



**ECFS CYSTIC FIBROSIS
NEONATAL SCREENING
WORKING GROUP MEETING**

Overview of last year's activities

29th European CF Conference
Copenhagen
Saturday 17th June 2006

*Carlo Castellani
Verona CF Centre
Italy*

ECFS NEONATAL SCREENING WORKING GROUP MEETING

Saturday June 17 12.30-14.00

29th ECFC Conference, Copenhagen 15-18 June 2006

Agenda of the meeting

Overview of last year's activities

Carlo Castellani, Italy

Information Branch: European survey preliminary results

France neonatal screening programme

Anne Munck, France

Positive Results Processing Branch: sweat test, pathway for difficult diagnoses

UK neonatal screening programme

Kevin Southern, UK

**Outcome Branch: neonatal screening costs evaluation
the Netherlands neonatal screening programme**

Jeanette Dankert-Roelse, the Netherlands



ECFS Neonatal Screening Working Group

- connecting professionals from laboratories and clinics working in CF neonatal screening
- sharing technical information on the IRT assay and on genetic analysis connected with CF neonatal screening protocols
- producing guidelines on some critical issues concerning CF neonatal screening
- helping European countries or regions interested in starting CF neonatal screening programs



ECFS Neonatal Screening Working Group

Co-ordination

Carlo Castellani, Verona

IRT laboratory branch

leader: Georges Travert, Caen

Molecular analysis branch

leader: Claude Ferec, Brest

PRP (Positive Result Processing) branch

Leaders: Kevin Southern, Liverpool and Anne Munck, Paris

Outcome branch

Leaders: Jeanette Dankert-Roelse, Baambrugge

Information branch

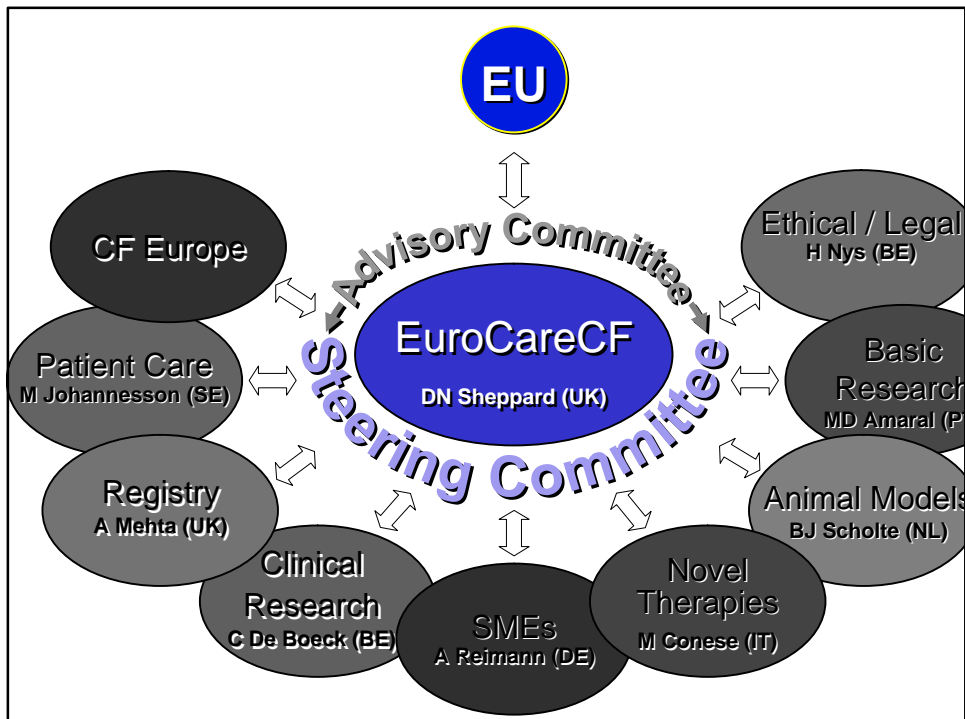
Leaders: Anne Munck, Paris and Carlo Castellani, Verona

