



ECFS Diagnostic Network Working Group Meeting Report

17th Annual Meeting Utrecht, The Netherlands - February 6th - 8th 2020
University Medical Center Utrecht (UMC Utrecht), Utrecht, The Netherlands

Thursday, 6th February 2020

At 13:30 the meeting started with Inez Bronsveld's and Nicholas Simmonds' welcome words to all members of the ECFS Diagnostic Network Working Group and participants of the 17th annual ECFS DNWG meeting in Utrecht (The Netherlands).

Session 1: The orange diagnostic journey - Part 1

(Chair: Nicholas Simmonds – Kors van der Ent)

The first presentation on the first meeting day was held by Jeffrey Beekman (Utrecht, The Netherlands) on organoid biobank resource for preclinical exploration of therapies. He extended his talk by explaining gene editing and variation in R117H. Afterwards Eyleen de Poel (Utrecht, The Netherlands) gave an insight in her study on rare CFTR-mutations in The Netherlands (Rainbow project). Peter van Mourik (Utrecht, The Netherlands) followed with his talk about HIT CF. He gave an update on this project where drug candidates of several companies will be tested on organoids, followed by clinical trials and biobanking.

Session 2: The orange diagnostic journey – Part 2

(Chair: Silke van Koningsbruggen-Rietschel – Inez Bronsveld)

Maarten Sinaasappel (Rotterdam, The Netherlands) opened this session with an overview of the history of the Diagnostic Network Working Group. The former coordinator and co-founder of DNWG looked back on the early beginnings of the working group and the course it has taken since then. Hugo de Jonge (Rotterdam, The Netherlands) gave an in-depth presentation on the developments of CFTR research throughout his career as a researcher.

He reflected on the differences before and after the discovery of the CFTR gene. Following, Harm Tiddens (Rotterdam, The Netherlands) gave an overview of the developments in medical imaging throughout his career and his contribution to it. He spoke about the transition from X-Ray as a gold standard to computer tomography and magnetic resonance imaging and the dogmas he had to deal with during this transition. Harry Heijerman (Utrecht, The Netherlands) closed the last session on this meeting day. He looked back on the introduction of the sweat test and the benefits of the sweat test compared to other endpoints such as lung function tests. He also described the positive results seen with 'triple therapy' CFTR modulators.

Friday, 7th February 2020

Session 3: Is CFTR functional testing 'still' the holy grail for diagnosing CF?

(Chair: Elke De Wachter – Bente Aalbers)

The second meeting day started with a presentation by Burkhard Tümmler (Hannover, Germany). He discussed his experience of NPD and ICM over the last decade. Subsequently, Martina Wilke (Rotterdam, The Netherlands), discussed clinical molecular diagnostics used in the Netherlands and highlighted new identified CFTR-variants in 2018. Afterwards, the first of this year's Young Investigators, Simon Gräber (Berlin, Germany), presented his work CFTR-mediated chloride secretion in rectal biopsies and argued that results of normal CFTR-function may be age-dependent. The second Young Investigator Yulia Melyanovskaya (Moscow, Russia) informed us about her study of the function of the chloride channel in patients with cystic fibrosis with the CFTR genetic variant E92K.

Session 4: The sweat gland anno 2020

(Chair: François Vermeulen – Bert Arets)

Natalia Cirilli (Ancona, Italy) started the 4th session with her talk about the ECFS DNWG sweat test project. She addressed the issues that needed a shared decision regarding the position paper of ECFS on the sweat test performance. These matters were discussed with the audience. Dorota Sands (Warsaw, Poland) presented her study comparing Nanoduct (conductivity method) and classical Gibson Cook sweat test (pilocarpine iontophoresis).

Teresinha Leal (Brussels, Belgium) talked about MucoSweatOmics covering proteomic peptidomic and metabolic analysis of sweat and the eccrine sweat gland. The goal of this study is to find new biomarkers of CF. Davide Treggiari (Verona, Italy), explained the possible benefit of measuring different sweat ions and their ratio for the diagnosis of cystic fibrosis.

