



European Cystic Fibrosis Society Diagnostic Network Working Group

Coordinator: Elke De Wachter
http://www.ecfs.eu/ecfs_dnwg



17th Annual Meeting, Utrecht, The Netherlands

6-8 February 2020

Organizing committee:

Inez Bronsveld, Universitair Medisch Centrum Utrecht, Utrecht, The Netherlands

Elke De Wachter, ECFS DNWG Coordinator, VUB-Brussel, Brussels, Belgium

Nick Simmonds, ECFS DNWG vice-coordinator, Royal Brompton Hospital, London, UK

Marlies Destoop, ECFS DNWG Assistant, VUB-Brussel, Brussels, Belgium

Scientific Program

Thursday 6 February 2020

13.00 Arrival and coffee

13.30 Welcome – Inez Bronsveld (Utrecht, The Netherlands)
Nicholas Simmonds (London, United Kingdom)

Session 1: The orange diagnostic journey - Part 1 (Chair: Nicholas Simmonds - Kors van der Ent)

13.40 Intestinal organoids: R117H gene expression and function
Jeffrey Beekman (Utrecht, The Netherlands)

14.10 Disease severity and CFTR testing
Eyleen de Poel (Utrecht, The Netherlands)

14.40 HIT CF - an update
Peter van Mourik (Utrecht, The Netherlands)

15:10 - Coffee break -

Session 2: The orange diagnostic journey - Part 2 (Chair: Silke van Koningsbrugge - Inez Bronsveld)

15.30 The history of the DNWG: Where did we come from?
Maarten Sinaasappel (Rotterdam, The Netherlands)

16.00 How CFTR changed my life
Hugo de Jonge (Rotterdam, The Netherlands)

16.30 How imaging changed my life
Harm Tiddens (Rotterdam, The Netherlands)

17.00 Triple excitement
Harry Heijerman (Utrecht, The Netherlands)

17.30 - Evening programme -

Friday 7 February 2020
Different shades of grey...

Session 3: Is CFTR functional testing 'still' the holy grail for diagnosing CF?
(Chairs: Elke de Wachter - Bente Aalbers)

- 9.00 10 years experience of NPD and ICM in a single centre
Burkhard Tümmler (Hannover, Germany)
- 9.30 Variants of unknown significance: how to make the unknown known?
Martina Wilke (Rotterdam, Netherlands)
- 10.00 YI1: CFTR-mediated chloride secretion in rectal biopsies is sage-dependent
Simon Gräber (Berlin, Germany)
- 10.15 YI2: Study of the function of the chlorine channel in patients with cystic fibrosis with the genetic variant E92K of the CFTR gene in the genotype
Yulia Melyanovskaya (Moscow, Russia)

10.30 - Coffee break -

Session 4: The sweat gland anno 2020
(Chairs: François Vermeulen - Bert Arets)

- 11.00 Sweat test: The position of ECFS
Natalia Cirilli (Ancona, Italy)
- 11.20 Is there still hope for sweat conductivity?
Dorota Sands (Warsaw, Poland)
- 11.40 Mucosweatomics
Teresinha Leal (Brussels, Belgium)
- 12.00 What is the role of sweat ions ratio in the diagnosis of Cystic Fibrosis
Davide Treggiari (Verona, Italy)

12.15 - Lunch break & Group picture -
- Sweat test demonstration -

**Session 5: DNWG as a partner of the ECFS-registry
(Chairs: Kevin Southern, Peter Middleton)**

- 13.30 Do we need a CFSPID-Registry? PRO
Susanna McColley (Chicago, USA)
- 13.50 Do we need a CFSPID-registry? CON
Lutz Nährlich (Giessen, Germany)
- 14.10 Discussion Pro-con
The group + moderators
- 14.25 Diagnosis of CFRD and definition in the CF-registry
Manfred Ballmann (Rostock, Germany)

14.55 - Coffee break -

**Session 6: In the grey zone
(Chairs: Kris De Boeck - Nico Derichs)**

- 15.30 CBAVD project: Is the Bombieri paper still relevant?
Elke De Wachter - Marlies Destoop (Brussels, Belgium)
- 15.45 CFTR-related disorders - time for new guidelines
Carlo Castellani (Genua, Italy)
- 16.00 Recurrent pancreatitis as a CFTR related disorder
Michael Wilschanski (Jerusalem, Israel)
- 16.30 Validation of intestinal organoids: correlation with disease severity in infants and adults with CF
Karin de Winter (Utrecht, The Netherlands)

17:00 - Evening programme -

Saturday 8 February 2020

Feeling blue after painting the town red

Session 7: The genetics round

(Chair: Carlo Castellani - Emmanuelle Girodon)

- 9.00 The secrets of the French CFTR database
Caroline Raynal (Montpellier, France)
- 9.30 Sequencing for lung disease - it's not all about CFTR!
Debbie Morris-Rosendahl (London, United Kingdom)
- 10.00 Where to draw the line: To treat, or not to treat?
Eitan Kerem (Jerusalem, Israel); talk by Michael Wilschanski
- 10.30 YI3: The genetic analysis of cystic fibrosis patients with seven new mutations in the central Anatolian region of Turkey
Zeynep Reyhan Onay (Ankara, Turkey)
- 10.45 YI4: Correlations between LCI and impulse oscillometry and spirometry in Polish pediatric patients with CF
Magdalena Postek (Warsaw, Poland)

11.00 - Coffee break -

Session 8: On the pink cloud? Newborn screening for CF

(Chairs: Karin de Winter - Silvia Gartner)

- 11.30 Benefits and pitfalls of PAP. The German experience
Olaf Sommerburg (Heidelberg, Germany)
- 12.00 Coaching parents after positive newborn screening
Jacqueline Noordhoek (Baarn, Netherlands)
- 12.30 5 years after the CFSPID designation: what have we learnt?
Kevin Southern (Liverpool, United Kingdom)
- 13.00 **Closing remarks**
Elke De Wachter & Inez Bronsveld

13.15 - End of meeting -

The meeting is kindly supported by:



