

PROGRAM
12th European Conference On Rare Diseases
“Living With A Rare Disease”
Spała 2014

Friday, July 04, 2014

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| Friday, July 04, 2014 | | |
| Breakfast | | 7:30 - 8:30 |
| | Registration | 8:00 - 9:00 |
| Teresa Matulka Anna Tylki-Szymańska <i>Warsaw, Poland</i> | Welcome Address | 9:00 - 9:30 |
| Session I | | |
| Chairs: | Grzegorz Węgrzyn, Ekaterina Zakharova | |
| Jörn Oliver Sass <i>Zürich, Switzerland</i> | Inborn Errors of Cobalamin Transport and Metabolism | 9:30-9:50 |
| Ladislav Kuchar <i>Prague, Czech Republic</i> | Tandem mass spectrometry: new possibilities for research and diagnosis of lysosomal storage disorders | 9:50-10:10 |
| Gajja S Salomons <i>Amsterdam, Netherlands</i> | Update on the D-2- and L-2-Hydroxyglutaric Acidurias | 10:10-10:30 |
| Johannes Berger <i>Vienna, Austria</i> | X-linked Adrenoleukodystrophy: From the basic research to possible therapeutic strategies. | 10:30-10:50 |
| Coffee break | | 10:50-11:20 |
| Session II | | |
| Chairs: | Maciej Machaczka, Jana Ledvinova | |
| Stefan Karlsson <i>Stockholm, Sweden</i> | Development of gene therapy in a mouse model for Gaucher disease | 11:20-11:50 |
| Hanna Rosenbaum <i>Haifa, Israel</i> | Gaucher disease: clinical course, diagnosis and innovation in treatment. | 11:50-12:10 |
| Jana Ledvinova <i>Prague, Czech Republic</i> | Diagnosis of lysosomal storage disorders in the Czech Republic | 12:10-12:30 |
| Nataliia Pichkur <i>Kyiv, Ukraine</i> | Hyperphenylalaninemia: bipterins deficiency | 12:30-12:45 |
| Lunch | | 12:45-14:00 |
| Session III | | |
| Chairs: | Joern Olivier Sass, Johannes Berger | |
| David J. Begley <i>London, UK</i> | Delivering enzyme replacement therapy for mucopolysaccharidosis and other lysosomal storage disorders to the CNS across the blood-brain barrier. | 14:00-14:20 |
| Ulrich Matzner <i>Bonn, Germany</i> | Advances in therapies for metachromatic leukodystrophy | 14:20-14:40 |
| Wioletta Kobiela <i>Gdansk, Poland</i> | Evaluation of the efficacy of different therapeutic approaches in the treatment of MPS I | 14:40-15:00 |
| Agnieszka Ługowska <i>Warsaw, Poland</i> | Does apolipoprotein E genotype and LRP1 polymorphisms influence the phenotype in patients with different clinical types of metachromatic leukodystrophy ? | 15:00-15:15 |
| Katarzyna Hetmańczyk <i>Warsaw, Poland</i> | Dipeptidyl peptidase-IV: a new biomarker for diagnosing of mucopolysaccharidoses – preliminary results | 15:15-15:25 |

