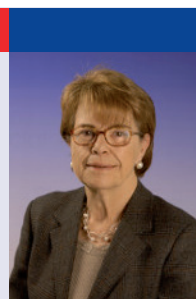


5 NOVIEMBRE
2016



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

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AGENDA

5 NOVIEMBRE

2016 | MADRID



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 Adult Group Session <ul style="list-style-type: none"> 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: <ul style="list-style-type: none"> 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 leukodystrophy Adult Niemann-Pick Type C mimicking Wilson Disease: the importance of oxysterols and lysosphingolipids pattern Industry Sponsored Symposia: Shire - Our Fabry Family Tree	Dr. M. Lopez Hospital de la Cruz Roja (Madrid)
09:30 09:45	Sesión II Industry Sponsored Symposia: Sigma-Tau - Carnitine Deficiencies and Fatty Acid Oxidation Nutrition & Dietetics Meeting <ul style="list-style-type: none"> Dietary Restriction in non-classical inborn errors of metabolism Side effects of dietary treatment in PKU, glycogen storage 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? 	Dr. P. Sanjurjo Hospital de Cruces (Bilbao)
09:45 10:00	Sesión III Parallel Session 1D: Nutricion and dietetics & miscellaneous <ul style="list-style-type: none"> A double-blind placebo-controlled trial of triheptanoin in adult polyglucosan body disease Longitudinal study examining nutritional status in children with organic acidaeimias on a modular feed using a protein free module espically 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 mice Industry Sponsored Symposia: Orphan Europe - Expert Viewpoints – Effective management of hyperammonaemia in organic acidaeimias Parallel Session 4B: Phelylketonuria & neurotransmitters disorders <ul style="list-style-type: none"> Secondary pterins alteration in patients with Phenylalanine Hydroxylase deficiency. Phenylalanine hydroxylase N-terminal domain is an allosteric binding site and can be target for pharmacologi-cal 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 	Dra. A. Belanger Dra. S. Mihaela Stanescu Hospital Ramón y Cajal (Madrid)
10:00 10:30	Sesión IV Industry Sponsored Symposia: BioMarin - The Complexities of Adult PKU: Case Discussions	Dr. J. Rocha Centro Hospitalar do Oporto (Portugal)
10:30 10:45	Sesión V 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 <ul style="list-style-type: none"> 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 <ul style="list-style-type: none"> 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-iduro-nidase and iduronate-2-sulfatase cDNAs from the albumin locus Intracerebroventricular 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 	Dra. M.L. Couce Hospital Clínico Universitario (Santiago de Compostela)
10:45 10:55	Sesión VI Parallel Session 2A: Aminoacid and urea cycle disorders <ul style="list-style-type: none"> 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 profound cognitive impairment 	Dr. V. Rubio Hospital La Fe (Valencia)

10:55 11:05	Sesión VII Industry Sponsored Symposia: Alexion - Hypophosphatasia: Treatment options across the ages Opening Lecture "Quo Vadis", redefinition of Inborn Metaboloic Diseases, a challenge for the future Industry Sponsored Symposia: Alexion - Emerging Novel Mutations: Implications In Lysosomal Acid Lipase Deficiency (LAL-D)	Dr. I. Vitoria Hospital La Fe (Valencia)
11:05 11:15	Sesión VIII Plenary Session 3: Therapeutical Chaperon Molecules and Beyond <ul style="list-style-type: none"> 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	Dr. L. Aldámiz Hospital de Cruces (Bilbao)
11:15 11:30	Sesión IX Plenary Session 1: “OMICs, Neonatal Screening and Beyond <ul style="list-style-type: none"> Untargeted metabolomics in Inborn error of Metabolism Lipidomics and innate immunity New frontiers of neonatal screening Parallel Session 1C: Miscellaneous disorders <ul style="list-style-type: none"> 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 validation 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 <ul style="list-style-type: none"> 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 	Dra. A. Ribes Hospital Clínic Centro de Diagnóstico Biomédico (Barcelona)

