ECFS Diagnostic Network Working Group
Meeting Report

14th Annual Meeting Ljubljana - February, 16th - 18th 2017

University Children's Hospital, Ljubljana, Slovenia

Thursday, 16th February 2017

At 14:00 all participants of the 14th Annual ECFS DNWG Meeting arrived at the University Children's Hospital in Ljubljana, Slovenia. At 14:30 the meeting started with Ana Kotnik Pirš' welcome words to all members of the ECFS Diagnostic Network Working Group and participants of the 14th ECFS DNWG meeting in Ljubljana.

Session 1: Building a structure for CF diagnosis and clinical care

(Chairs: Uroš Krivec, Lutz Nährlich)

The first presentation on the first meeting day was held by Ana Kotnik Pirš (Ljubljana, Slovenia) about the current situation, challenges and future perspektives of CF newborn screening in Slovenia. Afterwards Katarina Trebušak Podkrajšek (Ljubljana, Slovenia) provided a detailed insight into new generation sequencing and it's advantages and disadvantages. Following Uroš Krivec (Ljubljana, Slovenia) presented data from the Slovenian CF Patient database.

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

(Chairs: Nico Derichs, Valerie Mühlbacher)

The second session was opened by Dorian and Duska Tjesic-Drinkovic (Zagreb, Croatia) who gave an overview about the situation of CF diagnosing and the difficulties in introducing CF Newborn screening program in Croatia. Elena Kondratyeva (Moscow, Russia) discussed the CF diagnosing criteria in Russia and presented data from the Russian CF Patient database. Following Ludmila Balanetchi (Chisinau, Moldova) spoke about CF diagnosing in Moldova. She described the difficulties in Moldova especially concerning the introduction of CF newborn screening in Moldova and the problems with high infection rates of CF patients. Afterwards Ayse Tana Aslan (Ankara, Turkey) presented the status-quo of sweat testing,

newborn screening and genetic analysis in Turkey. Nico Derichs (Berlin, Germany) and Ana Kotnik Pirš (Ljubljana, Slovenia) closed the last session on this meeting day.

Friday, 17th February 2017

Session 3: Diagnosing CF after newborn screening and beyond: state-of-the-art 2017

(Chairs: Kevin Southern, Inez Bronsveld)

The second meeting day started with the presentation of the New CFF Diagnosis Consensus Guidelines. Nico Derichs (Berlin, DE) gave an update about the final outcome of the CFF Diagnosis guidelines. Afterwards Kevin Southern (Liverpool, GB) talked about the expansion and performance of national newborn screening programmes for CF in Europe. A discussion about whether CF mutation carriers should know about the fact they are carrier followed. Next presentation was a Pro-Con debate whether CFSPID cases should be called and treated as patients. Nicholas Simmonds (London, GB) gave a favourable opinion on this topic from the view of an Adult-CF-physician in regard to upcoming pancreas illness and infertility in adulthood of CF patients while Kevin Southern (Liverpool, GB) presented the negative impact of being a "patient". That talk gave much input for a following discussion on that topic within the group. Silvia Gartner (Barcelona, Spain) raised the question how to follow-up CFSPID infants into school age and what the possibilities are. Afterwards Anne Munck (Paris, France) closed the 3rd session by discussing open questions and needs concerning CF newborn screening.

Session 4: Diagnostic use of CFTR Biomarker

(Chairs: Michael Wilschanski, Nicholas Simmonds)

Inez Bronsveld (Utrecht, Netherlands) and Myriam Mesbahi (Paris, France) opened the 4th session with their talk about the definition of a CF-typical NPD. They presented the final results from the multicenter ECFS NPD SOP validation study. Following, the first of this year four Young Investigators presented her work: Bente Aalbers (Utrecht, Netherlands) showed her NPD measurement results in suspected CF patients with 5T polymorphism. The second Young Investigator Hana Goldstein (Jerusalem, Israel) presented her single center study

about nasal potential difference would being more predictive than sweat testing. Afterwards Rebecca Hyde (Hannover, Germany) as the meeting's third Young Investigator showed diagnostic features of subjects with CFTR-related disorder in her talk. Talking about the Lung Clearance Index, Erika Asperges (London, GB) – fourth Young Investigator – discussed the LCI variability from a pediatric perspective.

Subsequently, Natalia Cirilli (Ancona, Italy) drew a conclusion from the ECFS DNWG survey about real life practice of sweat testing in Europe and talked about the next steps and improvement initiatives that need to be done. Petr Kuban (Brno, Czech Republic) introduced a new sweat test method based on chloride/potassium ratio and disclosed the differences to common sweat testing methods. Hugo De Jonge (Rotterdam, Netherlands) closed the session with his talk about bicarbonate and chloride in CF.

Saturday, 18th February 2017

Session 5: Extended *CFTR* genotyping: road to somewhere?

(Chairs: Caroline Raynal, Harry Cuppens)

The last meeting day started with the presentation of Harry Cuppens (Leuven, Belgium) about complete *CFTR* gene mutation analysis in European patients with Cystic Fibrosis. Following Caroline Raynal (Montpellier, France) and Emmanuelle Girodon (Paris, France) provided an insight into the French *CFTR* genotyping network and talked about the search for rare mutations and their interpretation as well as further technological development. Following, Valerie Mühlbacher (Berlin, Germany) presented first results from CFTR3, a project which aims at a personalised characterization of rare CF genotypes. Refiloe Masekela (Durban, South Africa) closed the 5th session by providing a detailed worldwide overview of *CFTR* genotypes.

Session 6: Documentation of CF diagnosis in registries

(Chairs: Ana Kotnik Pirš, Isabelle De Monestrol)

Isabelle De Monestrol (Stockholm, Sweden) started the last meeting session with her talk about lessons learned for CF diagnosis from the ECFS Patient registry, what is missing and what could be useful mentioned in registries. Lutz Nährlich (Giessen, Germany) continued

with presenting CF diagnosis items and categories in terms of the Global Registry Harmonisation Project. Afterwards Tereza Doušová (Prague, Czech Republic) demonstrated the use of CF patient registry data for optimal clinical care, particularly with regard to the Czech CF Patient registry.

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 - Ana Kotnik Pirš - for the great organisation of this meeting and closed the ECFS DNWG Meeting 2017.

We thank all speakers and participants for their ambitious and informative contribution to this fruitful meeting. We are looking forward to meeting you at our next ECFS DNWG Meeting in 2018!

February 24th, 2017

Valerie Mühlbacher – ECFS DNWG Assistant

Nico Derichs – ECFS DNWG Coordinator

ECFS DNWG Young Investigators 2017

(left to right: Rebecca Hyde (Hannover, DE), Hana Goldstein (Jerusalem, IL), Erika Asperges (London, GB), Bente Aalbers (Utrecht, NL))

ECFS Diagnostic Network Working Group

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