I INTERNATIONAL MEETING ON MENKES

AND OTHER **RARE COPPER DISEASES**

(MARCO I)

& charity gala

12-13
SEPTEMBER 2024

MÁLAGA, SPAIN
(RUSSIAN MUSEUM COLLECTION)

COLECCIÓN DEL MUSEO RUSO

málaga

Ciudad de Málaga

Engrail THERAPEUTICS

Fundación Yusuf y Farida Hamied

SOCIEDAD ESPAÑOLA DE NEUROLOGÍA PEDIÁTRICA S.E.N.E.P.

Cu Mla
MENKES INTERNATIONAL

BASIC DETAILS

Hybrid modality (in-person and online)

Thursday 12 September
Friday 13 September 2024 (full days)

Friday 14 2024 at 20hrs – charity gala dinner

Simultaneous interpretation
English-Spanish available

Link to live streaming:
vimeo.com/event/4380080

Recommended hotel:
Hotel Maestranza (malaga@mshoteles.com)

Price: 30 euros (meeting) & 100 euros (gala dinner)

Payment: BIC/Swift: CAIXESBBXXX- IBAN: ES40 2100 0928 1302 0006 3432

ORGANISING COMMITTEE

Francesc Palau

Aurora Mateos

Elena Godoy

Stephanie Lotz

Denis Broun

Copper(less) Committee

Thursday 12 September 2024: Menkes Day

Morning session (9:30 - 13:30 hrs)

(Simultaneous interpretation English-Spanish available)

08:30-09:30 **Registration of participants**

9.30-09:50 **Opening and welcoming addresses**

09:50-10:30 **Inaugural lecture: Copper metabolism and the brain**

Svetlana Lutsenko, Professor of Physiology at the Johns Hopkins School of Medicine and Associate Director for basic science and clinical relations at the Institute for Basic Biomedical Sciences.

10:30-11:00 **Menkes disease: the challenges of a rare disease**

Francesc Palau. Director of the Department of Genetic Medicine and Pediatric Institute of Rare Diseases (IPER and CIBERER), Hospital Sant Joan Déu, Barcelona, Spain.

(11.00-11.20 **Coffee break**)

11.30-12.00 **Ethics of prenatal sequencing of the whole genome for rare disorders: the case of Menkes disease**

Anne-Marie Gerdes, Professor, Dept of Genetics Rigshospitalet, Copenhagen University, Denmark.

12:00-12:30 **The role of hospital compounding in access to treatments**

Miquel Villaronga, Hospital Pharmacist, Hospital Sant Joan de Déu, Barcelona, Spain.

Rubén Varela, Hospital Pharmacist, Complejo hospitalario León, Spain.

12:30-13:00 **Drug repurposing in rare diseases: the case of elesclomol-copper**

Vishal Gohil. Professor, Department of Biochemistry & Biophysics, Texas A&M University; College Station, TX, USA.

13:00-13:40 **Approaches to early diagnosis and clinical management of Menkes patients**

José Miguel Ramos-Fernández, Neuropediatrician, Head of the Child Neurology Unit. Department of Pediatrics, Regional University Hospital, Málaga, Spain.

Elena Godoy Molina, Attending Pediatrician, Complex Chronic Children and Palliative Care Unit, Department of Pediatrics, Regional University Hospital, Málaga, Spain.

(13:30-15:00 **Lunch**)

Side meeting room (level 1) Spanish and English

WORKSHOP FOR CLINICIANS and MEDICAL STUDENTS WITH MENKES PATIENTS

13:45-14:30 **Approaches to early diagnosis of Menkes disease in clinical practice**

By José Miguel Ramos-Fernández. Head of Pediatric Neurology, Hospital Materno-Infantil, Málaga and Diego Martinelli, Head of Metabolic Diseases Unit at the Bambino Gesù Paediatric Hospital, Rome, Italy.

(Food and drinks available during the workshop)

Afternoon session (15:00-19:00 hrs)

(Simultaneous interpretation English-Spanish available)

15:00-15:30 **The role of philanthropy in rare diseases: the case of Menkes disease**

Yusuf Hamied, philanthropist, CEO CIPLA Therapeutics.

15:30-16:15 **Round table 1: The Copper(less) Committee: a unique initiative of international scientific cooperation**

Moderator: **Denis Broun.** Director Givopax, former executive director of UNTAID, World Health Organization, Medical Director of Menkes International.

Michael J. Petris. Professor of Biochemistry, Departments of Ophthalmology and Biochemistry, University of Missouri, Columbia, MO, USA.

Francesc Palau. Director, Department of Genetic Medicine and Pediatric Institute of Rare Diseases (IPER and CIBERER), Hospital Sant Joan Déu, Barcelona, Spain.

Aurora Mateos. Founder and Director, Menkes International

Nina Horn. Professor of Molecular Medicine. Head of Laboratory and Research at Kennedy Center (rtd).

16:15-16:45 Challenges of a clinical trial for Menkes disease

Vikram Sudarsam, CEO, Engrail Therapeutics.

