



# 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 1<sup>st</sup>**

- 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 2<sup>nd</sup>**

- 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 3<sup>rd</sup>**

#### 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 25<sup>th</sup>, 2020

COORDINATION

POST-GRADUATE EDUCATION

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