11:30 12:00 | COFFEE BREAK

12:00 12:15	Sesión X 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 <ul style="list-style-type: none"> Update Lecture: What is new in methylation disorders Adenosylhomocysteine alters methylation of cellular RNA Interaction and characterization of the cb1F (LMBD1) and cb1J (ABCD4) membrane proteins Disturbed regulation of methylenetetrahydrofolate reductase by S-adenosylmethionine The low-density lipoprotein (LDL) receptor-related protein 2 is essential for the exosome-dependent cerebral folate transport Parallel Session 4C: Lysosomal & autophagy disorders <ul style="list-style-type: none"> 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 progres-sive 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 	Dr. G. Pintos Hospital Geman Trias i Pujol (Badalona)
12:15 12:35	Sesión XI I-NTD Group Meeting E-IMD Group Meeting metabERN Group Meeting Plenary Session 2: “Celular Signaling” <ul style="list-style-type: none"> 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 <ul style="list-style-type: none"> 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 acidemia Propionate anions, accumulated in Propionic Acidemia, affect cardiac excitation-contraction coupling, gene regulation and cellular growth, which may contribute to heart dysfunction Axonal peripheral neuropathy in propionic acidemia: 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 cb1A-type methylmalonic aciduria from 67 new patients and func-tional MMAA missense mutation characterization Plenary Session 6: Cellular Trafficking <ul style="list-style-type: none"> Disorders of intracellular trafficking Autophagy, inborn errors of metabolism and human diseases Autophagy and mitochondrial disorders 	Dra. A. García Cazorla Hospital Sant Joan de Deu (Barcelona)

12:35 12:50	Sesión XII Industry Sponsored Symposia: Vitafo - Nutritional Choices in IEM Parallel Session 1B: Glycosylation and carbogydrate disorders <ul style="list-style-type: none"> 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 <ul style="list-style-type: none"> 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 	Dr. D. Gonzalez-Lamuño Hospital Marqués de Valdecilla (Santander)
12:50 13:05	Sesión XIII Parallel Session 1A: Mitochondrial and fatty acid oxidation disorders <ul style="list-style-type: none"> 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 caused 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 <ul style="list-style-type: none"> 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-threatening event (ALTE) Industry Sponsored Symposia: Actelion - Improving our understanding of a rare disease: collective efforts expand our horizons in Niemann-Pick disease Type C	Dra. M. O'Callaghan Hospital Sant Joan de Deu (Barcelona)
13:05 13:15	Sesión XIV Industry Sponsored Symposia: Shire - New directions in Gaucher disease management Plenary Session 4: Liver Directed Therapies in IEM <ul style="list-style-type: none"> Liver transplantation Gene therapy Treatment of molybdenum cofactor deficiency 	Dr. J. Aguirre Hospital Torrecardenas (Almería)
13:15 13:30	Sesión XV Industry Sponsored Symposia: Sanofi-Genzyme - The Impact of New Initiatives in Rare Disease Diagnostics Plenary Session 5: Clinical Novelties <ul style="list-style-type: none"> Inherited disorders of metal metabolism Hyperinsulinisms New therapeutical challenges in lysosomal storage disorders Late Breaking News <ul style="list-style-type: none"> Biallelic 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 <ul style="list-style-type: none"> Mitochondrial Disorders, a journey through the magic circle and beyond Garrod Award Lecture <ul style="list-style-type: none"> Paediatric single mitochondrial DNA deletion disorders: an overlapping spectrum of disease 	Dra. M. del Toro Hospital Vall d'Hebron (Barcelona)
13:30 14:00	Sesión XVI Industry Sponsored Symposia: BioMarin - Diagnose CLN2 Disease Earlier: Connecting the dots between Genetics and Neurology	Dra. M. Pineda Hospital Universitario 12 Octubre (Madrid)
13:30 14:00	Sesión XVII Industry Sponsored Symposia: BioMarin - Long-Term treatment outcomes and impact on natural disease course - Esperience in Morquio A and Maroteaux-Lamy	Dr. P. Harmatz Oakland Children Hospital - CA, USA

Aforo limitado. Se ruega confirmar asistencia en este e-mail: postssiem2016@skylineventos.com