



CME WEBINAR

EIGHTH EUROPEAN COURSE IN CLINICAL DYSMORPHOLOGY “WHAT I KNOW BEST” and EURODYSMOCLUB

ROME, OCTOBER 1-2-3, 2020

DIRECTORS: Karen Gripp, Fiorella Gurrieri, Tjitske Kleefstra

EURODYSMOCLUB: Marcella Zollino

EURODYSMOCLUB

(not accredited CME)

October 1st

- 11:30 Introduction *Marcella Zollino (Italy)*
- 11:40 Clinical Presentations *Chair: Tjitske Kleefstra (Netherlands)*
- 12:40 **Pause**
- 12:55 Clinical presentations *Chair: Marcella Zollino (Italy)*
- 13:55 **Pause**
- 14:10 Clinical presentations *Chair: Angelo Selicorni (Italy)*
- 15:10 End of the session

WHAT I KNOW BEST COURSE

(accredited CME)

October 2nd

- 11:40 Presentation of the Course
Karen Gripp, Fiorella Gurrieri, Tjitske Kleefstra

MAIN LECTURE

Judith Hall (Canada)

“The old and the new in arthrogyriposis”

Chair: *Giovanni Neri (Italy)*

SESSION 1

Syndromes with neurodevelopmental disorders or epileptic encephalopathies

Introduction and chair: *Marco Tartaglia (Italy)*

- 12:15 The ITHACA network
Tjitske Kleefstra (Netherlands)
- 12:40 Syndromic neurodevelopmental K+ channelopathies (Zimmerman Laband and so on)
Kerstin Kutsche (Germany)
- 13:05 Shaaf – Yang Syndrome
Christian Schaaf (Germany)
- 13:30 **Pause**
- 14:30 Pitt-Hopkins syndrome
Marcella Zollino (Italy)
- 14:55 Ayme-Gripp syndrome
Karen Gripp (USA)

SESSION 2

Neuropsychiatric Genetics

Chair: *Christian Schaaf (Heidelberg)*

- 15:20 Syndromes with (high chance) comorbid psychiatry
Tjitske Kleefstra (Netherlands)
- 15:45 From phenotype to SNVs and back
David Skuse (UK)
- 16:10 **Pause**
- 16:25 The Phenotypic Presentation of Young People with Copy Number Variants Associated with High Risk of Neurodevelopmental Disorder (ND-CNVs): Overview of Cardiff Research Findings
Marjanne vd Bree (UK)
- 16:50 SATB2 associated phenotypes
Yuri Zarate, (USA)

October 3rd

SESSION 3

Rare and recently identified syndromes

Chair: *Karen Gripp*

- 14:00 Fontaine syndrome and Gorlin-Chaudhry-Moss syndrome
Karin Witzl (Slovenia)
- 14:25 Mulibrey nanism
Kristiina Avela (Finland)
- 14:50 **Pause**
- 15:05 Sifrim-Hitz-Weiss syndrome
Karin Weiss (Israel)
- 15:30 MECT syndrome
Jeanne Amiel (France)

SESSION 4

WES diagnoses and further cohort identification

Chair: *Tjitske Kleefstra*

- 16:05 ANKRD11/KBG syndrome
Karen Low (UK)
- 16:30 DDX3X syndrome
Elliot Sherr (USA)
- 16:55 **Pause**
- 17:10 Adams-Oliver syndrome
Martin Zenker (Germany)
- 17:35 Conclusions
Karen Gripp, Fiorella Gurrieri, Tjitske Kleefstra

ECM/CME credits

Medical Doctors, Biologists (Pediatrics, Genetics, Neurology, Child Neuropsychiatry)

REGISTRATION FEE: € 180,00

REGISTRATION DEADLINE: September 25th, 2020

COORDINATION

POST-GRADUATE EDUCATION

Università Campus Bio-Medico di Roma

Via Álvaro del Portillo, 21 - 00128 Roma

Tel. (+39) 062.2541.9400

postlauream@unicampus.it