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| Karl-Eugen Mengel <i>Mainz, Germany</i> | Diagnosis, manifestation and management of GM1-Gangliosidosis in Germany | 15:25-15:45 |
| Shunji Tomatsu <i>Wilmington, USA,</i> | Newborn screening and diagnosis of mucopolysaccharidoses | 15:45-16:05 |
| Coffee break | | 16:05 -16:30 |
| Session IV | | |
| Chairs | Michael Beck, Rafał Płoski | |
| Grzegorz Węgrzyn <i>Gdansk, Poland</i> | Gene Expression-Targeted Isoflavone Therapy: Facts, Controversies and Further Possibilities | 16:30-16:50 |
| Maurizio Scarpa <i>Wiesbaden, Germany</i> | Pathophysiology of neurodegeneration in Lysosomal Storage Diseases and approaches to therapy of the CNS | 16:50-17:10 |
| Maciej Machaczka <i>Stockholm Sweden</i> | Thirty years' experience of allogeneic hematopoietic stem cell transplantation for inborn errors of metabolism in Sweden | 17:10-17:30 |
| Krzysztof Kałwak <i>Wroclaw, Poland</i> | Hematopoietic stem cell transplantation in metabolic disorders. What's new in the world and in Poland? | 17:30-17:50 |
| Grill-dinner | | 20:00 |
| Saturday , July 5, 2014 | | |
| Breakfast | | 7:30 - 8:30 |
| Session V | | |
| Chairs | Gajja Salomons, Agnieszka Ługowska | |
| Rafał Płoski <i>Warsaw, Poland</i> | Whole exome sequencing for diagnosis of rare diseases - 1,5 year experience | 8:30-8:50 |
| Paulina Jędrak <i>Gdansk, Poland</i> | The significance of the level of mitochondrial DNA in cells of patients suffering from Huntington's disease | 8:50-9:10 |
| Barbara Zapala <i>Krakow, Poland</i> | A novel <i>TAZ</i> gene mutation and maternal mosaicism in Polish family with Barth syndrome | 9:10-9:30 |
| Hanna Mierzewska <i>Warsaw, Poland</i> | MRI in diagnostics of rare diseases with the nervous system involvement. | 9:30-9:50 |
| Coffee break | | 9:50-10:20 |
| Session VI | | |
| Chairs | Maurizio Scarpa, Anna Tyłki-Szymańska | |
| Michael Beck <i>Mainz, Germany</i> | The Center for Lysosomal Storage Disorders at the University of Mainz (Villa Metabolica): A model for a Center of Rare Diseases | 10:20-10:50 |
| Christina Lampe <i>Wiesbaden, Germany</i> | Transition of Adolescents in Adulthood | 10:50-11:10 |
| Representatives of the state administration | Rare diseases in Poland | 11:10-11:50 |
| Roundtable meeting | Representatives of the state administration - representatives of patient organizations - <i>Knights' Hall</i> | 11:50 -13:00 |
| Arndt Rolfs <i>Rostock, Germany</i> | New insights in the diagnosis and treatment of Gaucher disease | 11:50-12:10 |
| Ivanka Sinigerska <i>Sofia, Bulgaria</i> | Biochemical Diagnosis of Lysosomal Storage Diseases in Bulgaria | 12:10-12:30 |

