Thursday September 10

| | |
|---|---|
| 2.00 pm | Opening of registration |
| 6.00 pm | Get together |
| 7.30. pm | Dinner |
| 9.00 pm | Opening / Welcome address |
| 9.15 pm | Opening lecture |
| | Peter Lobel |
| | Lysosomes, proteomics and disease |

Friday September 11

**8.15 am - 10.10 am Scientific session I : Cell biology of lysosomes**

| | |
|---|---|
| 8.15 am - 8.35 am | Sandra Pohl, Marisa Encarnacao, Thomas Braulke |
| | GlcNac-1-phosphotransferase: Key enzyme in the M6P-dependent transport of lysosomal enzymes |
| 8.35.am – 8.55 am | Katrin Kollmann, Anna Katharina Strohwald, Sandra Pohl, Stephan Storch, Torben Lübke, Thomas Braulke |
| | Generation of a novel knock-in mouse model for mucolipidosis II alpha/beta |
| 8.55 am – 9.15 am | Krista Lakomek, Matthias Kettwig, Achim Dickmanns, Ralf Ficner, Torben Lübke |
| | Characterization of a novel putative acid amidase of the lysosome |
| 9.15 am – 9.35 am | Bernd Schröder, Christian Wrocklage, Andrej Hasilik, Paul Saftig |
| | Novel lysosomal membrane proteins: Molecular characterization of transmembrane protein 192 (TMEM192) and disrupted in renal carcinoma (DIRC2) |
| 9.35 am – 9.55 am | Frank Rutsch, Susann Gailus et al |
| | Identification of the gene for the cblf defect of vitamin B12 metabolism reveals a novel lysosomal membrane protein with homology to lipocalin receptors |
| 9.55 am –10.10 am | Corinne Sagne, Samantha Papal, Frank Rutsch, Bruno Gasnier Functional characterization of LMBD 1, the protein defective in vitamin B12 storage disease |
| 10.10 am – 10.40 am | Coffee break |

**10.40 am to 12.20 am Scientific session II Neuronal Ceroid Lipofuscinosis**

| | |
|---|---|
| 10.40 am – 11.00 am | S. Storch, S. Herder et al. |
| | Lysosomal transport of CLN7/MFSD8 is mediated by both dileucine and tyrosine based sorting signals |
| 11.00 am- 11.20 am | Robert Steinfeld, Aritra Pal, Ralf Krätzner, Jutta Gärtner, Georg Sheldrick |
| | The molecular basis of late infantile neuronal ceriod lipofuscinosis is substantiated by the crystal structure of trpeptidyl peptidase 1 (TPP1) |
| 11.20 am – 11.40 am | Sara Mole, Sandra Codlin, Rebecca Haines, Claudia Kitzmüller |
| | Using Schizosaccharomyces to study Batten disease |
| 11.40 am – 12.00 am | A. Jalanko, A Kyttälä, von Schantz C, Schmiedt ML et al. |