Session 5: DNWG as a partner of the ECFS-registry

(Chair: Kevin Southern – Peter Middleton)

A Pro-Con debate whether we need a CFSPID registry or not was held by Susanna McColley (Chicago, USA) explaining the advantages of a CFSPID registry and Lutz Nährlich (Giessen, Germany) defending the CON debate. This talk gave much input for a lively discussion within the group. Manfred Ballmann (Rostock, Germany) discussed the diagnosis of CF Related Diabetes (CFRD) and its definition in the CF registry. He highlighted areas where there is room for improvement within management and treatment of CFRD.

Session 6: In the grey zone

(Chair: Christiane De Boeck – Nico Derichs)

Marlies Destoop (Brussels, Belgium) presented her study on whether the recommendations to exclude underlying CF in CBAVD men are applied in fertility clinics. This talk opened the discussion whether these recommendations designed by Bombieri and coworkers should be updated. This presentation was followed by a talk by Carlo Castellani (Genoa, Italy), discussing the limitations of the existing guidelines for the diagnosis of CFTR-RD and the prospects for developing new recommendations in the near future. Michael Wilschanski (Jerusalem, Israel) highlighted recurrent pancreatitis as a CFTR related disorder. Karin de Winter (Utrecht, The Netherlands) closed the last session on this meeting day with a presentation on the validation of intestinal organoids and more specifically on the correlation with disease severity in infants and adults with CF.

Saturday, 8th February 2020

Session 7: The genetics round

(Chair: Carlo Castellani – Emmanuelle Girodon)

The last meeting day started with the presentation of Caroline Raynal (Montpellier, France) who gave us an insight into the French CFTR database. She discussed the unique working of CFTR-France and explained how this database could contribute to better CF care. Debbie Morris-Rosendahl (London, United Kingdom) presented the development of a comprehensive gene panel for rare inherited respiratory conditions, not focusing only on CF. Michael Wilschanski (Jerusalem, Israel) replaced Eitan Kerem in the presentation 'Where to draw the line: to treat or not to treat?' Based on real-life cases the audience debated on whether children with very mild symptoms, or CFTR-RD, should be treated with CFTR-modulators. Zeynep Reyhan Onay (Ankara, Turkey), the third Young Investigator, discussed the genetic analysis of CF-patients with seven new mutations in the central Anatolian Region of Turkey. The fourth Young Investigator, Magdalena Postek (Warsaw, Poland), talked about correlations between LCI and impulse oscillometry and spirometry in Polish pediatric patients with CF.

Session 8: On the pink cloud? Newborn screening for CF

(Chair: Karin de Winter – Silvia Gartner)

Olaf Sommerburg (Heidelberg, Germany) gave a clear overview on the benefits and pitfalls of pancreatitis-associated protein (PAP) in CF NBS. Jacquelin Noordhoek (Baarn, The Netherlands), the president of CF Europe, summarized a coaching program for parents with a positive NBS baby and those with a baby with CF after NBS. Kevin Southern (Liverpool, United Kingdom) closed this topic, discussing what we have learnt from ""CFSPD", 5 years after the introduction of this new designation.

Elke De Wachter (Brussels, Belgium), closed the 17th Annual meeting after summarizing this very interesting meeting and giving future perspectives for the DNWG as an ECFS-working group. She thanked the host – Inez Bronsveld and all sponsors - for the great organisation of this successful meeting and Nicholas Simmonds for his great help as vice-coordinator.

We thank all speakers and participants for their fantastic contribution to make the 17th Annual ECFS Diagnostic Network Working Group Meeting a great success!

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

14th February 2020

Inez Bronsveld – Local host

Elke De Wachter – ECFS DNWG Coordinator

Nicholas Simmonds – ECFS DNWG Vice-coordinator

Marlies Destoop – ECFS DNWG Assistant



ECFS DNWG Young Investigators 2020

(left to right: Nick Simmonds, Inez Bronsveld, Zeynep Reyhan Onay, Yulia Melyanovskaya, Magdalena Postek, Simon Gräber, Elke De Wachter)



ECFS Diagnostic Network Working Group

The meeting was kindly supported by:

