

Final Program
ECFS Diagnostic Network Working Group
Meeting Jerusalem, February 2013



Wednesday, 13 February 2013

Arrival
Wilschanski Lab Open House (optional)

Thursday, 14 February 2013

Session 1 CF Genetics (Chair: Michael Wilschanski)

- 9.00 Welcome: Dr. Osnat Lev – Zion, Director, Hadassah Hospital-Mount Scopus
 Welcome: Shira Zagori, Director, Israel CF Foundation
- 9.15 Keynote Lecture:
 Diagnosis of CF: 23 years after the discovery of the gene - Batsheva Kerem (Israel)
- 10.00 Young Investigator: Bente Aalbers (Netherlands)
 Nasal Potential Difference in suspected CF patients with 5T polymorphism –
 CFTR function testing and follow up in 40 patients from Israel
- 10.15 CFTR2: Update and future plans - Carlo Castellani (Italy)

- 10.45 Coffee break

Session 2 CF Newborn Screening (Chair: Carlo Castellani)

- 11.15 Diagnostic dilemmas resulting from Newborn Screening – Kevin Southern (UK)
- 11.45 Young Investigator: Patrick Staffler (Israel)
 Impact of CF population carrier screening - the Israeli experience
- 12.00 Young Investigator: Sara Caldrer (Italy)
 CFTR 186-8T/C allele in a subject diagnosed as affected by CF
- 12.15 Challenge the experts: 3 Diagnostic cases (5min+5min discussion)

- 12.45 Lunch

Session 3 *CFTR Function (Chair: Kris De Boeck)*

14.00 Diagnostic NPD criteria in Europe: survey results - Lutz Nährlich (Germany)

14.15 ECFS NPD SOP: Design of Multicentre diagnostic validation - Inez Bronsveld (Netherlands)

14.30 ECFS ICM SOP: Results of Multicentre diagnostic validation - Nico Derichs (Germany)

15.00 ECFS CTN certification for clinical trials - Isabelle Sermet/Isabelle Fajac (France)

15.30 Coffee break

Session 4 *CFTR Modulators (Chair: Batsheva Kerem)*

16.00 Young Investigator: Sheila Scheinert (Germany)
Reliability of Intestinal Current Measurement as outcome biomarker
for CFTR modulating therapies

16.15 Organoid research - Hugo de Jonge (Netherlands)

16.45 Diagnostic aspects of CFTR research in Italy - Paola Melotti (Italy)

17.15 A new educational tool for CF caregivers: CFTR.info - Kris De Boeck (Belgium)

Evening programme

Friday, 15 February 2013

Session 5 CF Diagnosis & Terminology (Chair: Inez Bronsveld)

9.00 CF Diagnosis in 2013: the clinical perspective - Eitan Kerem (Israel)

9.45 Young Investigator: Aleksandra Norek (Poland)
ENaC-related disorder is a new CF-like clinical entity

10.00 CF Diagnostic Algorithms are useful – Pro/Con (all)

10.45 Coffee break

Session 6 CF Diagnosis & Registries (Chair: Nico Derichs)

11.15 Who is reported in national CF registries?
Defining the needs for diagnostic documentation and terminology - Kris De Boeck (Belgium)

12.00 10 years ECFS Diagnostic Network Working Group - Michael Wilschanski (Israel)

12.15 Perspectives for the future

12.30 End of meeting
