ECFS Working group
Neonatal Screening

Outcome branch

Fields of interest

- Follow-up after diagnosis through neonatal screening
- cost-effectiveness studies
- evaluation of advantages and disadvantages of CF neonatal screening

General planning

Outcome branch

- Develop a data registry aimed at
  1. collecting data about the *effectiveness of NBS* during 2 consecutive years (2006-2007)

  2. collecting data on *costs* during 1 year

Effectiveness of NBS

1. General data of NBS

For each participating country/region

- Total birth cohort
- Total number neonatal screening tests for CF
- Screening strategy
- Number of positive tests
- Number of CF patients detected by screening
- Number of CF-patients not-detected by screening

Effectiveness of NBS

2. Outcome data

For each identified CF-patient

- Age at diagnosis
- Genotype
- Number in ECFS-registry to be able to retrieve data from the ECFS-registry, such as
- Survival
- Number of hospital admissions and length of stay
- no. of outpatient clinic visits, planned and unplanned
- weight at birth and at age 6 months, 1 year, (18 months)
- height at birth and at age 6 months, 1 year, (18 months)

Costs of NBS

2. collecting data on *costs* during 1 year

- A questionnaire about the costs will be mailed soon to participants of the ECFS-Working Group
- After the results of this questionnaire have been obtained the data registry on effectiveness of NBS will start

Questionnaire on costs

1. Which screening test strategy is currently in use in your country/area?
   - ? IRT/IRT
   - ? IRT/DNA
   - ? IRT/DNA/IRT
   - ? other:…….

2. Can you give an approximation of the mean current costs per screening test, or if available an exact figure (preferably in €) and please indicate if the figure is an estimate or exact) of the following steps in the screening-program?

2.2

First Tier (please describe it)

•obtaining informed consent	€	(estimate/exact)
•sampling heel prick blood sample for CF screening	€	(estimate/exact)
•mailing blood sample	€	(estimate/exact)
•IRT-analysis (all costs included)*	€	(estimate/exact)
•Other (please specify)	€	(estimate/exact)

*all costs included: including materials, personnel and technical equipment

Questionnaire on costs

Second Tier (please describe it)

Sampling second heel prick	€	(estimate/exact)
DNA-analysis (all costs included)*	€	(estimate/exact)
mailing blood sample	€	(estimate/exact)
informing GP if screen positive	€	(estimate/exact)
informing parents if screen positive	€	(estimate/exact)
Other (please specify)	€	(estimate/exact)

Questionnaire on costs

Third tier (please describe it)

Sampling second heel prick	€	(estimate/exact)
Mailing blood sample	€	(estimate/exact)
IRT-analysis (all costs included)*	€	(estimate/exact)
Extended gene analysis (all costs included)*	€	(estimate/exact)
Other (please specify)	€	(estimate/exact

Questionnaire on costs

Screen positive tests

Sweat test	€	(estimate/exact)
Consultation at specialized center	€	(estimate/exact)
Genetic counselling	€	(estimate/exact)
Costs of further exams	€	(estimate/exact)

Participants of Outcome branch

Follow-up	
	AUSTRIA
Irmgard Eichler	Nationwide
Sabine Renner	
	CZECH REP
Veronika Slalicka	½ of Czech Republic
	FRANCE
Anne Munck	Nationwide
Muriel le Bourgeois	
	ITALY
Ettore Provenzano	Calabria
Carla Colombo	Lombardia
Rolando Gagliardini	Marche
Elisabetta Bignamini	Piemonte
Giovanni Taccetti	Toscana
Luciana Lapichino	Sicilia
Carlo Castellani	Veneto- Trentino AA
	POLAND
Dorota Sands	Pilot not yet operative
	SPAIN
Silvia Gartner	Catalunya
Barbara Judge	UK
Kevin Southern	
	East Anglia
	Leeds
	Northamptonshire
Anil Mehta	Scotland
	South Yorkshire,
	Ulster, Wales

A pilot study of neonatal screening for CF in the Netherlands

Background

In 2005, the Dutch Health Council advised to incorporate screening for CF in the routine newborn screening program.