(16:45-17:15. **Tea break**)

17:15-18:00 Round table 2: Clinical follow-up and support for Menkes patients with exceptional treatment with elesclomol-copper

Moderator: **Francesc Palau.** Director. Department of Genetic Medicine and Pediatric Institute of Rare Diseases (IPER and CIBERER), Hospital Sant Joan Déu, Barcelona, Spain.

Elena Godoy. Attending Pediatrician, Complex Chronic Children and Palliative Care Unit, Department of Pediatrics, Regional University Hospital, Málaga, Spain.

Stephanie Lotz. Clinical Researcher, Department of Genetic Medicine and Pediatric Institute of Rare Diseases (IPER and CIBERER), Hospital Sant Joan Déu, Barcelona, Spain.

Rosa Marqués. Neurological Physiotherapist, expert in Menkes children, Dulce Nombre de Maria Psychopedagogic Institute, Málaga.

18:00-19:00 Round table 3: the voice of Menkes families

Moderator: **Aurora Mateos.** Founder and Director, Menkes International.

Association Angelli per la Vita (Menkes Italy)
Family of a child following Menkes exceptional treatment
Foundation Amigos de Nono (Menkes Spain)
Other Menkes families

Friday 13 September 2024

Morning session (9:00-13:30 hrs)

(Simultaneous interpretation English-Spanish available)

9:00-09:30 New treatment pathways for copper related rare diseases

Michael J. Petris, Professor of Biochemistry, Departments of Ophthalmology and Biochemistry, University of Missouri, Columbia, MO, USA

09:30-10:00 Progress in gene therapy for rare copper diseases

Cristina Fillat, Group Leader, Institut d'Investigacions Biomèdiques August Pi i Sunyer (IDIBAPS)-Hospital Clínic, Barcelona

10:00-10:30 Wilson's disease in pediatric patients: diagnosis and treatment

Aurelia Poujois. Coordinator of the Rare Disease Reference Centre "Wilson's disease and other copper-related rare diseases"Neurology Department Rothschild Foundation Hospital, head and neck expertise.

10:30-11:00 The Huppke-Brendel Syndrome and pre-clinical studies related to the disease

Victoriano Mulero. Professor, Coordinator of the research group on immunity, inflammation and cancer, University of Murcia.

(11.00-11.30 **Coffee break**)

Side meeting room (level 1) Only in Spanish

WORKSHOPS FOR FAMILIES

10:30-11:15 Workshop on nutrition

by **MD Javier Blasco**

– Ad hoc nutrition aspects for patients with copper rare diseases.
– Natural food enrichment. Texturing and thickeners.

11:15-12:00 Workshop on physiotherapy

by **Rosa Marqués**, Neurological Pediatric Physiotherapist
and Trainer Head of Clinical Pediatric Physiotherapist Teams.

– The role of physiotherapists in copper rare diseases treatments and homework with patients.

(Food and drinks available during the workshop)

12:15-13:45 Inspirational stories: The importance of civil society for rare disease new therapies

Nick Sireau, Beacon for rare diseases, CEO and Chair of Trustees at the AKU Society (UK)

11:30-12:15 Round table 4: The role of national drug regulatory agencies in new treatments for rare diseases

Manuel Ibarra, Head of the Department of Medicines Inspection and Control at the Spanish Agency for Medicines and Health Products (AEMPS).
Milena Peraita, Head of Compassionate Use at the Spanish Agency for Medicines and Health Products (AEMPS).

12:45-13:00 Round table 5: The voice of other copper rare diseases families

Moderator: **Aurora Mateos,** founder and director, Menkes International.

Faustino Giménez, Wilson Spain Association (Asociación Española de Familiares y Enfermos de Wilson)
Mª Paz Bustos, Association Huppke-Brendl Syndrome Spain (Princesa Gabriela)
Other families of patients with rare copper diseases.

(13:30-15:00. **Lunch**)

Afternoon session (15:00-17:15 hrs)

15:00-15:30 Informed medical consent and rare diseases

Joaquín Pérez Catalán, Director, Division of International Relations, Spanish Agency of Data Protection

15:30-16:15 Round table: Saving lives by predicting rare diseases through newborn screening

Moderator: **Raquel Yahyaoui,** Head, Dept. of Clinical Biochemistry. Clinical Laboratory Geneticist (EBMG). Hospital Regional Universitario de Málaga, IBIMA-Plataforma BIONAND. SPAIN.

James (Jim) Bonham, president elect of the International Society of Newborn Screening

Cristiano Rizzo, Principal Biologist of the Metabolic Diseases and Drug Biology Laboratory. Bambino Gesu Children's Hospital, Rome, Italy.

16:15-16:45 Closing lecture: Central role of metals in biomedicine

Raphaël Rodriguez. Skłodowska-Curie Chair of Chemical Biology, Research Director CNRS. Institut Curie, France.

16:45-17:15 Closing ceremony

Charity Gala (20:00 hrs)

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