# European Cystic Fibrosis Society

## Diagnostic Network Working Group

**(Coordinator: Nico Derichs, Berlin)**

# Final Program

## 11th Annual Meeting, 13-15 February 2014

**Hörsaalruine, Medical History Museum, Virchowweg 16**
**Charité University Berlin,**
**Charitéplatz 1, 10117 Berlin, Germany**

**Thursday, 13 February 2014**

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| 14.30 | Arrival – Snacks |
| ***Welcome*** | |
| 15.15 | Prof. Dr. Annette Grüters-Kieslich, Dekanin, Charité University Berlin<br>Prof. Dr. Philippe Stock, Director Paediatric Pulmonology and Immunology<br>PD Dr. Doris Staab, Head of Christiane Herzog CF Centre Berlin |
| ***Session 1*** | ***Structures for implementation of CF Newborn Screening (Chair: Kevin Southern, Liverpool)*** |
| 15.30 | Structures in existing European CF NBS programs (Jürg Barben, St. Gallen) |
| 15.45 | Implications from German Regional CF NBS experiences<br>(Jutta Hammermann, Dresden; Olaf Sommerburg, Heidelberg) |
| 16.15 | German National CF NBS: Choice of protocol, logistic aspects and follow up structures<br>(Manfred Ballmann, Bochum) |
| 16.35 | Socio-economical and political aspects of CF NBS (Andreas Reimann, Bonn) |
| 16.50 | Discussion (all) |
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| 17.00 | Coffee break |
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| ***Session 2*** | ***Current practice and challenges of diagnosing CF in Europe***<br>***(Chairs: Andreas Reimann, Mukoviszidose Institute Germany; Snezana Bojcin, CF Europe)*** |
| 17.30 | Slovenia (Ana Kotnik Pirs, Ljubljana) |
| 17.45 | Bulgaria (Rouzha Pancheva, Varna) |
| 18.00 | Estonia (Tiina Kahre, Tartu) |
| 18.15 | Ukraine (Halyna Makukh, Lviv) |
| 18.30 | Russia (Elena Amelina, Moscow) |
| 18.45 | DNWG Diagnostic Webforum (Inez Bronsveld, Utrecht) |
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| 19.00-19.45 | Charité Medical History Tour |
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| 20.00 | Dinner |

**Friday, 14 February 2014**

***Session 3*** ***CF Diagnosis: From Terminology to Registries (Chair: Kris De Boeck, Leuven)***

9.00 CF diagnosis in the US CF Patient Registry (Bruce C. Marshall, CFF, USA)

9.45 CF diagnosis in the European CF Patient Registry (Edward McKone, Dublin)

10.15 *Young Investigator:* Measures to improve documentation of CF diagnosis in CF Registries (Michaela Kleinschmidt, Brussels)

10.30 Results from the ECFS NSWG Delphi Consensus: Introducing the diagnostic category „CF Screen Positive, Inconclusive Diagnosis" (Kevin Southern, Liverpool)

10.40 CF and its new relatives: Update of the European CF Diagnostic Guideline (Nico Derichs, Berlin)

10.50 Discussion (all)

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11.00 Coffee break

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***Session 4*** ***CFTR Function: Sweat gland & Intestine (Chair: Nico Derichs, Berlin)***

11.30 New perspectives for sweat test performance (Lutz Nährlich, Giessen)

12.00 *Young Investigator:* ECFS ICM SOP multicentre validation: reference data for CF diagnosis (Lea Pinders, Berlin)

12.15 CTN-TDN ICM SOP for clinical trials: Training, certification, centralised analysis (Nico Derichs, Berlin)

12.30 Intestinal organoids: Implications for CF diagnosis (Jeffrey Beekman, Utrecht)

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13.00 Group picture / Lunch

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***Session 5*** ***CFTR Function: Respiratory (Chair: Michael Wilschanski, Jerusalem)***

14.00 NPD Experiences from Brazil (Izabela Sad, Rio de Janeiro)

14.30 *Young Investigator:* Revealing the potential: the search for NPD outcomes to support a CF diagnosis (Gloria Tridello, Verona)

14.45 *Young Investigator:* Interpretation of NPD measurements in difficult cases of possible CF and the role of published equations (Mike Waller, London)

15.00 ECFS NPD SOP multicentre validation: reference data for CF diagnosis (Inez Bronsveld, Utrecht)

15.30 CTN-TDN NPD SOP for clinical trials: Training, certification, centralised analysis (Isabelle Sermet, Paris)

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16.00 Coffee break

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***Session 6*** ***Multiple Breath Washout (Chair: Jane Davies, London)***

16.30 Background, techniques and implementation in routine CF care
(Philipp Latzin, Basel)

17.00 Challenges for CF diagnostics and clinical trials (Jane Davies, London)

17.30 CTN-TDN MBW SOP for clinical trials: Training, certification, centralised analysis
(Francois Vermeulen, Leuven)

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18.00 Evening programme

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**Saturday, 15 February 2014**

***Session 7*** ***CFTR Genetics (Chair: Burkhard Tümmler, Hannover)***

9.00 How to better classify *CFTR* mutations *in vitro* into mutation classes?
(Burkhard Tümmler, Hannover)

9.30 CFTR 2: Defining the most common CF-causing mutations helps to predict the individual clinical phenotype
PRO (Anne Munck, Paris), CON (Kevin Southern, Liverpool)

10.00 CFTR 3: Personalised characterisation of rare *CFTR* mutations (Sheila Scheinert, Berlin)

10.15 *Young Investigator:* Novel stop codon mutations give insight on CFTR function
(Aurelie Hatton, Paris)

10.30 New age of molecular diagnostics for CF with Next Generation Sequencing?
(Aleksandra Norek, Berlin)

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10.45 Coffee break

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***Session 8*** ***Translational Use of CF Diagnostic Tools (Chair: Inez Bronsveld, Utrecht)***

11.00 Follow-up of equivocal cases after CF NBS in Spain (Silvia Gartner, Barcelona)

11.30 *Young Investigator:* The 5T project: update after a year (Bente Aalbers, Utrecht)

11.45 CFTR repair by genistein and curcumin in cystic fibrosis patients carrying the S1251N channel gating mutation (Hugo de Jonge, Rotterdam)

12.15 CF Research in Australia (Peter Middleton, Sydney)

12.45 DNWG Perspectives (Nico Derichs, Berlin)

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13.00 Snack - End of meeting

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The meeting is kindly supported by:

(7500 €) (5000 €) (5000 €)

(4000 US $) (1500 €)