Recent Advances in Rare Diseases:
**Frequently misdiagnosed hereditary disorders (FREMIDIS)** – multidisciplinary translational research affects global clinical impact

Preliminary program

**SCIENTIFIC ADVISORY COMMITTEE:**

Prof. Ari Zimran/Israel; Prof. Dr. Mia Horowitz/Israel; Prof. Dr. Uma Ramaswami/UK; Prof. Elena Lukina/Russia; Prof. Dr. Roberto Giugliani/Brazil; Dr. Ludwig Luis Antonio Albornoz Tovar/Colombia; Prof Joaquín Carrillo Farga/Mexico; Dr. Paula Rozenfeld/Argentina; Dr. L Monteiro/Brazil

Thursday, June 20, 2019

09:00 – 09:15  Welcome and introduction to Bogota – NN
09:15 – 10:00  Opening Lecture: “Let’s un-rare the rare diseases” – A Rolfs, Germany
10:00 – 10:45  New horizons in the therapy of genetic diseases – LLA Albornoz Tovar, Colombia

**HOT TOPICS (CHAIRS: N TAYEBI, USA; L MONTEIRO, BRAZIL)**

10:45 – 11:15  Stem cell therapy for the central nervous system in lysosomal storage diseases – JH Wolfe, USA
11:15 – 11:45  Many genes are involved – lessons to be learnt for the ER in LSD – M Horowitz, Israel
11:45 – 12:15  Epigenetics in lysosomal storage disorders – ESidransky, USA
12:15 – 12:45  Long-term complications in Gaucher disease – S Revel Vilk, Israel
12:45 – 14:00  LUNCH
14:00 – 14:30  LSD in LATAM – an example for unmet needs in rare diseases – RGugliani, Brazil
14:30 – 15:00  Therapy of Gaucher disease in Pakistan – HCheema, Pakistan
15:30 – 16:00  Treatment of rare hereditary diseases – lessons for emerging countries – URamaswami, UK
Friday, June 21, 2019

PLENARY LECTURE (CHAIR: A ZIMRAN, ISRAEL)
08:00 – 08:30  Strategies for clinical implementation of screening for hereditary disorders – P Bauer, Germany
08:30 – 09:00  What is new in iPS cells in metabolic diseases – R Feldman, USA
09:00 – 09:30  Experimental therapeutics in animal models in LSDs – R D’Hooge, Belgium
09:30 – 10:00  Why is early and simple diagnosis critical in LSDs – C Kurschat, Germany
10:00 – 10:30  Hereditary Cardiomyopathies: decision-making about genetic testing – C Louis, USA
10:30 – 11:00  COFFEE BREAK

DMD AND LGMD (CHAIR: E SIDRANSKY, USA)
11:00 – 11:30  Genetic diagnosis in Duchenne muscular dystrophy – J Saute, Brazil
11:30 – 12:00  Clinical and molecular characterization of a cohort of Colombian patients with Duchenne Muscular Dystrophy – F Suarez Obando, Colombia
12:00 – 12:30  Duchenne muscular dystrophy: CRISPR/Cas9 treatment – D Duan, Colombia
12:30 – 13:00  Cardiac outcome in Pompe disease – AT van der Ploeg, Netherlands
12:30 – 13:00  Epidemiology, clinical manifestation and diagnostic options in limb-girdle dystrophies (LGMD) – C Angelini, Italy
13:00 – 14:00  LUNCH

PATIENT ADVOCACY (CHAIR: C KURSCHAT, GERMANY)
14:00 – 14:20  Charity – is this the right attitude in rare diseases? – Z Zeelig, Israel
14:20 – 14:40  Role of patient advocacy in rare diseases – T Collin-Histed, UK
14:40 – 15:00  What do I expect from my partners in the orphan drug world? – T House, USA
15:00 – 15:20  Patient data – support and fear – J McNary, USA
15:20 – 16:00  Panel Discussion “Collaborations: Advocacy, Pharma and Academia”
16:30 – 16:30  COFFEE BREAK
16:30 – 17:30  Poster presentations 1
PARALLEL SESSIONS

Session 1. Myopathies and CLN (CHAIR: LLA ALBORNOZ TOVAR, COLOMBIA)
09:00 – 09:30 Importance of the neurocognitive assessment in the neurodegenerative LSDs – H Amartino, Argentina
09:15 – 10:00 Progranulin, lysosomal regulation and neurodegenerative disease – EJ Huang, USA
10:00 – 10:30 Future therapeutic approaches for neuronal ceroid lipofuscinosis – A Rahim, UK
10:30 – 11:00 Treatment opportunities in patients with metabolic myopathies – J Vissing, Denmark

Session 2. Tandem MS/MS in hereditary metabolic disorders - basic science impacts clinical decisions (CHAIR: JC FARGA, MEXICO)
09:00 – 09:30 Metabolic disorder screening by mass spectrometry – what are we learning – G La Marca, Italy
09:30 – 10:00 Metabolomics for clinical diagnosis of inborn errors of metabolism – A Rahman, Saudi Arabia
10:00 – 10:30 LSD screening: mass spectrometry is the future – C Cozma, Germany
10:30 – 11:00 Tandem MS/MS: blood or urine? – C Auray-Blair, Canada

Session 3. Lysosomal storage disorders (MACIEJ MACHACZKA, SWEDEN/POLAND)
09:00 – 09:30 Screening strategies in lysosomal disorders – P Rozenfeld, Argentina
09:30 – 10:00 Gaucher disease – can we expect a further improvement of therapeutic goals – O Goker-Alpan, USA
10:00 – 10:30 Controversies in Gaucher disease – A Zimran, Israel
10:30 – 11:00 Late onset manifestation in NPC – M Patterson, USA

Session 4. aHUS and PNH (CHAIR: R GIUGLIANI, BRAZIL)
09:00 – 09:30 Genetics and differential diagnostics in aHUS – L Monteiro, Brazil
09:30 – 10:00 Genotype-phenotype correlations of low-frequency variants in the complement system – A de Breuk, The Netherlands
10:00 – 10:30 Clinical predictors of atypical syndrome phenotype and outcome – P Muus, UK
10:30 – 11:00 General practice in PNH treatment – J Szer, Australia
11:00 – 11:30 COFFEE BREAK
11:30 – 13.00  Unsolved cases session – FP Vairo, USA
13:00 – 14:00  LUNCH
14:00 – 16.30  Poster presentations 2

16:30 – 16.45  Conference summary and closing
Lecture highlighting the future avenues for diagnosis and therapy (A Rolfs, Germany)

19:00 – 22.00  Networking Dinner / Poster Prizes