



ECFS Diagnostic Network Working Group Meeting Report

18th Annual Meeting - April 29th - 30th 2021

Virtual meeting via Zoom

Thursday, 29th April 2021

Day 1: Diagnostic challenges: Can we do even better?

Chair: Elke De Wachter (*UZ Brussel, Brussels, Belgium*) – **Jürg Barben** (*Children's Hospital of Eastern Switzerland, St Gallen, Switzerland*)

The virtual meeting started with welcome words by the coordinator of the ECFS Diagnostic Network Working Group, Elke De Wachter, giving an overview of the aims, achievements, and past meetings of the working group. Jürg Barben (*Children's Hospital of Eastern Switzerland, St Gallen, Switzerland*) shared with us some news of the Neonatal Screening Working Group (NSWG) and he presented the new book written by the working group in collaboration with the international Journal of Newborn Screening (IJNS) called "Newborn Screening for CF".

The first presentation was held by Natalia Cirilli (*United Hospitals, Salesi Children's Hospital, Ancona, Italy*), in which she gave us more insight in the sweat test project by the DNWG. The project is called: "A Quality Improvement Tool for Sweat Testing". The aim is to make a tool to guide CF centres in performing sweat test.

Following, Shoab Afzal (*Copenhagen University Hospital, Copenhagen, Denmark*) gave an in-depth presentation on his recently published paper called "Morbidity and mortality in carriers of the cystic fibrosis mutation CFTR Phe508del in the general population". The speaker illustrated the findings of his study and concluded that Phe508del carriers have a normal lifespan, however also have an increased risk of certain CF associated symptoms.

Mike Waller (*King's College, London, UK*) gave an overview on the relationship of upper and lower airways electrophysiology with lung function. He started by comparing nasal potential difference (NPD) to lower airway PD (LAPD). During his presentation he gave us more insight in the relationship between NPD and LAPD with FEV₁, the correlation between nasal sodium indices and LCI, and the correlation between chloride transport in the nose and lower airway.

Afterwards, the first of this year's Young Investigators, Sophia Pallenberg (*Clinic for pediatric pneumology, allergology and neonatology, Hanover Medical School, Hanover, Germany*), presented her work on the evaluation of the β -adrenergic sweat rate assay as a therapeutic monitoring tool for highly efficient CFTR modulation. In her study, β -adrenergic sweat secretion assay, pilocarpine iontophoresis sweat tests, spirometry and multiple breath washout were assessed in CF patients before and three months post treatment initiation with Kaftrio and in healthy individuals.

The second Young Investigator Ellie Russo (*Department of Women's and Children's Health, University of Liverpool, Liverpool, UK*) informed us about her study on CFSPID in the UK CF registry in which they assessed the extent to which infants with an inconclusive diagnosis were added to the UK CF Registry, how many of the infants later had a CF diagnosis and what data are available on follow-up sweat testing.

The next speaker, Anne Munck (*Hôpital Necker Enfants Malades, CF centre Paris, France*) stressed in her talk the importance of monitoring the performance of NBS for CF and compare the different protocols of NBS to optimize them. In order to evaluate the performance of NBS protocols, the research team established key outcomes and parameters that are required to do so.

Marlies Destoop (*VUB, Brussels, Belgium*) was the last speaker of the first day and talked about her survey study: "Do fertility clinics use the current recommendations to exclude CF in CBAVD men?".

Nicholas Simmonds, the vice-coordinator of the working group, closed the first day of the 18th annual DNWG meeting after summarizing the meeting.

Friday, 30th April 2021

Day 2: CFTR-RD: What's in a name?

Chairs: Carlo Castellani (*Giannina Gaslini Institute, Genoa, Italy*) - **Nick Simmonds** (*Royal Brompton Hospital, London, UK*)

The second meeting day started with welcome words by the vice-coordinator of the ECFS Diagnostic Network Working Group, Nick Simmonds, giving an overview of the program of the second day of the meeting.

The first presentation was given by Carlo Castellani (*Giannina Gaslini Institute, Genoa, Italy*). He gave us an update on the CFTR-RD project. Throughout his presentation, he guided us through all the questions that needed to be solved by the working group in order to make an updated version of the current guidelines.

Following, Isabelle Sermet (*Hôpital Necker-Enfants Malades, CF centre Paris, France*) discussed what biomarkers tell us if CF diagnosis is not straightforward. She enlightened us on how we can assess the risk of developing CF in CFTR related disease patients based on biomarkers. During her presentation, she mainly focused on NPD and ICM as biomarkers.

Afterwards, Caroline Raynal (*CHU de Montpellier, Montpellier, France*) informed us on aquagenic wrinkling and whether we should consider this a CFTR-RD. To answer this question, she gave us an in-depth presentation of the results of her CFTR genetic analysis in a cohort of French patients with aquagenic wrinkling.

Following, Michael Wilschanski (*Hadassah Hebrew University Medical Center, Jerusalem, Israel*) gave a clear overview on primary sclerosing cholangitis. He highlighted the unclear character of the etiology of the disease and what role CFTR dysfunction might have in the pathophysiology.

Next, a Pro-Con debate on “bronchiectasis as CFTR-RD” was held by Nick Simmonds (*Royal Brompton Hospital, London, UK*) explaining the advantages of considering bronchiectasis as CFTR-RD and Kevin Southern (*Alder Hey Children's Hospital, Liverpool, UK*) defending the CON debate by explaining the advantages of considering bronchiectasis as CF. This talk gave much input for a lively discussion within the group moderated by Kris De Boeck (*UZ Leuven, Leuven, Belgium*).

This presentation was followed by a talk by *Eitan Kerem (Hadassah Hebrew University Medical Center, Jerusalem, Israel)*, discussing the ethical aspects of treating patients with CFTR-RD with CFTR modulators.

Elke De Wachter closed the 18th Annual meeting after summarizing this very interesting meeting and giving future perspectives for the DNWG as an ECFS working group. She thanked Helen Chadwick and David Debisschop for the great organisation of this successful meeting and Nicholas Simmonds for his great help as vice-coordinator. Marlies Destoop, ECFS DNWG Assistant, was also thanked for all her excellent support. The ECFS was also thanked for sponsoring the surprise boxes that were send to the participants.

We thank all speakers and participants for their fantastic contribution to make the 18th Annual ECFS Diagnostic Network Working Group Meeting a great success!

We are looking forward to seeing you at our next ECFS DNWG Meeting.

30th April 2021

Elke De Wachter – ECFS DNWG Coordinator

Nicholas Simmonds – ECFS DNWG Vice-coordinator

Marlies Destoop – ECFS DNWG Assistant



'Surprise box' send to the participants (sponsored by ECFS)



Virtual get-together

The meeting was kindly supported by:

