18th UPDATE IN THE MANAGEMENT OF GAUCHER DISEASE AND OTHER LYSOSOMAL DISORDERS. May 13th-16th, 2013.

Program goals

To provide participants with an update on diagnosis, treatment and management of Gaucher disease, Niemann-Pick C and other lysosomal disorders.

At the end of the course, participants will be able to:

- Analyse currently available options for lysosomal disease therapy, taking into account the safety and efficacy profiles of various therapeutic agents.
- Critically review the significance of different responses in Gaucher disease as defined by guidelines established and identify factors that influencing in response and discuss interindividual response.
- Discuss the different approaches of enzymatic and genetic diagnosis of lysosomal diseases, as well as provide recommendations for genetic counselling.
- Critically review clinical evidence and current therapeutic algorithms, including the FEETEG recommendations, to make appropriate treatment decisions in Gaucher disease.

Target audience

This course is directed to physicians involved in diagnosis and treatment of lysosomal diseases.

Accreditation

- An accreditation of this educational programme will be provided to participants.

Instructors

Training will be provided by experts in multidisciplinary areas related to the particular features of storage diseases.

Dr. Pilar Alfonso, PhD
Traslational Research Laboratory. Hospital Universitario Miguel Servet. CIBERER
Dr. Marcio Andrade Campos, MD,
Haematology Department. Hospital Universitario Miguel Servet.

Prof. Javier Banzo, MD, PhD
Nuclear Medicine Service. Hospital Clínico Universitario de Zaragoza.

Dr. José Luis Capablo, MD, PhD
Head of Neurology Department. Hospital Universitario Miguel Servet. CIBERER

Dr. Jesús Fraile, MD, PhD
Head of Otolaryngology Department. Hospital Universitario Miguel Servet

Dr. Javier Gervas Arruga, PhD
Traslational Research Laboratory. Hospital Universitario Miguel Servet. CIBERER

Dr. Pilar Giraldo MD, PhD
Head of Hematology Department. Trasational Research Unit. Hospital Universitario Miguel Servet. CIBERER

Dr. Pilar Irún, PhD
Biochemistry and Molecular/Celular Biology Department. Universidad de Zaragoza. CIBERER.

Dr. Paz Latre, MD
Primary care physician.

Dr. Blanca Medrano, PhD
Pharmacists and Nutritionist. Trasational Research Unit. Hospital Universitario Miguel Servet. CIBERER

Prof. Miguel Pocovi, PhD
Biochemistry and Molecular/Celular Biology Department. Universidad de Zaragoza. CIBERER.
Dr. Mercedes Roca, MD PhD
International Skeletal Society. CIBERER

Dr Luis Sarria, MD
Radiodiagnostic Department. Hospital Universitario Miguel Servet

Dr. Alicia Saenz de Cabezón MD
Neurophysiology Department. Hospital Universitario Miguel Servet. CIBERER

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PROGRAM

Co-Directors
Pilar Giraldo, MD, PhD - Miguel Servet University Hospital. CIBERER
Miguel Pocovi, Professor - Zaragoza University, Spain. CIBERER

May 13th 2013 (Monday)

Unidad de Investigación Traslacional. Hospital Universitario Miguel Servet (main floor, main entrance)
Address: Paseo de Isabel La Católica 1-3. 50009 Zaragoza

To all attendees
16:00-16:10
Welcome/Opening
Dr. Pilar Giraldo and Dr. Miguel Pocovi

16:10-16:30
Lysosomal Diseases: An overview, emerging new treatments.
Dr. Pilar Giraldo

16:30-17:00
Introduction to enzymatic and genetic diagnosis of Lysosomal Diseases.
Dr. Miguel Pocovi

Coffee Break

17:20-17:45
Biomarkers in the diagnosis and follow-up of Gaucher Disease Fabry and NPC.
Dr. Pilar Irúń
17:45-18:15
Evaluating Gaucher Disease by imaging techniques.
Dr. Mercedes Roca

18:15-18:45
Neurological exam of Gaucher Disease and other lysosomal disorders.
Dr. José Luis Capablo

18:45-19:00
Neurophysiological examination: an overview.
Dr. Alicia Saenz de Cabezón

