

ECFS Diagnostic Network Working Group Meeting Report

DNWG meeting during the 46th ECFS Conference 2023
Vienna, Austria,
June 9th, 2023

Chair: Elke De Wachter (Brussels, Belgium) – **Nick Simmonds** (London, UK)

The welcome words, given by the coordinator Elke De Wachter, summarized the work that has been done in the previous months and the future projects of the working group.

Carlo Castellani (Genova, Italy) opened this session with a summary on the current status of the project “ECFS standards of care on CFTR-related disorders”. The aim of this project, driven by a core group of DNWG members (Carlo Castellani, Elke De Wachter, Christiane De Boeck, Nick Simmonds, Kevin Southern and Isabelle Sermet), is to update the recommendations of CFTR-RD. His talk was followed by a presentation given by **Natalia Cirilli** (Ancona, Italy). She gave an overview on the sweat test project. The aim of this project is to improve the quality of the performance of sweat testing in established and emerging CF services, including lower- and middle-income countries (LMIC) in and outside Europe.

Next, **Lieven Dupont** gave an update on the Belgian Unconfirmed Diagnosis project. He started his talk by giving an overview on the Belgian CF registry. The aim of this project is to review diagnostic criteria, genotype and clinical disease severity of patients registered in the registry without confirmed CF diagnosis and subsequently potentially identifying a subpopulation with CFTR-RD.

In the second part of the meeting, difficult cases were presented. **Michael Waller** (London, UK) gave an interactive presentation on two interesting cases involving the variant c.3874-4522A>G. The two cases stressed the importance of non-coding genomic regions and phenotypic variability and raised the question whether the above-mentioned variant should be included in future targeted diagnostic gene panels.

The second case was presented by **Anders Lindblad** (*Gothenburg, Sweden*). The title of the presentation was “Can we rule out CF in a child with an abnormal sweat test? Both presentations led to an interesting discussion.

Elke De Wachter closed the DNWG session during the ECFS conference and invited all the participants to the next DNWG meeting (8 - 10th February 2024) which will take place in Hannover, Germany.

We would like to thank ECFS for this successful meeting. We thank all speakers and participants for their fantastic contribution and for making the meeting such a great success!

We are looking forward to seeing you at our brainstorm meeting at the NACFC in Phoenix, USA, and our next annual DNWG Meeting in Hannover.

9th June 2023

Elke De Wachter – ECFS DNWG Coordinator

Nicholas Simmonds – ECFS DNWG Vice-coordinator

Marlies Destoop – ECFS DNWG Assistant

The meeting was kindly supported by:



Photos taken during the meeting:



Left to right: Nick Simmonds (London, UK), Elke De Wachter (Brussels, Belgium)



