I Luso-Hispanic Meeting for Congenital Disorders of Glycosylation (CDG)

Barcelona , 21-23 of October 2011

Vanessa Ferreira contact: (0034)628 101 693

Day 21 October

19.00: JAEKEN'S FAN CLUB (Vanessa, Liliana, Yone and Elena) will pick him up at the hotel. A guided tour to the Famous Raval will be done.

20.15: Welcome dinner at Restaurante L'Antic Magatzem: Calle Aldana, 8 (08015, Barcelona).

Day 22 October (Where: Casa del Mar, Calle Albareda, 1-13, Barcelona)

To all speakers:

During all Seminars it is extremely fruitful your participation in the discussions and answers! Thanks!

ROLE OF ALL SCIENTIFIC COLLABORATORS:

- To present a summary of the CV of our speakers in the Rare Disease field, with a main focus on CDG.
- To Control the time. **VERY IMPORTANT! Otherwise many speakers will not have the opportunity to give their talk!**
- To moderate and motivate families to do questions (Families questions have priority!!!!)

INTERVIEWS DONE BY EURORDIS (the Voice of Rare Disease Patients in Europe):

Having Eurordis with us is a unique and great opportunity. The interviews take **5** minutes maximum! The **subjects** you can see below. **If you do not agree with the subject let me know as soon as possible.** Thanks!

8.00: Vanessa and Liliana will pick up Professor Jaeken at the Hotel

8.00-9.00: Bring kids to kindergarten (The volunteer service is kindly offered by Cruz Roja)

8.30-9.00: **Registration (OBLIGATORY to all participants).** You will receive identification and documentation needed for the Meeting.

9.00-11.00: "Congenital Disorders of Glycosylation (CDG): Past, Present and Future"

Scientific collaborator: Dr Rafael Artuch (Clinical-Biochemistry Department, Hospital Sant Joan de Déu, Barcelona

9.00-9.15: Introduction by ***Vanessa Ferreira*** (Portuguese and Spanish CDG Associations)

9.15-9.20: **Dr Rafael presents a summary of Dr Belén Pérez Dueñas CV in the Rare Disease area, focused on CDG.** (articles, projects were she was involved etc)

Conference by

Dr Jaak Jaeken (Pediatric Department and Professor at the Faculty of Medicine at the University of Leuven, Belgium)

Translation by

Dr Belén Pérez-Dueñas (Neurology department, Hospital Sant Joan de Déu, Barcelona, Spain)

11.00-11.10: Living with CDG in Australia presentation by *Merell and Morgan Liddle*, CDG Australian patient representative.

11.10-11.30: *Coffee*

11.30-12.55: "The Hispano-Luso-Argentina landscape about CDG".

11.30-11.40: Scientific collaborator: Dr Vilaseca (Clinical Biochemistry Unity, Hospital Sant Joan de Déu, Barcelona, España)

Round table with: **15 minutes** each oral presentation and following the order:

11.40-11.55: ***Dr Mercedes Pineda*** (Neuropediatrician department,Hospital Sant Joan de Déu, Barcelona, España)

11.55-12.10: ***Dr Paz Briones*** *(*Molecular Genetics and Biochemistry Unity, Instituto de Bioquímica Clínica, España)

12.10-12.25: ***Dr Dulce Quelhas*** (Genetic biochemistry Unity, Instituto de Genética Jacinto Magalhaes, Portugal)

12.25-12.40: ***Dr Carla Asteggiano*** (Researcher at Centro de Estudio de las Metabolopatías Congénitas, Facultad de Ciencias Médicas, Universidad Nacional de Córdoba (UNC) , Argentina)

12.40-12.55: Questions-answers

12.55-13.10: Guía Metabólica: a powerful and interactive 2.0 network targeted to families affected by Inborn Metabolic Diseases

by *Dra Mercedes Serrano* *(*Neurology department, Hospital Sant Joan de Déu, Barcelona, Spain)

13.10-15.00: Lunch at Casa del Mar

Interviews by Eurordis: Meeting point: Casa del Mar Terrace. The interviews take **5** minutes maximum.
13.15: **Dr Vilaseca** and **Dr Serrano**: The role of Guia Metabólica.
13.25: **Dr Paula Videira**: The importance of Glycoimmunology for CDG.
14.00: **Maria Luisa Pendas Sanchez**: The role of Early intervention to CDG patients and families.
14.15: **Dr Terricabras and Dr Ruth García**: How your working areas can benefit CDG patients?
14.30: **Vanessa Ferreira, Rachel Levillain and Marta Sánchez:** The role of CDG patient's voice.

