



ECFS Diagnostic Network Working Group Meeting Report

15th Annual Meeting St. Gallen, CH - February, 8th - 10th 2018
Kantonsspital St. Gallen, Switzerland

Thursday, 8th February 2018

At 13:30 all participants of the 15th Annual ECFS DNWG Meeting arrived at the Kantonsspital in St. Gallen, Switzerland. At 14:00 the meeting started with Jürg Barben's welcome words to all members of the ECFS Diagnostic Network Working Group and participants of the 15th annual ECFS DNWG meeting.

Session 1: Newborn Screening – looking over the precipice

(Chairs: Kevin Southern, Karin De Winter)

The first presentation on the first meeting day was held by Ralph Fingerhut (Zurich, Switzerland) about issues around IRT as an important value in newborn screening. Afterwards Danieli B. Salinas (Los Angeles, United States) provided a detailed insight into the Californian experience of newborn screening. Next presentation was a Pro-Con debate whether the recognition of CFSPID is beneficial for families. Danieli B. Salinas (Los Angeles, United States) gave a favourable opinion on this topic while Sarah Mayell (Liverpool, GB) presented the negative impact of being a "CFSPID"-called patient in CF care.

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

(Chairs: Kris De Boeck, Valerie Mühlbacher)

The second session was opened by Amale Hassani (Rabat Morocco) who gave an overview about the situation of CF diagnosing and the difficulties in CF care in Morocco. Tsitsino Parulava (Tbilisi, Georgia) discussed the CF diagnosing criteria in Georgia and presented Georgia's CF care system. Following Dijana Plaseska-Karanfilska (Skopje, Macedonia) spoke about CF diagnosing in Macedonia and all the difficulties behind. Olafur Thorvaldsson (Reykjavik, Iceland) gave an insight of the situation in Iceland, which is because of the small

amount of CF patients very special. Jürg Barben (St. Gallen, Switzerland) closed the last session on this meeting day.

Friday, 9th February 2018

Session 3: Taking sweat testing to the next level

(Chairs: Nico Derichs, François Vermeulen)

The second meeting day started with a presentation about the new CLSI sweat test guidelines by Vicky LeGrys (Chapel Hill, North Carolina, US). Following the question if sweat collection from two sites was essential to reduce QNS samples had been discussed. Vicky LeGrys provided advantages of sweat testing on two sides, while Kevin Southern (Liverpool, UK) spoke about the disadvantages. Afterwards Jürg Barben (St. Gallen, Switzerland) raised the question if it was time to unwrap the Nanoduct and gave a detailed overview over Nanoduct sweat testing system. Natalia Cirilli (Ancona, Italy) gave a talk about the improvement of sweat test performance in Europe and the possibility of establishing development of new ECFS DNWG training resources. Thao Nguyen-Khoa (Paris, France) presented results of evaporimetry in atypical and Lumacaftor/Ivacaftor treated CF patients. This session was closed by the discussion of defining top 3 priorities for sweat testing in Europe where all the participants of the DNWG meeting were asked to contribute their opinion.

Session 4: Current and future CF diagnostic markers

(Chairs: Nick Simmonds, Silvia Gartner)

Valerie Mühlbacher (Berlin, Germany) started the 4th session of this year's meeting with her talk on the ICM responsiveness to oral Lumacaftor/Ivacaftor treatment in CF patients. Lutz Nährlich (Giessen, Germany) followed with an update from the International CF Registry Group and gave special attention to the aim of designing future documentation of CF diagnosis and related phenotypes. Subsequently, Manfred Ballmann (Rostock, Germany) talked about the diagnosis of CF-related diabetes and what we have learned from the results of a recent multicentre study.

Following, the first of this year four Young Investigators presented her work: Magdalena Postek (Warsaw, Poland) presented the synergy of basic lung function tests (spirometry, MBNW, plethysmography) in diagnostics and monitoring CF patients. The second Young Investigator Olga Novoselova (Moscow, Russia) informed about Cytochrome genes polymorphism and effectiveness of antibacterial therapy in CF patients. Afterwards Reratilwe Mphahlele (Durban, South Africa) as the meeting's third Young Investigator discussed the complete CFTR gene mutation analysis in Black African patients with CF. Talking about the connection of CF carriership and tuberculosis, Lander Bosch (Cambridge, UK) – fourth Young Investigator – provided an insight into the geographic und demographic CF research.

Session 5: Extended CFTR genotyping: road to somewhere?

(Chairs: Emmanuelle Girodon, Harry Cuppens)

Sabina Gallati (Berne, Switzerland) started this day's last session with her talk about the assessment of the historical and current CFTR genetic identity in our population. Following Caroline Raynal (Montpellier, France) presented the impact of functional classification of the multi-faceted nature of CFTR mutations. Last talk for this meeting day was held by Elke De Wachter (Brussels, Belgium) about the question how it would be possible to get improved quality for genetic results.

Saturday, 10th February 2018
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Session 6: Intestinal organoids as (diagnostic) markers of CFTR function

(Chairs: Inez Bronsveld, Hugo De Jonge)

The last meeting day started with the presentation of Kris De Boeck (Leuven, Belgium) who raised the question of organoids were an additional helpful technique for diagnosing CF. Following Jeffrey Beekman (Utrecht, Netherlands) presented the correlation between intestinal organoids and sweat tests. Paola Melotti (Verona, Italy) described the relation between intestinal organoids and respiratory CFTR function (NPD). Hugo De Jonge (Utrecht, Netherlands) continued with presenting the rescue of CFTR-dependent bicarbonate transport in CF organoids.

Session 7: Functional and structural markers of CF lung disease

(Chairs: Jürg Barben)

Florian Singer (Zürich, Switzerland) talked about the multiple breath washout and whether it was ready for the inclusion into CF registries. Following, Kathryn Ramsey (Berne, Switzerland) gave a presentation on airway mucus properties as a biomarker in lung disease. Philipp Latzin (Berne, Switzerland) informed about respiratory rate and functional MRI in early CF lung disease.

Finally, Nico Derichs (Berlin, Germany) summarized the meeting and gave an overview about the current and future DNWG projects and collaborations. He thanked the host – Jürg Barben - for the great organisation of this meeting and closed the ECFS DNWG Meeting 2018.

We thank all speakers and participants for their fantastic contribution to make the 15th Annual ECFS Diagnostic Network Working Group Meeting a great success!

We are looking forward to meeting you at our next ECFS DNWG Meeting in 2019.

28th April, 2018

Valerie Mühlbacher – ECFS DNWG Assistant

Nico Derichs – ECFS DNWG Coordinator



ECFS DNWG Young Investigators 2018

(left to right: Reratilwe Mphahlele, Magdalena Postek, Lander Bosch, Olga Novoselova)



ECFS Diagnostic Network Working Group

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