| | |
|---|---|
| | Finnish variant of neuronal ceroid lipofuscinosis: new insights into the CLN5 protein, molecular interactions and disease process |
| 12.00 am- 12.20 am | Sarah Pressey, David Smith, Frances M Platt, Jonathan Cooper<br><br>Comparing the pathogenesis of Niemann Type C, Sandhoff disease and neuronal ceroid lipofuscinosis |
| 12.20 pm to 1.50 pm | Lunch |
| **1.50 pm to 3.30 pm** | **Scientific Session III : Pathophysiology of lysosomal storage diseases** |
| 1.50 pm- 2.10 pm | Idit Ron, Mia Horowitz<br><br>ER associated Degradation in Gaucher disease: its possible implication to the development of Parkinsons disease |
| 2.10 pm – 2.30 pm | Oprisoreanu AM, Sidransky, E, Horwitz, M, Sandhoff R., Harzer, K, Sauerbruch T, Yildiz, Y.<br><br>Role of GBA2 in Morbus Gaucher |
| 2.30 pm – 2.50 pm | Thomas Kuehl, Janos Groh, Antje Kroner, Rudolf Martini, Jon Cooper<br><br>Pathogenic impact of immune related cells in neuronal ceroid lipofuscinosis |
| 2.50 pm - 3.10 pm | Langeveld M, Hollak, CEM, Aerts, JM<br><br>Glycosphingolipids and insulin resistance |
| 3.10 pm to 3.30 pm | Jean Michel Heard, Sandrine Vitry, Michael Hocquemiller, Stephanie Bigou, Jerome Aussiel<br><br>Biogenesis of abnormal lysosomes in MPSIIIB neurons |
| 3.30 pm – 4.00 pm | Coffee break |
| 4.00 pm- 5.00 pm | **Scientific Session IV : Pathophysiology of lysosomal storage diseases** |
| 4.00 pm- 4.20 pm | David A. Zeevi, Ayala Frumkin, Vered Offen-Glasner, Aviram Kogot-Levin, Gideon Bach<br><br>A potentially dynamic lysosomal role for the endogenous TRPML proteins |
| 4.20 pm – 4.40 pm | Alexander Pshezhetsky, Volkan Seyrantepe, Aleksander Hinek et al<br><br>Novel role of lysosomal carboxypeptidase cathepsin A/Protective protein in regulation of vasoconstrictiona dn elastogenesis. |
| 4.40 pm- 5.00 pm | Elina Kuokkanen, Hilde Mf Riise-Stensland, Oivind Nilssen, Pirkko Heikinheimo<br><br>Intracellular targetting of lysosomal a-mannosidase variants associated with a-mannosidosis. |
| . | |
| 5.00 pm to 6.30 pm | Attended Poster session |
| 7.00 pm | Dinner |
| 9.00 pm | Bryan Winchester<br><br>History of the ESGLD |

Saturday September 12

| | |
|---|---|
| **8.30 am- 9.10 am** | **Scientific Session IV : Pathophysiology of lysosomal storage diseases / continued** |
| 8.30 am to 8.50 am | Korolenko TA, Cherkanova, MS, Filatova, TG, Bravve IY<br>Chitotriosidase, MMP activity, cystatin C and intralysosomal storage of lipids in atherosclerosis |
| 8.50 am – 9.10 am | Rebecca van Zyl, Volkmar Gieselmann, Matthias Eckhardt<br>Sulfatide with C18:0 fatty acid causes lethal audiogenic seizures in transgenic mice |
| **9.10 am- 10.30 am** | **Scientific Session V : Therapy of lysosomal storage diseases** |
| 9.10 am – 9.30 am | Caroline Sevin, Francoise Piguet, Marie Vanier et al<br>Intracerebral gene therapy for metachromatic leukodystrophy |
| 9.30 am- 9.50 am | Delai S, Visigalli I, Stok M, et al<br>Therapeutic efficacy of hematopoetic stem cell gene therapy for Hurler Type 1 mucopolysaccharidosis |
| 9.50 a.m 10.10 a.m | Fiora Wilkinson , Alex Langford Smith, William Bennet et al<br>Lentiviral Vector mediated stem cell gene therapy for MPSIIIA |
| 10.10 a.m – 10.30 a.m | Angela Gritti, Annalisa Lattanzi, Claudio Maderna et al.<br>Direct intra brain lentiviral-mediated gene delivery leads to widespread transgene distribution and high level enzymatic correction in the whole CNS of twitcher mice |
| 10.30 am to 11.00 a.m | Coffee break. |
| **11.00 am- 12.15 am** | **Scientific Session VI : Therapy of lysosomal storage diseases** |
| 11.00 am - 11.15 am | Andrew Wong, Sarah Ahamdi, Ahad Rim et al<br>Neural stem cell and viral vector therapies for neuronal ceroid lipofuscinosis |
| 11.15 am –11.30 am | Frank Matthes, Stephan Schröder et al<br>Transendothelial trasnfer of arylsulfatase A in a cell culture model of the blood brain barrier |
| 11.30 am – 11.45 am | David Aviezer, Yoseph Shaaltiel, Raul Chertkoff et al<br>Novel enzyme replacement therapy for Gaucher disease: On Going Phase II clinical trials with recombinant human glucocerebrosidase expressed in plant cells |
| 11.45 am- 12.00 am | DP Germain et al<br>Long Term Safety, Tolerability and Pharmacodynamic Data for AT1001, a pharmacological Chaperone for the treatment of Fabry disease |
| 12.00 am – 12.15 am | Thomas Lemmonier, Stephane Blanchard, Diana Toli, Jean Michel Heard, Delphine Bohl<br>Induced pluripotent stem cells from adult skin fibroblasts of MPSIIIB human patients |
| 12.15 pm to 1.45 pm | Lunch |
| 2.00 pm to 6.00. pm | Social program (Cologne Cathedral or Wine Festival)<br>(Busses leave at 2.00 pm) |
| 6.00 pm to 7.30 pm | Attended Poster session |
| 8.00 pm | Gala Dinner |