15.00-16.00: SALA 3 (first floor on the right side): **Room is prepared to project ppt presentations. An overview of their research work and the future plans related to CDG will be presented.** **Participants: Pf. Jaeken, Dr Artuch, Dr B P Dueñas, Dr Pineda, Dr Quelhas, Dr Briones, Dr Asteggiano, Dr Giròs, Dr B Pérez González).**

Scientific collaborator: Dr Rafael Artuch (Clinical-Biochemistry Department, Hospital Sant Joan de Déu, Barcelona

Role: to control the time and moderate discussion.

15.00-15-15: Dr Paula Videira. (Medicine Faculty, Chronic Diseases Research Centre, Lisbon, Portugal. Faculdade de Ciências Médicas, Universidade Nova de Lisboa, Portugal)

15.15-15.30: Discussion

15.30-15.45: Dr Javier Corral (Servicio de Hematología y Oncología Médica. Hospital Universitario Morales Meseguer. Departamento de Medicina de la Universidad de Murcia**)**

15.45-16.00: Discussion

16.00: RETURN TO CONFERENCE ROOM

15.00-16.30: "The clinical manifestations of CDG: Prevention and treatment. Part I

Scientific collaborator: *Dr Mercedes Serrano* *(*Neurology department, Hospital Sant Joan de Déu, Barcelona, Spain)

15.00-15.30: Dra. Ruth García Romero (Gastroenterology, Hepathology and child nutrition department, Metabolic Diseases Unity, Hospital Sant Joan de Déu, Barcelona)

15.30-16.00: Dr. Luis Terricabras Carol (Orthopedic department at Hospital Sant Joan de Déu, Barcelona)

16.00-16.30: Questions-answers

16.30-16.45: Launch of CDG Online Community: A project in collaboration with Eurordis, Nord, Families, Researchers and Medical Doctors.

Presentation by *Denis Costello* **(**Web Communications Officer, Eurordis, Paris, France)

16.45-18.00: "The clinical manifestations of CDG: Prevention and treatment. Therapies. Part I I.

Scientific collaborator: **Dra Giròs** (Molecular Genetics and Biochemistry Unity ,Instituto de Bioquímica Clínica, España)

16.45-17.15: Seminar with *Dr Belén Pérez* *: Searching for new therapeutic targets based on the genotype.*

17.15-18.00: Round table about Therapeutical options:

Using Dr Belén presentation as a background, a round table based on Questions-answers will done.

with

Dr Belén Pérez *(G*enetic diagnosis of inherited metabolic disorders Department, Universidad Autónoma de Madrid, España)

Dr Jaak Jaeken (Pediatric Department and Professor at the Faculty of Medicine at the University of Leuven, Belgium)

Dr Belén Pérez-Dueñas (Neurology department, Hospital Sant Joan de Déu, Barcelona, Spain). **Responsible for translating Pf Jaeken and will also participate in the discussion with her experience.**

18.00-18.30**:** Coffee

Interviews by Eurordis: Meeting point: Casa del Mar Terrace.
18.00-18.10: Pf. Jaeken, Dr Artuch, Dr B Dueñas: How do you imagine CDG field in 10 years? **Pf Jaeken**: will answer focusing at the International level **Dr Artuch and Dr Belén Pérez Dueñas**: focus in the Spanish Scenario
18.10-18.15: Dr B Pérez: How advances in technology will help CDG diagnosis?
18.15-18.25: Dr Pineda, Dr Quelhas, Dr Briones, Dr Asteggiano: How the studies in their countries will benefit CDG patients?

18.30-20.00: "CDG Brainstorming with families, Researchers and Medical Doctors": fundraising, future research projects, establishment of collaborations between the key stakeholders, etc..

20.15: Dinner at Casa del Mar.

Día 23 Octubre (only for families and held in PRBB**: Barcelona Biomedical Research Park** (PRBB)- C/ Dr. Aiguader, 88-08003 Barcelona).

11.00-14.00: General Assembly for Spanish CDG Association. **Guided tour by Vanessa Ferreira to the laboratories at Barcelona Biomedical Research Park**

14:00: Lunch at FresCO (Salvador Espriu, 31-35, 08005 Barcelona)