*
- *Evaluation of long-term safety and efficacy of pegvaliase treatment for adults with phenylketonuria: updated year 4 results*
- *New generation of chemical scaffolds able to bind to human phenylalanine hydroxylase*
- *Clinical, biochemical and genetic approaches to improve the diagnosis of neurodevelopmental diseases related to neurotransmitter metabolism*

10:00 10:30 **Sesión IV** — *Dr. J. Rocha*, Centro Hospitalar do Oporto (Portugal)

Industry Sponsored Symposia:
BioMarin *- The Complexities of Adult PKU: Case Discussions*

10:30 10:45 **Sesión V** — *Dra. M.L. Couce*, Hospital Clínico Universitario (Santiago de Compostela)

Industry Sponsored Symposia:
BioMarin *- PKU in the Real World: Meeting the Needs of Diverse Patient Groups*

E-HOD Group Meeting

Parallel Session 2D: New therapies
- *Update Lecture: Gene therapy in hyperoxaluria*
- *Interim data from a randomized, placebo controlled, phase 1 study of ALN-AS1, and investigational RNAi therapeutic for the treatment of acute hepatic porphyria*
- *Effect of enzyme replacement therapy on osteoporosis in several CBS-deficient homocystinuric mouse models*
- *The utility of patient-derived hepatocytes for developing liver-directed therapies in propionic acidemia*
- *Aminoadipate semialdehyde synthase (AASS) as a therapeutic target for pysidoxine dependent epilepsy by substrate reduction*

Parallel Session 3B: New Therapies in Lysosomal Disorders
- *Update Lecture: Gene therapy in lysosomal storage diseases*
- *Long-term Outcomes with rhGUS ina a Phase I/II Clinical Trial in MPS VII*
- *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*
- *Intracerbroventcricular cerliponase alfa (BMN 190) in childrem with CLN2 disease: Results from a Phase 1/2, open-label, dose-escalation study*
- *Novel treatment for Fabry disease - IV administration of plant derived alpha-gal-a enzyme safety and efficacy, 1 year experience*

10:45 10:55 **Sesión VI** — *Dr. V. Rubio*, Hospital La Fe (Valencia)

Parallel Session 2A: Aminoacid and urea cycle disorders
- *Update lecture: Inherited disorders of proline metabolism*
- *A new metabolic disorder in human cationic amino acid transporter-2-mimicking arginase deficiency in newborn screening*
- *Minimal NTBC concentrations necessary to prevent formation of succinylacetone in Tyrosinemia type 1 patients*
- *Genetic cause and prevalence of hydroxiprolinemia*
- *Gain of function mutation in GLS1 causes infantile onset cataract and profund cognitive impairment*

10:55 11:05 **Sesión VII** — *Dr. I. Vitoria*, Hospital La Fe (Valencia)

Industry Sponsored Symposia:
Alexion *- Hypophosphatasia: Treatment options across the ages*

Opening Lecture
"Quo Vadis", redefinition of Inborn Metabloic Diseases, a challenge for the future

Industry Sponsored Symposia:
Alexion *- Emerging Novel Mutations: Implications In Lysosomal Acid Lipase Deficiency (LAL-D)*

11:05 11:15 **Sesión VIII** — *Dr. L. Aldámiz*, Hospital de Cruces (Bilbao)

Plenary Session 3: Therapeutical Chaperon Molecules and Beyond
- *New technologies to find new active molecules*
- *Cysteamine: A new scenario for an old molecule*
- *Samll molecules and clinical trials in Inborn Error of Metabolism*

Industry Sponsored Symposia:
Amicus *- Chaperone Therapy: Escorting Fabry Disease into a new era*

11:15 11:30 **Sesión IX** — *Dra. A. Ribes*, Hospital Clínic Centro de Diagonóstico Biomédico (Barcelona)

Plenary Session 1: "OMICS, Neonatal Screening and Beyond
- *Untargeted metabolomics in Inborn error of Metabolism*
- *Lipidomics and innate immunity*
- *New frontiers of neonatal screening*

Parallel Session 1C: Miscellaneous disorders
- *Multi-omics tools for the diagnosis and treatment of rare neurological disease*
- *Investigating applications of next generation sequencing in newborn screening*
- *A newborn screening method for Cerebrotendinous Xanthomatosis: data from a pilot validatioin study*
- *Progressive cognitive deterioration and pathological hallmarks in murine model creatine trasporter deficiency*
- *GNAO1 mutation: a new cause of transmembrane signalling derangement causing early onset movement disorder*
- *Developments in the diagnosis and treatment of PNPO deficiency*