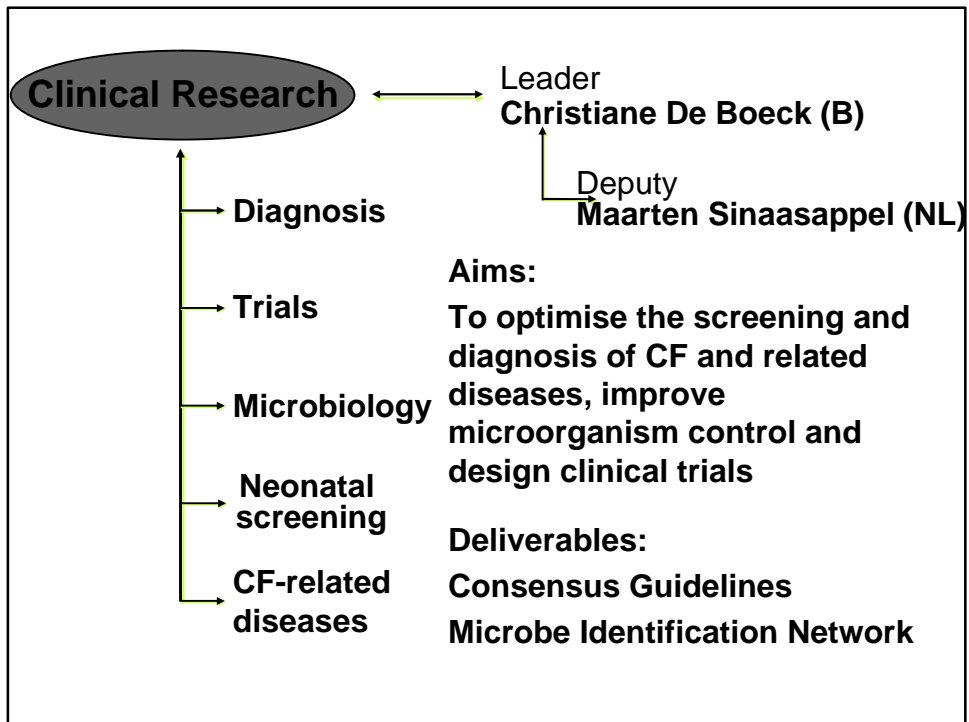
Members at 31/03/06: 117

CF neonatal screening net of contacts

IRT
Molecular genetics
Diagnosis
Follow-up

28
SCREENING
CENTRES IN
Austria
Czech rep
France
Italy
Poland
Spain
UK





Workpackage 3

Coordination of Clinical Research

Work Plan (continued...)

Microbiology:

- To produce reference material for the correct identification of CF microbes and to distribute this reference material.
- To organise workshops providing specialist training in molecular diagnostics and microbiological quality control.
- To disseminate widely in Europe expertise for the molecular typing of microbiological specimens.

Neonatal Screening:

- To organise workshops to analyse the experiences of European groups in neonatal screening.

Workpackage 3

Coordination of Clinical Research

Work Plan (continued...)

CF-Related Diseases:

- To organise workshops to analyse the experience of groups in Europe on CF-related diseases.
- To build a database of CF-related disease phenotypes.

All Groups:

- To contribute to the EuroCareCF newsletter and website with reports, suggestions and a discussion forum to ensure the rigorous review and development of consensus documents.
- To publish consensus guidelines.

Agenda Item 4:

Structure of EuroCareCF

- a) Steering Committee
- b) Advisory Committee
- c) Overview of the different workpackages and their activities
- d) Interaction of EuroCareCF with other EU projects
- e) Project start / end dates

