10th International meeting on Neuroacanthocytosis Syndromes

Barcelona, March 25-27, 2020

PRELIMINARY PROGRAM

TUESDAY, MARCH 24, 2020:

Optional: Arrival of scientists, patients and caregivers

WEDNESDAY, MARCH 25, 2020:

11:00 - 17:00: Registration

Patients and Caregivers Session I

11:00-11:15  Introduction
11:15-13:00  Roundtable introduction of all patients
13:00-14:30  Lunch
14:30-15:00  Patient’s history
15:00-15:30  Short Clinical overview
15:30-15:45  New developments in translational research
15:45-16:30  Medical Q&A

Scientists/Patients and Caregivers Session

17:00-17:20  Meeting Opening:
17:20-19:30 h  Introduction

1.1. Neuropathology and pathogenesis of Chorea and movement disorders.
1.2. Neuroacanthocytosis syndromes, a critical update.
1.3. Chorea-Acanthocytosis as core neuroacanthocytosis syndrome

19:30  Reception
20:00  Dinner
THURSDAY, MARCH 26, 2020:

**Scientists/Patients and Caregivers Session**

**9:00 -10:30** - VPS13 genes in movement disorders

- 2.1. Genetics of choras and Neuroacanthocytosis syndromes
- 2.2. Pathogenic VPS13C mutations in movement disorders.
- 2.3. Mutations in VPS13D and movement disorders.

**10:30-10:45**  Coffee break

**10:45 - 12:15** - VPS13 proteins in cell processes (I).

- 3.1. Role of vps13 in organelle interactions and membrane contact sites.
- 3.2. Role of VPS13A and VPS13C in the endoplasmic reticulum.
- 3.3. Role of VPS13B in vesicle trafficking.

**12:15-13:45**  Lunch

**Parallel Sessions: Scientists**

**13:45- 14:45** - VPS13 proteins in cell processes (II).

- 3.4. Role of yeast vps13 in trans-Golgi network and mitochondria.
- 3.5. VPS13A in mitochondrial morphology and lipid droplet motility.
- 3.6. Role of VPS13A in mitochondria and lysosome.

**15:15- 16:45** - VPS13 proteins in health and disease (I).

- 4.1. Red blood cells in neurodegeneration.
- 4.2. Mice models in chorea-acanthocytosis
- 4.3. Defective intracellular signaling pathways in chorea-acanthocytosis

**Parallel Sessions: Patients and Caregivers Session**

**13:45-16:45**

**16:15- 16:30**  Coffee break

**Scientists/Patients and Caregivers Session**

**16:30 -18:00** - VPS13 proteins in health and disease (II).

- 4.4. VPS13A knock-out and mouse neuronal dysfunction.
- 4.5. VPS13B in membrane trafficking during acrosome biogenesis.
18:00 - 18:20 Coffee break
20:00 Dinner

FRIDAY, MARCH 27, 2020:

Patients and Caregivers

10:00-13:00 Trip to Gaudi’s world

Scientists Session

8:30 - 9:00 – Glenn Irvine prize to young investigator to give a short communication.

9:00 - 11:00 Clinics and standard of care in chorea-acanthocytosis

5.2. Targeting Lyn kinase and PI3K pathways to treat chorea-acanthocytosis.
5.3. Deep brain stimulation in a patient with chorea-acanthocytosis.
5.4. Future avenues for treatment of chorea-acanthocytosis.

11:00-11:15 Coffee break

11:15 - 12:00 Workshop.

6. Future directions of scientific work and conclusions