Sunday September 13

| | |
|---|---|
| 8.30 am to 10.30 am | **Scientific session VII Therapy pf lysosomal storage diseases** |
| 8.30 am – 8.50 am | James Dodge, Seng Cheng<br><br>Efficacy of intracerebroventricular infusion of lysosomal enzymes at correcting the the CNS manifestations in mouse models of neuronopathic lysosomal storage disorders. |
| 8.50 am- 9.10 am | Kenneth Valenzano, Richie Khanna, John Flanagan, et al<br><br>Exploring the combination of pharmacological chaperones and ERT as a potential therapeutic strategy for lysosomal storage diseases. |
| 9.10 am- 9.30 am. | Ullrich Matzner, Frank Matthes, Annika Böckenhoff et al<br><br>Enzyme replacement therapy of metachromatic leukodystrophy: CNS effects in the arylsulfatase A deficient aggravated mouse model |
| 9.30 am – 9.50 am | Carla M Hollak, Wilam Donker, Johann E Groener et al<br><br>Progressive Gaucher disease due to neutralizing antibodies: succesful treatment with rituximab (anti-CD20) followed by high dose imiglucerase |
| 9.50 am – 10.10 am | Marchesan D, Deegan PB, Cox T<br><br>Effect of serum and cell type on mannose-6-phosphate mediated delivery of enzyme therapy in cell models of Fabry disease |
| 10.10 am – 10.30 am | Bruno Bembi, Marco Confalonieri, Giovanni Ciana, et al<br><br>Enzyme replacement therapy with a-glucosidase alfa in late onset phenotype of glycogenosis type 2 |
| 10.30 am to 11.00 am | Coffee break |
| 11.00 am to 12.40. am | **Scientific session VII Clinical aspects of lysosomal storage diseases** |
| 11.00 am – 11.20 am | OP van Diggelen, LF Oemardien, AM Boer, I de Graaf, C Weykamp<br><br>Quality Assessment of enzyme analysis for lysosomal storage disorders: Four years QA-LSD, European pilots with 36-59 participants |
| 11.20 am – 11.40 am | Krägeloh Mann I ,Kehrer C, Kustermann-Kuhn B, Groeschel S Grodd W<br><br>Clinical course description of metachromatic leucodystrophy in children using standardized motor and neuroimaging scores |
| 11.40 am – 12.00 am | Rombach SM, Dekker N, Linthorst GE, et al<br><br>Elevated lysoceramidetrihexoside in Fabry disease: a new biomarker |
| 12.00 am – 12.15 am | Stephan vom Dahl, Ludger Wilhelm Poll, Dieter Häussinger, Ulrich Mödder<br><br>MRI bone marrow findings in non-neuronopathic Gaucher disease: Patterns, determinants and relationship with severe skeletal complications in 63 adult patients – 13 years of experience from a single center |
| 12.15 am – 12.30 am | Quirine Teunissen, J ohanna van der Lee, Carla Hollak et al<br><br>International consensus on a mucopolysaccharidosis type I phenotype severity scale |
| 12.30 pm to 1.00 pm | Young scientists award |
| 1.00 pm – 2.00 pm | Lunch and departure |