Official start date: 1st January 2006

Effective start date: 1st April 2006

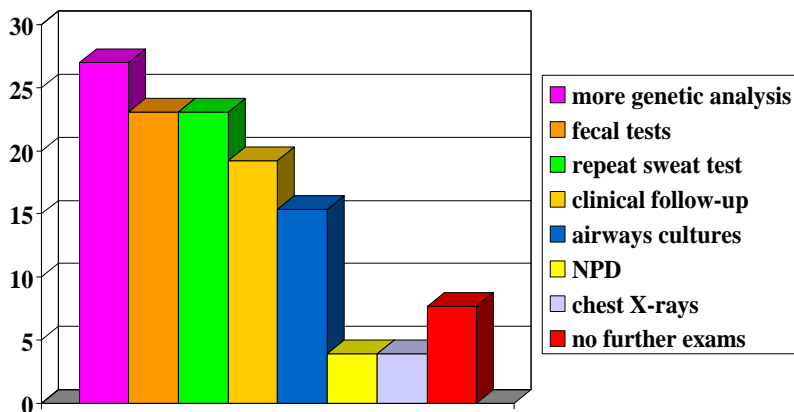
Official end date: 31st December 2008

Subject to our performance, an extension might be possible

WORKSHOP ECFDN 2006



Further exams in uncertain diagnoses



% do not add up to 100 as both missing data and multiple responses are present

Difficult diagnoses are growing in CF NBS ISSUES

- **definition of diagnosis**
- **prediction of outcome**
- **communication**

FOLLOW-UP

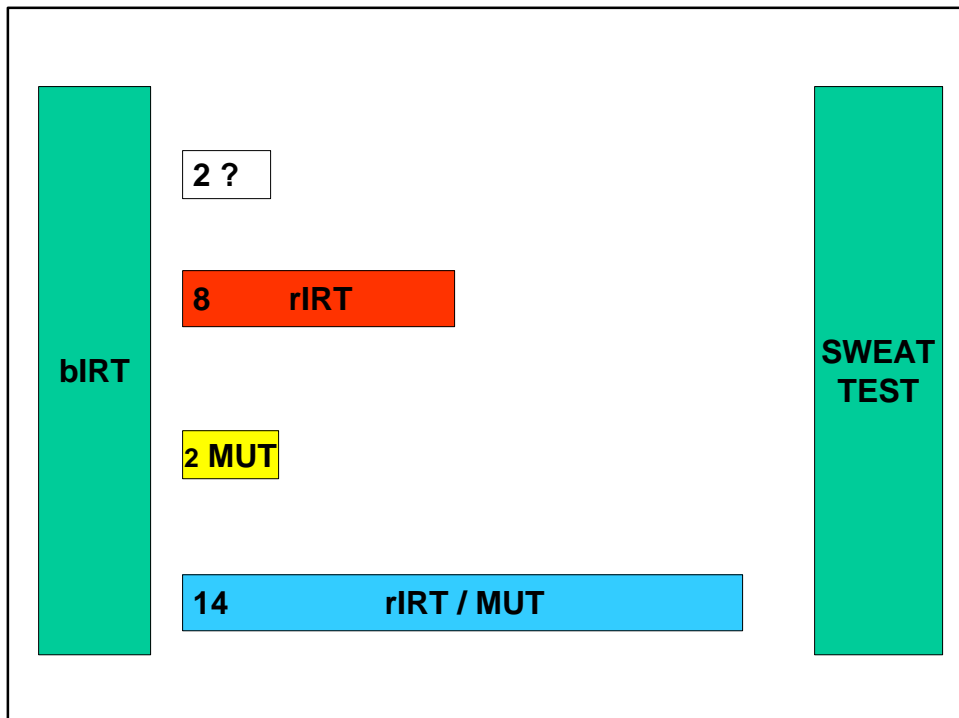
A Survey of Newborn Screening for Cystic Fibrosis in Europe

SOUTHERN KW, MUNCK A, POLLITT R, TRAVERT G,
ZANOLLA L, DANKERT-ROELSE J and CASTELLANI C,
on behalf of the ECFS CF Neonatal Screening Working Group

**JCF
in press**

Members of European Cystic Fibrosis Society Neonatal Screening Working Group who contributed to the survey

Aitken David, *Newborn Screening Laboratory, Institute of Medical Genetics, Glasgow, UK*
 Alonso Ramos Maria Jesus, *Genetic Screening Service, Castilla-Leon, Spain*
 Antonozzi Italo, *Genetic and metabolic disease section, University Hospital, Rome, Italy*
 Balascakova Miroslava, *Institute of Biology and Medical Genetics, Charles University Prague Czech Republic*
 Bernardi Filippo, *Clinica Pediatrica S. Orsola Malpighi, Bologna, Italy*
 Bignamini Elisabetta, *Pediatric Cystic Fibrosis Center, Turin, Piedmont, Italy*
 Bodamer Olaf, *University Children Hospital, Vienna, Austria*
 Bradley Don, *Newborn Screening Service, Wales, UK*
 Bucchi Micaela, *Clinica Pediatrica S. Orsola Malpighi, Bologna, Italy*
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 Colon Mejeras Cristobal, *Unidad de Deteccion Precoz Neonatal, Santiago de Compostela, Spain*
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 Gartner Silvia, *Cystic Fibrosis Center, Catalonia, Spain*
 Holubova Andrea, *Institute of Biology and Medical Genetics, Charles University Prague, Czech Republic*
 Iapichino Luciana, *Centro Regionale Fibrosi Cistica, Palermo, Italy*
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 Leavy Anne, *Royal Belfast Hospital for Sick Children, Belfast, UK*
 Lelli Alessandra, *Italian Red Cross Central Laboratory, Rome, Italy*
 Leslie Hilary, *Royal Belfast Hospital for Sick Children, Belfast, UK*
 Lillia Franco, *Malattie del Metabolismo e Screening Neonatale, Centro Microcitemie, Cagliari, Italy*
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 Vavrova Vera, *Department of Pediatrics, Charles Univ. Prague-2. School of Medicine, Czech Republic*
 Votava Felix, *Department of Pediatrics, Charles Univ. Prague-3. School of Medicine, Czech Republic*
 Zemkova Dana, *Department of Pediatrics, Charles Univ. Prague-2. School of Medicine, Czech Republic*



Protocols comparison

	Screening centers	Years of screening (min-max)	Neonates screened per year	Incidence	False negatives per year
-	2	4.4 (0.8-8)	12,500 (11000-14000)	-	-
rIRT	8	5.5 (0.9-21)	29,250 (16000-80000)	1/4450 (1/2850-10500)	0.5 (0.29-0.86)
MUT	2	4 (0-8)	-	-	-
rIRT MUT *	13	9 (2-32)	33,000 (7900-9 2000)	1/2775 (1/2250-5200)	0.3 (0-1)

* France not included