19:00-19:15
Discussion

May 14th 2013 (Tuesday)

Group 1
Laboratorio de Bioquímica, Biología Molecular y Celular
Facultad de Ciencias. Universidad de Zaragoza
Campus Plaza San Francisco

9:00-13:30
Tour to Laboratory and Clinic
Enzymatic and genetic diagnosis of Gaucher, Fabry and Niemann-Pick C Diseases
Coordinated by Dr. Pilar Alfonso (CIBERER)

Group 2
Hospital Universitario Miguel Servet
Hematology Day Hospital, 1st floor
Paseo de Isabel la Católica, 1-3
9:00-13:00
Tour of the Hospital Universitario Miguel Servet premises

Coordinated by Dr. Pilar Giraldo

CLINICAL TRAINING:

- Clinical evaluation of Gaucher Disease patients  
  Dr. Pilar Giraldo

- Neurological exam of Gaucher Disease patients  
  Dr. José Luis Capablo

- Neurophysiological examination of Gaucher Disease patients  
  Dr. Alicia Saenz de Cabezón.

- ENT examination  
  Dr. Jesús Fraile

14:00-16:00
Lunch with the course instructors

To all attendees

FEETEG
Lacarra de Miguel, 35 2º izda
50008 Zaragoza

Evaluation of liver deposits by MRI
Dr. Luis Sarria. Radiodiagnóstics Department. Hospital Universitario Miguel Servet

Evaluation of hearing loss in GD
Dr. Jesús Fraile. Otolaryngology Department. Hospital Universitario Miguel Servet

16:00-18:00

FEETEG Registry and Clinical Trials
Case discussion
Dr. Pilar Giraldo
Dr. Paz Latre
May 15th 2013 (Wednesday)

**Group 2**
Laboratorio de Bioquímica, Biología Molecular y Celular
Facultad de Ciencias. Universidad de Zaragoza
Campus Plaza San Francisco

9:00-13:30
Tour to Laboratory and Clinic
*Enzymatic and genetic diagnosis of Gaucher, Fabry and Niemann-Pick C Diseases*
Coordinated by Dr. Pilar Alfonso (CIBERER)

**Group 1**
Hospital Universitario Miguel Servet
Servicio de Hematología
Paseo Isabel la Católica, 1-3

9:00-13:00
Tour of the Hospital Universitario Miguel Servet premises
Coordinated by Dr. Pilar Giraldo

**CLINICAL TRAINING:**

- *Clinical evaluation of Gaucher Disease patients*
  Dr. Pilar Giraldo

- *Neurological exam of Gaucher Disease patients*
  Dr. José Luis Capablo

- *Neurophysiological examination of Gaucher Disease patients*
  Dr. Alicia Saenz de Cabezón.

- *ENT examination*
  Dr. Jesús Fraile
14:00-16:00
Lunch with the course instructors

To all attendees
FEETEG
Lacarra de Miguel Street, 35 2nd floor-left
50008 Zaragoza

Bone density studies in Gaucher Disease
Dr. Marcio Andrade Campos. Haematology Department.

Functional Neuroimaging applications in GD
Dr. Javier Banzo. Nuclear Medicine Department. Hospital Clínico Universitario.

16:00-18:00
Molecular basis of therapy
Dr. Miguel Pocovi

Gaucher Disease. Therapeutic decisions
Dr. Pilar Giraldo

Iron overload management
Dr. Blanca Medrano

May 16th 2013 (Thursday)

To all attendants
Laboratorio de Bioquímica, Biología Molecular y Celular
Facultad de Ciencias. Universidad de Zaragoza
Campus Plaza San Francisco
9:30-14:30

Practices:

*Genetic Analysis of GD, FD, Niemann-Pick C. (Continuation)*

*Biomarkers determinations*

**Coordinated by Dr. Pilar Alfonso (CIBERER)**

14:00-16:00

Lunch

**To all attendants**

Centro de Diagnóstico Dra. Roca

_Carrera del Sábado_ Street, 4

50006 Zaragoza

16:30-19:30

*Practices of Radiological Studies: MRI, US Densitometry*

Dr. Mercedes Roca