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| Radka Tincheva <i>Sofia, Bulgaria</i> | Healthcare of children with treatable rare diseases in Bulgaria" | 12:30-12:50 |
| Chris Hendriksz <i>Manchester, UK</i> | Enzyme Replacement therapy for Morquio (MPS IVA) | 12:50-13:10 |
| Lunch break | | 13:00 - 14:00 |
| Session VII Dysmorphology meeting | | |
| Chairs | Małgorzata Krajewska-Walasek, Ewa Obersztyn | |
| Aleksandra Jezela-Stanek <i>Warsaw, Poland</i> | What dysmorphologists should keep in mind during evaluation of patients with blepharophimosis and mental disability? | 14:10-14:30 |
| Małgorzata Krajewska-Walasek <i>Warsaw, Poland</i> | Genitopatellar syndrome contra Ohdo-like /SBBYS syndrome: two disorders – one gene. Understanding clinical similarities and differences according to known <i>KAT6B</i> defects | 14:30-14:50 |
| Robert Śmigiel <i>Wroclaw, Poland</i> | Ohdo syndrome caused by mutation in <i>KAT6B</i> gene - case report and phenotype analysis on the basis of the literature | 14:50-15:05 |
| Jacek Pilch <i>Katowice, Poland</i> | Microdeletion 2q31.1-32.2 in a boy with blepharophimosis and mental retardation – a phenotype analysis | 15:05-15:20 |
| Coffee break | | 15:20 - 15:40 |
| Moderators: Małgorzata Krajewska-Walasek, Robert Śmigiel | Presentations of unknown cases | 15:40 -17:50 |
| Gala Dinner | | 20:00 |
| Sunday , July 6, 2014 | | |
| Breakfast | | 7:30 – 8:30 |
| Session VIII | | |
| Chairs | Zita Krumina, Anna Latos-Bieleńska | |
| Zita Krumina <i>Riga, Latvia</i> | Difficulties in diagnosis of rare genetic disorders in Latvia | 9:10-9:30 |
| Julia Zakharchuk <i>Chelyabinsk, Russia</i> | The clinical case of MPS VI type Maroteaux – Lamy | 9:30-9:50 |
| Aleksandra Żurowska <i>Gdansk, Poland</i> | Do we have a Wonder Drug for atypical hemolytic-uremic syndrome ? | 9:50-10:10 |
| Olga Cirstea <i>Chisinau, Moldova</i> | Wegener's Granulomatosis in children: Case report and the need for a multidisciplinary approach | 10:10-10:30 |
| Svetlana Volgina <i>Kazan, Russia</i> | Fabry disease in the children: from the onset of clinical symptoms until diagnosis (speech in Russian) | 10:30-10:50 |
| Coffee break | | 10:50 - 11:20 |
| Session IX | | |
| Arunas Valiulis <i>Vilnius, Lithuania</i> | Alpha-1 Antitrypsin Deficiency Network as Pattern of Management of Rare Disease in Central - Eastern Europe" | 11:20-11:40 |
| Anna Tylki-Szymańska <i>Warsaw, Poland</i> | Inborn errors of metabolism – manifestation in newborns | 11:40-12:00 |
| Ekaterina Zakharova <i>Moscow, Russia</i> | Undiagnosed patient | 12:00-12:20 |
| Teresa Matulka Anna Tylki-Szymańska <i>Warsaw, Poland</i> | Summary of the conference | 12:20-13:00 |

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| Lunch | 13:00 - 14:00 |
| Medical consultations | 9:00 - 16:30 |
| Holy Mass in intention of patients with rare diseases and their families | 17:00 - 18:00 |
| Dinner | 19:00 |
| Monday, July 7, 2014 | |
| Breakfast | 8:00 - 9:00 |
| Transfer to the airport | |

Session for Patients, Parents and Care givers

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| Saturday , July 5, 2014 | | |
| Breakfast | | 8:30 - 9:30 |
| Session for Patients, Parents and Care givers | | |
| Chairs | Anna Pruszyńska, Paulina Zalewska | |
| Barbara Czartoryska <i>Warsaw, Poland</i> | The role of the laboratory in the diagnosis of lysosomal storage diseases | 9:30-10:00 |
| Jolanta Wierzba <i>Gdańsk, Poland</i> | Why children are unique genetic. | 10:00-10:30 |
| Coffee break | | 11:00 - 12:00 |
| Jolanta Marucha <i>Warsaw, Poland</i> | The role of physiotherapy in progressing dysfunction of musculoskeletal system in patients with selected metabolic diseases | 12:00-12:30 |
| Beata Prorok <i>Łódź, Poland</i> | Palliative care of patients with rare diseases | 12:30-13:00 |
| Lunch | | 14:00 - 15:00 |
| Maciej Machaczka <i>Stockholm Sweden</i> | Karolinska experience in allogeneic hematopoietic stem cell transplantation for inherited metabolic disorders | 15:00 - 15:30 |
| Tylki Szymańska Anna <i>Warsaw, Poland</i> | Living with a rare disease | 15:30 - 16:00 |
| Summary and the end of the conference | | 16:00 - 16:20 |
| <i>Medical consultations, optical examinations</i> | | 16:20 - 18:00 |
| Gala Dinner | | 20:00 |
| Monday, July 7, 2014 | | |
| Breakfast | | 8:00 - 9:00 |
| Medical consultations - paediatric, metabolic, genetic, neurological, psychological, orthopaedic and genetic oncology | | 9:00-16:00 |