



## ECFS Diagnostic Network Working Group Meeting Report

**13<sup>th</sup> Annual Meeting London - February, 11<sup>th</sup> - 13<sup>th</sup> 2016**

Royal Brompton Hospital, London

Hosts: Jane Davies, Nicholas Simmonds

**Thursday, 11<sup>th</sup> February 2016**

At 13:30 all participants of the 13<sup>th</sup> Annual ECFS DNWG Meeting arrived at the Royal Brompton Hospital/Guy Scadding Building (National Heart and Lung Institute) in Kensington, London, UK.

**Session 1: When it's not CF: State of the art diagnostics for other chronic suppurative lung diseases** (Chairs: Nick Simmonds, Isabelle Sermet-Gaudelus)

The meeting started at 14.00 with Jane Davies' welcome words to all members of the ECFS Diagnostic Network Working Group and participants of the 13th ECFS DNWG meeting.

The first presentation on the first meeting day was held by Claire Hogg (London, UK) about the advances in imaging of ciliary defects concerning especially electron microscopy, tomography and immunofluorescence. Afterwards Hannah Mitchison (London, UK) gave an update in primary ciliary dyskinesia genetics. Austin Worth (London, UK) provided a detailed insight into immune testing in bronchiectasis. Following Suzanne Crowley (Oslo, Norway / London, UK) discussed secondary defects in CFTR in non-CF chronic suppurative lung disease and presented data on nasal PD and inflammatory cytokines of patients with PCD. The first session of the meeting was closed by a discussion on the audience's questions.

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

(Chairs: Nico Derichs, Jacquélien Noordhoek)

The second session was opened by Olav Trond Storrøsten (Oslo, Norway) who gave an overview about the situation of Newborn screening program in Norway and presented data regarding the first three years of Norwegian CF Newborn Screening since it had been introduced in 2012. Reka Bodnár (Budapest, Hungary) discussed the situation in Hungary and

concluded that Hungary needed to establish the Newborn Screening, that the technique of sweat testing had to be modified and that they considered to establish CF centers in Hungary. Elpis Hatziagorou (Thessaloniki, Greece) reflected the CF diagnosing situation in Greece and reported on the need of a NBS program in Greece. Samia Hamouda (Tunis, Tunisia) presented the actual situation of CF diagnosing and treating in Tunisia. In the end she concluded that the physicians' awareness of CF had to be raised to have an earlier diagnosis and that the main challenges were rare available pulmonary function tests and difficulties in regular monitoring with bacteriological sputum. Jacqueliën Noordhoek, president of CF Europe, closed the second and last session on Thursday by presenting which possibilities CF Europe has to support diagnosing CF in European countries.

**Friday, 12<sup>th</sup> February 2016**

**Session 3: Precisely defining and communicating the diagnosis of CF and associated phenotypes (Chairs: Kris De Boeck, Carlo Castellani)**

The second meeting day started with the presentation of the present status of the outcome of the CFF Diagnosis Consensus Meeting that was held in October 2015. Clement Ren (Indianapolis, USA) presented the screened populations, Patrick Sosnay (Baltimore, USA) talked about non-screened populations. A Pro-Con debate concerning the extended use of 'CFTR-related disorders' as label for multiorgan CF-like disease, not fulfilling CF diagnostic criteria followed. Kevin Southern (Liverpool, UK) gave a favourable opinion on this topic while Jürg Barben (St. Gallen, Switzerland) presented the counter-arguments. Following Jane Davies (London, UK) raised the question why a diagnosis of CFSPID could be harder to handle than a diagnosis of CF and gave ideas how that could be improved in the future. Following Jane Davies talked with the pediatric Psychologist Dr. Michele Puckey, who presented the difficult situation of handling the label CFSPID from her point of view. Nicholas Simmonds (London, UK) described from the perspective of an adult physician the difficulties in diagnosing CF in adults. Furthermore he invited the group to be part of a skype conference with a South African patient (who moved to London) who talked about her life with all the challenges as a CF patient. Nico Derichs (Berlin, Germany) closed the 3<sup>rd</sup> Session by presenting the ECFS Diagnosis Consensus Delphi Process 2016 as an update of the ECFS Diagnostic Algorithm from 2006.

