



**European Cystic Fibrosis Society**  
**Diagnostic Network Working Group**

(Coordinator: Nico Derichs)

[http://www.ecfs.eu/ecfs\\_dnwg](http://www.ecfs.eu/ecfs_dnwg)



**14<sup>th</sup> Annual Meeting, Ljubljana, Slovenia, 16<sup>th</sup>-18<sup>th</sup> February 2017**

**Meeting venue:**

**University Medical Center Ljubljana, University Children's Hospital**

**Bohoričeva 20, 1000 Ljubljana, Slovenia**

**Organizing committee:**

**Ana Kotnik Pirš - University Children's Hospital, Ljubljana, Slovenia**

**Valerie Mühlbacher – ECFS DNWG Assistant, Charité University Berlin, Germany**

**Nico Derichs – ECFS DNWG Coordinator, Charité University Berlin, Germany**

## Scientific Program

**Thursday, 16<sup>th</sup> February 2017**

14.00 Arrival – Snacks

14.30 Welcome: Ana Kotnik Pirš & Local team

### **Session 1: Building a structure for CF diagnosis and clinical care**

**(Chairs: Uroš Krivec, Lutz Nährlich)**

14.45 CF newborn screening in Slovenia: where are we and where would we like to be?  
- Ana Kotnik Pirš: (Ljubljana, Slovenia)

15.15 New generation sequencing in the diagnostics of CF: friend or/and foe?  
- Katarina Trebušak Podkrajšek (Ljubljana, Slovenia)

15.45 Data from the Slovenian CF Patient Database – Uroš Krivec (Ljubljana, Slovenia)

16.15 - Coffee break -

### **Session 2: Current practice and challenges of diagnosing CF in ...**

**(Chairs: Snezana Bojcin, CF Europe; Nico Derichs)**

16.45 Croatia – Dorian & Duska Tjesic-Drinkovic (Zagreb, Croatia)

17.10 Russia – Elena Kondratyeva (Moscow, Russia)

17.35 Moldova – Svetlana Sciuca (Chisinau, Moldova)

18.00 Turkey – Ayse Tana Aslan (Ankara, Turkey)

18.25 The perspective of CF Europe – Snezana Bojcin (Macedonia)

19.00 - Evening program -

**Friday, 17<sup>th</sup> February 2017**

**Session 3: Diagnosing CF after newborn screening and beyond: state-of-the-art 2017**

**(Chairs: Kevin Southern, Inez Bronsveld)**

- 9.00 New CFF Diagnosis Consensus Guidelines: The final outcome – Nico Derichs (Berlin, Germany)
- 9.30 The expansion and performance of national newborn screening programmes for CF in Europe - Jürg Barben (St. Gallen, Switzerland)
- 10.00 “CFSPID cases should be called and treated as patients” – Pro: Nicholas Simmonds (London, UK), Con: Kevin Southern (Liverpool, UK)
- 10.30 Discussion
- 11.00 - Coffee break -
- 11.30 How to follow-up CFSPID infants into school age? - Silvia Gartner (Barcelona, Spain)
- 12.00 Identifying the open questions for CF newborn screening - Anne Munck (Paris, France)
- 12.30 - Group picture / Lunch -

**Session 4: Diagnostic use of CFTR Biomarker**

**(Chairs: Michael Wilschanski, Nicholas Simmonds)**

- 14.00 Real life practice of sweat testing in Europe: Lessons learned from the ECFS DNWG survey & next steps – Natalia Cirilli (Ancona, Italy)
- 14.30 A new sweat test method based on chloride/potassium ratio - Petr Kuban (Brno, Czech Republic)
- 15.00 Young Investigator 1: Nasal Potential Difference in suspected CF patients with 5T polymorphism - Bente Aalbers (Utrecht, Netherlands)
- 15.15 Young Investigator 2: Electrophysiological tests in CF diagnosis: nasal potential difference is more predictive than sweat test: single center study - Hana Goldstein (Jerusalem, Israel)
- 15.30 Young Investigator 3: Diagnostic features of subjects with CFTR-related disorder – Rebecca Hyde (Hannover, Germany)
- 15.45 Young Investigator 4: Lung Clearance Index: understanding variability from a paediatric perspective - Erika Asperges (London, UK)

16.00 - Coffee break –

16.30 How to define a CF-typical NPD? Final results from the multicenter ECFS NPD SOP validation study - Inez Bronsveld (Utrecht, Netherlands) & Myriam Mesbani (Paris, France)

17.15 Future perspective of CFTR biomarkers for clinical trials – Francois Vermeulen (Leuven, Belgium)

17.45 HCO<sub>3</sub> and Cl<sup>-</sup> in CF: Update 2017 - Hugo De Jonge (Rotterdam, Netherlands)

18:15 Short guided tour of CF center Ljubljana

19:30 - Evening program -

**Saturday, 18<sup>th</sup> February 2017**

**Session 5: Extended *CFTR* genotyping: road to somewhere?**

**(Chairs: Caroline Raynal, Harry Cuppens)**

- 9.00 Complete *CFTR* gene mutation analysis in European patients with Cystic Fibrosis – Harry Cuppens (Leuven, Belgium)
- 9.30 The French *CFTR* genotyping network - Emmanuelle Girodon (Paris, France) & Caroline Raynal (Montpellier, France)
- 10.00 Personalised characterization of rare CF genotypes: examples from CFTR3 - Valerie Mühlbacher (Berlin, Germany)
- 10.30 Worldwide overview of *CFTR* genotypes – Refiloe Masekela (Durban, South Africa)
- 11:00 - Coffee break –

**Session 6: Documentation of CF diagnosis in registries**

**(Chairs: Ana Kotnik Pirš, Isabelle de Monestrol)**

- 11.30 Lessons learned for CF diagnosis from the ECFS Patient registry - Isabelle de Monestrol (Stockholm, Sweden)
- 12.00 The Global Registry Harmonisation Project: CF Diagnosis items and categories – Lutz Nährlich (Giessen, Germany)
- 12:30 The use of CF registry data for optimal clinical care - Tereza Doušová (Prague, Czech Republic)
- 13.00 Conclusion & DNWG perspectives – Nico Derichs (Berlin, Germany)

- End of meeting -

The meeting is kindly supported by:

