European Cystic Fibrosis Society
Diagnostic Network Working Group

(Coordinator: Nico Derichs)

http://www.ecfs.eu/ecfs_dnwg

14th Annual Meeting, Ljubljana, Slovenia, 16th-18th 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
Thursday, 16\textsuperscript{th} 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 – Ludmila Balanetchi (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, 17th 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 How to define a CF-typical NPD? Final results from the multicenter ECFS NPD SOP validation study - Inez Bronsveld (Utrecht, Netherlands) & Myriam Mesbahi (Paris, France)

14.45 Young Investigator 1: Nasal Potential Difference in suspected CF patients with 5T polymorphism - Bente Aalbers (Utrecht, Netherlands)

15.00 Young Investigator 2: Electrophysiological tests in CF diagnosis: nasal potential difference is more predictive than sweat test: single center study - Hana Goldstein (Jerusalem, Israel)

15.15 Young Investigator 3: Diagnostic features of subjects with CFTR-related disorder – Rebecca Hyde (Hannover, Germany)

15.30 Young Investigator 4: Lung Clearance Index: understanding variability from a paediatric perspective - Erika Asperges (London, UK)
15.45 - Coffee break –

16.15 Real life practice of sweat testing in Europe: Lessons learned from the ECFS DNWG survey & next steps – Natalia Cirilli (Ancona, Italy)

16.45 A new sweat test method based on chloride/potassium ratio - Petr Kuban (Brno, Czech Republic)

17.15 HCO₃ and Cl in CF: Update 2017 - Hugo De Jonge (Rotterdam, Netherlands)

18:00 Short guided tour of CF center Ljubljana

19:30 - Evening program -
Saturday, 18th 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:

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