Parallel Session 2C: Laboratory diagnosis of lysosomal disorders
- *Update Lecture: Contribution of tandem mass spectrometry to the diagnosis of lysosomal storage disorders*
- *A reliable multiplex mass spectrometry analysis of glyconsaminoglycans for mucopolysaccharidoses*
- *Increased collagen glycosylated hydroxylysine in the urine of MPS I, II and VI patients*
- *Identification of a new biomarker in Fabry disease by plasma proteomic analysis*
- *Low reliability of functional (enzymatic) diagnostics of lysosomal storage disorders in Dry Blood Spots (DBS) compared to fibroblasts*

11:30 12:00 | COFFEE BREAK

12:00 12:15 **Sesión X** — *Dr. G. Pintos*, Hospital Geman Trias i Pujol (Badalona)

Industry Sponsored Symposia:
Centogene *- Biomarkers in metabolic diseases – what is best for your patients in early diagnosis and long-term monitoring*

Parallel Session 3A: Methylation disorders
- *Update Lecture: What is new in methylation disorders*
- *Adenosylhomocysteine alters methylation of cellular RNA*
- *Interaction and characterization of the cblF (LMBD1) and cblJ (ABCD4) membrane proteins*
- *Disturbed regulation of methylenetetrahydrofolate reductase by S-adenosylmethionine*
- *The low-density lipoprotein (LDL) receptor-related protein 2 is essenctial for the exosome-dependent cerebral folate transport*

Parallel Session 4C: Lysosomal & autophagy disorders
- *Ammonia activates hepatic autphagy in vivo and its enhancement protects against acute and chronic hyperammonemia*
- *Initial, 24 weeks 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 IIIB*
- *Farber disease: acid ceramidase deficiency is more common than previously thought and slowly progressive disease may only be diagnosed in adulthood*
- *TAR-DNA binding protein 43 (TDP-43) pathology in Niemann Pick type C disease*
- *The emerging neurocognitive profile of classic infantile Pompe disease. J.M.P.*
- *The immunological basis of Vici syndrome*

12:15 12:35 **Sesión XI** — *Dra. A. García Cazorla*, Hospital Sant Joan de Deu (Barcelona)

I-NTD Group Meeting
E-IMD Group Meeting
metabERN Group Meeting

Plenary Session 2: "Celular Signaling"
- *Defective cellular signaling in mevalonate kinase deficiency*
- *Role of NO in urea cycle defects*
- *miRNA signaling in human diseases and in Inborn Errors of Metabolism*

Parallel Session 4A: Organic Acidurias
- *A fish model for propionic acidemia: increased survival and improvement of neurological phenotype by anaplerotic diet*
- *Medium term outcome of liver transplantation for children with propionic acidaemia*
- *Propionate anions, accumulated in Propionic Acidemia, affect cardiac excitation-contraction coupling, gene regulation and cellular growth, which may contribute to heart dysfunction*
- *Axonal peripheral neurpathy in propionic acidaemia: a severe side effect of long-term metranidazole treatment*
- *Stable isotope breath testing to assess in vivo metabolite flux in methylmalonic acidemia (MMA). From mouse models to patients*
- *Insights into disease mechanisms of cblA-type methylmalonic aciduria from 67 new patients and functional MMAA missense mutation characterization*

Plenary Session 6: Cellular Trafficking
- *Disorders of intracellular trafficking*
- *Autophagy, inborn errors of metabolism and human diseases*
- *Autophagy and mitochondrial disorders*

12:35 12:50 **Sesión XII** — *Dr. D. Gonzalez-Lamuño*, Hospital Marqués de Valdecilla (Santander)

Industry Sponsored Symposia:
Vitaflo *- Nutritional Choices in IEM*

Parallel Session 1B: Glycosylation and carbogydrate disorders
- *The development and validation of a semi-automated enzyme panel for muscle glycolytic disorders*
- *A Conserved phosphatase destroys toxic glycolytic side-products in mammals and yeasts*
- *Is G6PC3, the enzyme deficient in severe congenital neutropenia type 4, really a glucose-6-phosphatase?*
- *A novel sugar metabolic pathway in human: ISPD synthesises CDP-ribitol.*
- *ISPD produces CDP-ribitol used by FKTN and FKRP to transfer ribitol-phosphate onto alfa-dystroglycan*
- *NANS-mediated synthesis of sialic acid is required for brain and skeletal development.*

Parallel Session 3C: Glycosylation and cellular network disorders
- *Update Lecture: What is new in CDGs*
- *A novel group of metabolic disorders due to tissue-specific defects in V-ATPase assembly*
- *Toward a folding therapy for PMM2-CDG*
- *MAGT1-deficiency: new insights into a controversial protein with a key role in N-glycosylation*
- *SLC39A8 deficiency is a novel treatable disorder of manganese metabolism and glycosylation*

12:50 13:05 **Sesión XIII** — *Dra. M. O'Callaghan*, Hospital Sant Joan de Deu (Barcelona)

Parallel Session 1A: Mitochondrial and fatty acid oxidation disorders
- *Characterizing the molecular architecture of mitochondrial energy metabolism apparatus*
- *Decreased stability of the OCTN2 carnitine transporter in patients with primary carnitine deficiency*
- *Cellular models for medium-chain acyl-CoA dehydrogenase deficiency based on induced pluripotent stem cell technology*
- *Clinical exome sequencing in 900 index cases: diagnostic rate and new disease genes*
- *Cytosolic phosphoenolpyruvate carboxykinase deficiency presenting wit acute liver failure following gastroenteritis*
- *Urinary organic acids in paediatric single mitochondrial DNA deletion disorders.*

Parallel Session 2B: Mitochondrial disorders
- *Update lecture: New treatments in mitochondrial disorders*
- *Update of combined D, L-2-Hydroxyglutaric aciduria: new cases and restoration of the defect in vitro*
- *TANGO2 deficiency, a novel neurometabolic disorder with recurrent encephalo-cardio-myopathic crises*
- *Decanoic acid treatment of fibroblasts from patients with nuclear-encoded complex I deficient Leigh syndrome: a step towards personalised medicine?*
- *A novel causative gene of mitochondrial respiratory chain disorders in an apparent life-threating event (ALTE)*

Industry Sponsored Symposia:
Actelion *- Improving our understanding of a rare disease: collective efforts expand our horizons in Niemann-Pick disease Type C*

13:05 13:15 **Sesión XIV** — *Dr. J. Aguirre*, Hospital Torrecardenas (Almeria)

Industry Sponsored Symposia:
Shire *- New directions in Gaucher disease management*

Plenary Session 4: Liver Directed Therapies in IEM
- *Liver transplantation*
- *Gene therapy*
- *Treatment of molybdenum cofactor deficiency*

13:15 13:30 **Sesión XV** — *Dra. M. del Toro*, Hospital Vall d'Hebron (Barcelona)

Industry Sponsored Symposia:
Sanofi-Genzyme *- The Impact of New Initiatives in Rare Disease Diagnostics*

Plenary Session 5: Clinical Novelties
- *Inherited disorders of metal metabolism*
- *Hyperinsulinisms*
- *New therapeutical challenges in lysosomal storage disorders*

Late Breaking News
- *Bialleic mutations in IARS, encoding cytosolic isoleucyl-tRNA sythetase, cause growth retardation with prenatal onset, itellectual disability, muscular hypotonia, and infantile hepatopathy*
- *CAD Mutations and Uridine-Responsive Epileptic Encephalopathy*
- *TBCE mutations cause early-onset progresive encephalopathy with distal spinal muscular atrophy*
- *MAP17 is necessary for transport activity of SGLT2: a novel cause of renal glucosuria*

Komrower Lecture
- *Mitochondrial Disorders, a journey through the magic circle and beyond*

Garrod Award Lecture
- *Paediatric single mitochondrial DNA delection disorders: an overlapping spectrum of disease*

13:30 14:00 **Sesión XVI** — *Dra. M. Pineda*, Hospital Universitario 12 Octubre (Madrid)

Industry Sponsored Symposia:
BioMarin *- Diagnose CLN2 Disease Earlier: Connecting the dots between Genetics and Neurology*

13:30 14:00 **Sesión XVII** — *Dr. P. Harmatz*, Oakland Children Hospital - CA, USA

Industry Sponsored Symposia:
BioMarin *- Long-Term treatment outcomes and impact on natural disease course - Esperience in Morquio A and Maroteaux-Lamy*

III POST SSIEM Roma Madrid 2016