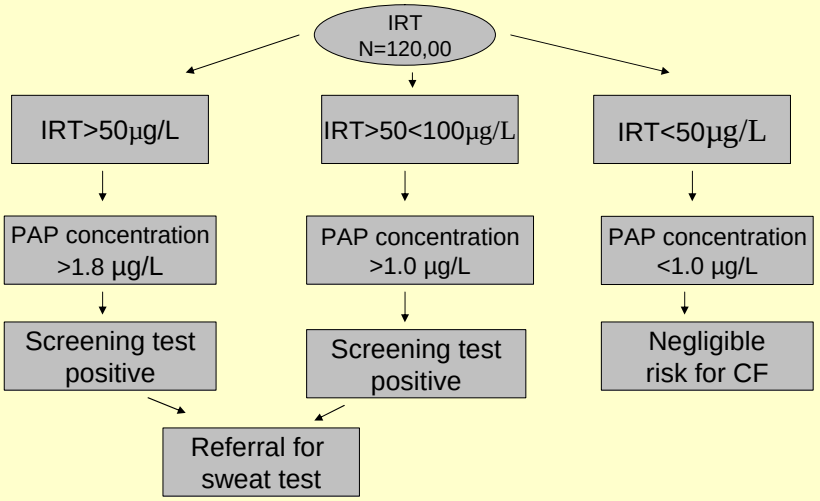
However, ***only*** when the specificity of newborn screening for CF would be better than in the current available screening strategies.

Before nationwide screening can be introduced in the Netherlands a pilot study will be carried out

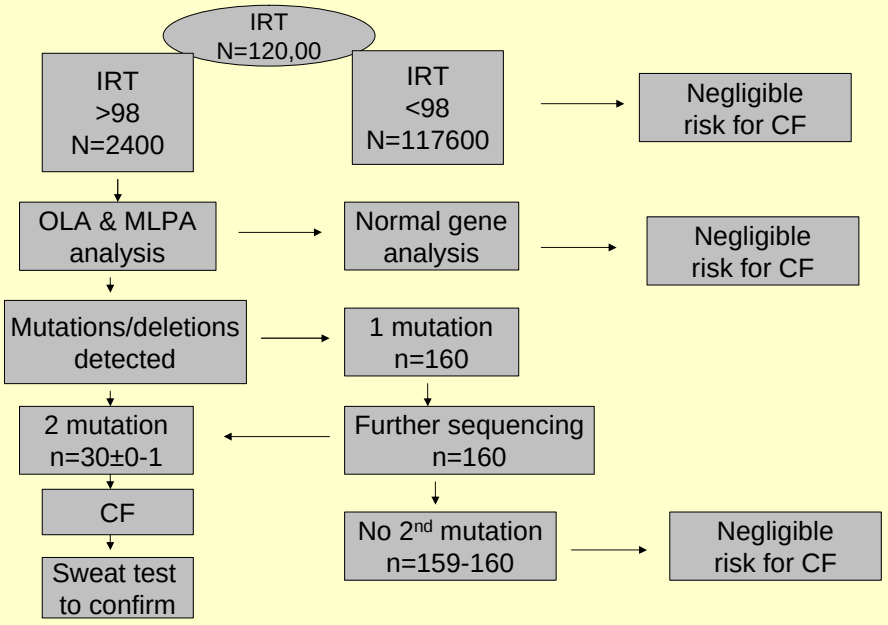
Aim of the pilot-study

- to enable informed decision making for implementation of newborn CF screening by studying test qualities, cost-effectiveness, and practical implications of two new screening strategies
- 2 screening strategies will be evaluated
  - IRT/PAP as described by Sarles et al, J Pediatrics 2005; 147:302-5.
  - IRT/ DNA/extended gene analysis

Alogorithm of IRT/PAP Stragegy



Algorithm of IRT/DNA/Extended gene analysis strategy



Questions to be answered

1. Which is the specificity, the sensitivity and predictive positive value of the IRT-DNA-EGA-approach compared to the IRT/PAP approach, and how perform these both approaches in comparison to current CF-newborn screening strategies as known from the literature?
2. What is the participation rate of routine newborn screening before and after the introduction of CF newborn screening?

Questions to be answered

3. What is the incremental cost-effectiveness of the IRT-DNA-EGA approach compared to the IRT/PAP approach?
4. What are the practical implications of adding CF newborn screening to the routine newborn screening programme:

a. are parents, obstetricians, nurses and GP's adequately informed about the purpose of newborn screening for CF?

Questions to be answered

b. how long does it take, after a positive screening test is reported to the GP, to definitely confirm or exclude the diagnosis CF?

c. is treatment of patients with CF identified by newborn screening adequately and timely started?

d. can adequate information reassure parents of a healthy baby with a positive screening test sufficiently and prevent longstanding anxiety?

Questions to be answered

e. do parents wish to be informed about their healthy children when these are identified as a CF-carrier during the screening process?

Comments from the audience?