RARD2019

Recent Advances in Rare Diseases:

Frequently misdiagnosed hereditary disorders (FREMIDIS) – multidisciplinary translational research affects global clinical impact

RECENT ADVANCES IN RARE DISEASES
JUNE 20-22, 2019 - BOGOTÁ

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: "Lets 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 –J **H 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 – **E Sidransky, 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 – **R Giugliani, Brazil**
14:30 – 15:00	Therapy of Gaucher disease in Pakistan – **H Cheema, Pakistan**
15:30 – 16:00	Treatment of rare hereditary diseases – lessons for emerging countries – **U Ramaswami, 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

Saturday, June 22, 2019

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**