#### **Session 4: Diagnostic use of CFTR Biomarker & Genetics**

**(Chairs: Michael Wilschanski, Inez Bronsveld)**

Natalia Cirilli (Ancona, Italy) opened the 4<sup>th</sup> Session with her talk about real life practice of sweat testing in Europe and presented the results of the ECFS DNWG survey as well as the further steps concerning the reevaluation of European sweat testing practice and planning of improvement initiatives. Isabelle Sermet-Gaudelus (Paris, France) talked about the definition of a CF-typical NPD on the basis of multicenter reference data from the diagnostic ECFS NPD SOP validation study. Nico Derichs (Berlin, Germany) spoke about the definition of a CF-typical ICM on the basis of multicenter reference data from the diagnostic ECFS ICM SOP validation study and the ECFS CTN ICM certification process. Afterwards the first of this year's six ECFS DNWG Young Investigators Michael Klosinski (Berlin, Germany) held his presentation about the diagnostic application of ECFS SOPs for NPD and ICM to patients with questionable CF or CFTR-related disorder. Following the second Young Investigator Barbara Bosch (Leuven, Belgium) talked about the relevance of organoids in a challenging diagnosis of atypical Cystic Fibrosis, presented the organoids methodology and her conclusions. Young Investigator Silvia Vercellone (Verona, Italy) talked about how to combine standardized and new CFTR functional tests for CF diagnosis. Afterwards Young Investigator Anne Bergougnoux (Montpellier, France) posed the question if ex vivo differentiated nasal epithelium qualified as a model for the functional characterization of splicing mutations in the *CFTR* gene and test of new targeted therapies. Following Elke De Wachter (Brussels, Belgium) held a talk about CFTR3 as a collection of clinical phenotype data in subjects with rare *CFTR* mutations. Anabela Ramalho (Leuven, Belgium) closed the last session on the second meeting day with her presentation about quantifying CFTR function in intestinal organoids of subjects with rare *CFTR* mutations.

<b>Saturday, 13<sup>th</sup> February 2016</b>
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#### **Session 5: Clinical trials and outcome measures in CF**

**(Chairs: Kevin Southern, Elke De Wachter)**

The last meeting day started with the presentation of Uta Griesenbach (London, UK) about CFTR gene therapy and future perspectives. Francois Vermeulen (Leuven, Belgium) talked about sweat test variability in real life. Jane Davies (London, UK) gave an update on the ECFS

CTN LCI Core facility and presented the plans for a multicenter clinimetrics study concerning lung clearance index. Katie Bayfield (London, UK) discussed further understanding differences between SF6 and nitrogen-based MBW techniques. Afterwards Michael Wilschanski (Jerusalem, Israel) closed the 5<sup>th</sup> meeting session with his talk about gastrointestinal outcome measurements in CF clinical trials.

### **Session 6: The ECFS Diagnostic Network Working Group - a growing global network**

**(Chair: Jane Davies)**

Refiloe Masekela (Durban, South Africa) started the last meeting session by giving an overview of the general situation of diagnosing CF in Africa. Cheryl Steward (Pretoria, South Africa) followed as a Young Investigator by talking about the molecular side of investigating CF in South Africa. She was followed by Carla Gomez (Campinas, Brazil) who spoke about pulsed direct and constant direct currents in the pilocarpine iontophoresis sweat chloride test. Following Ana Kotnik Pirs (Ljubljana, Slovenia) gave an update of the 1<sup>st</sup> Latin American Newborn and Early Screening of CF patients (LANES) Meeting 2015 and talked about the DNWG collaboration on this project.

Finally, Nico Derichs (Berlin, Germany) summarized the meeting and gave an overview about the current and future DNWG projects and collaborations. Sheila Scheinert has finished her position as DNWG Assistant, and Nico warmly welcomed Valerie Mühlbacher (Berlin) as the new DNWG Assistant. Martin Schwarz (Manchester) was honored as a founding member of the DNWG who is retiring now and leaving the group. Nico also thanked the hosts – Jane Davies and Nick Simmonds – for the great organisation of this meeting and closed the ECFS DNWG Meeting 2016.

We thank all speakers and participants for their ambitious and constructive contribution to this interesting meeting. We are looking forward to meeting you at our next ECFS DNWG

Meeting 16-18 February 2017 in Ljubljana!

April 4<sup>th</sup>, 2016

Valerie Mühlbacher – ECFS DNWG Assistant

Nico Derichs – ECFS DNWG Coordinator



### ECFS DNWG Young Investigator Travel Award Winners 2016

(left to right: Cheryl Stewart (Pretoria, South Africa), Anne Bergougoux (Montpellier, France), Carla Gomez (Campinas, Brazil), Barbara Bosch (Leuven, Belgium), Silvia Vercellone (Verona, Italy), Michael Klosinski (Berlin, Germany))



### ECFS Diagnostic Network Working Group

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