Thursday, May 18, 2017

08:45 - 09:00  Welcome and introduction to Moscow (Elena Lukina)

09:00 - 09:45  Opening Lecture “Lysosomes and lysosomal storage disorders” (Bryan Winchester)

Brain and more Gaucher - or what did we learn in the last 10 years

09:45 - 10:15  UPR or not UPR - what is the lesson for Gaucher disease? (Mia Horowitz)

10:15 - 10:45  The role of LIMP2 for the delivery of glucocerebrosidase to lysosomes - why is this of importance for neurologists? (Michael Schwake)

10:45 - 11:15  Coffee break

11:15 - 11:45  Glucocerebrosidase gene - what can we learn for Parkinson’s disease? (Christine Klein)

11:45 - 12:15  GBA-associated Parkinsonism using mouse models (Nahid Tayebi)

12:15 - 12:45  Prodromal pre-motor Parkinsonian features in carriers of Gaucher disease (David Arkadir)

12:45 - 13:45  Lunch

13:45 - 14:15  Glucocerebrosidase chaperones and its significance for the normalization of α-Synuclein in dopaminergic neurons (Ellen Sidransky)

14:15 - 14:45  What are we learning from the neuronopathic type in young Gaucher disease patients (Uma Ramaswami)

14:45 - 15:15  Why do we need better subgroups in Gaucher disease - what about the neurology? (Han-Wook Yoo)

15:15 - 15:45  Management of patients with neuronopathic Gaucher disease (Aya Narita)

15:45 - 16:15  Afternoon review: To biomarker or not to biomarker? (Arndt Rolfs)
Recent advances in rare disease:
Gaucher disease as a model RARD 2017

Friday, May 19, 2017

**Gaucher disease - new therapeutic modalities**

08:00 - 08:15 Opening remarks (*Ari Zimran*)

08:15 - 08:45 The role of Complement activation in glucosylceramide accumulation and tissue inflammation in Gaucher disease (*Jörg Koehl*)

08:45 - 09:15 Application of oral glucan particle macrophage mediated gene therapy for treatment of Gaucher disease (*Ed Ginns*)

09:15 - 09:45 Impaired Dopaminergic Development in an iPSC Model of Gaucher Disease (*Ricardo A. Feldman*)

09:45 - 10:15 Complexity in Lysosomal Diseases: insights from Niemann-Pick type C (*Frances Platt*)

10:15 - 10:45 Coffee break

**Current therapies**

10:45 - 11:15 How to treat newly diagnosed patients with Gaucher disease? (*Ari Zimran*)

11:15 - 11:45 Iminosugars for the treatment of Gaucher disease (*Hans Aerts*)

11:45 - 12:15 Co-morbidities and challenging cases in Gaucher disease (*Jeff Szer*)

12:15 - 12:45 Anti-coagulation in adult thrombocytopenic patients with Gaucher disease (*Barry Jacobson*)

12:45 - 13:45 Lunch

13:45 - 14:45 Poster session

14:45 - 15:45 Panel discussion: Is there any need for further treatment improvement in the ERT era?

Panelists: Debby Elstein, Jack Goldblatt, Maciej Machaczka, Elena Lukina, Hanna Rosenbaum, Ida Schwartz, Ari Zimran
Recent Advances in Rare Diseases: Gaucher disease as a model (RARD 2017)

Final program

Saturday, May 20, 2017

Plenary session

08:30 - 08:45 Opening remarks (Maciej Machaczka)

08:45 - 09:15 Newborn screening for lysosomal storage diseases - the special challenge of Gaucher disease (Roberto Giugliani)

09:15 - 09:45 Role of glycolipids in inflammation and immunity (Yaron Ilan)

09:45 - 10:15 Pregnancies in Gaucher disease patients (Heather Lau)

10:15 - 10:45 Coffee break

Parallel sessions

Session 1: Complement mediated diseases: paroxysmal nocturnal hemoglobinuria (PNH) and aHUS (Chair: Elena Lukina)

10:45 - 11:15 General practice in PNH treatment (Jeff Szer)

11:15 - 11:45 PNH and primary p.Cys89Tyr mutation in CD59: Differences and similarities (Dror Mevorach)

11:45 - 12:15 Genetics and differential diagnostics: TTP and aHUS (Lilian Monteiro)

12:15 - 12:45 Genetics in PNH - data from a global study (Arndt Rolfs)

Session 2: Fabry disease (Chair: Maciej Machaczka)

10:45 - 11:15 Prevention is the best therapy: the geneticist's approach (Gheona Altarescu)

11:15 - 11:45 X-chromosome inactivation in female patients with Fabry disease (Dominique Germain)

11:45 - 12:15 Kidney and what does it mean for treatment of Fabry’s disease (Christine Kurschat)

12:15 - 12:45 In vitro experimental results for the clinical interpretation in Fabry disease (Jan Lukas)

12:45 - 13:45 Lunch

13:45 - 15:15 Oral presentations of participants

15:15 - 15:45 Poster prizes - Conference summary and closing