Survey of the information provided for parents about newborn screening for CF in European programmes

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on behalf of the ECFS CF Newborn Screening Working Group

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The ECFS Newborn Screening (NBS) Group provides a forum for CF professionals to produce guidelines on critical issues and assisting implementing CF NBS across Europe.

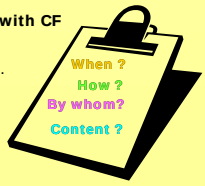
Aim

Determine current practice on the provision of information delivered to parents in Europe.

Material and Methods

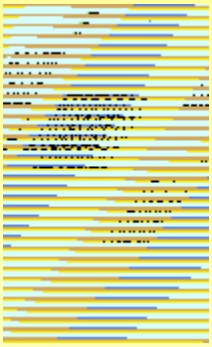
A questionnaire was sent to all established CF NBS programmes in Europe known to the focus group:

- **Information for all parents about CF NBS** *(both ante and post natal)*.
- **Information for parents of infants referred to a CF center for further assessment**.
- **Information for parents of infants with CF** *(positive diagnosis from NBS)*.
- **Information for parents of carriers**.
- **Information for parents of non-affected infants** *(negative NBS)*



Results

➢ **21 questionnaires were sent in 2005-2006 and 19 returned (90%)**



Italy (9 regions): Dr L. Palillo (W Sicily), Dr E. Provenzano Dr G. Parlatto (Calabria), Dr I. Antonozzi Dr S. Quattrucci (Lazio), Dr R. Gagliardini (Marche), Pr Bernardi Dr Bucci (Emilia Romagna), Dr C. Angelo (Umbria), Dr R. Cerone Dr L. Minicucci (Liguria), Dr G. Taccetti Dr L Zavatero (Tuscany), Dr C. Castellani (North Eastern)

Spain (4 regions): Dr CC Mejeras (Galicia), Dr MJ Alonso (Castilla-Leon), Dr S Gartner (Catalunia), Dr J Figuerola (Baleares)

Poland (1 region): Dr D. Sands

Wales : Dr I. Doull

Czech Republic (1 region): Dr M. Macek

UK: Dr KW. Southern (Liverpool) on behalf of national programme

Austria: Dr O. Bodamer on behalf of national programme

France: Dr A. Munck (AFDPHE) on behalf of national programme

Protocol strategies	Number
• Sweat testing for all infants with a raised IRT	1
• IRT/IRT measurement in the 4th week of life	5
• DNA analysis for common CFTR mutations on samples with a raised IRT (one programme used a second IRT measurement on infants with just 1 recognised mutation)	13

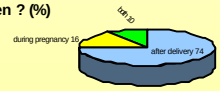
Information for all parents about CF NBS

➢ **Information recommended 58% mandatory 42%**

➢ **Parental informed consent for DNA analysis**

written consent	38%	written refusal	16%
nothing required	23%	oral consent	23%

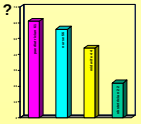
When ? (%)



How ?

• With a booklet	53%
(available in other community languages : 30%)	
• Orally only	47%
• Videotape	0%

By whom ?



Content ?

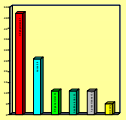
Very similar information are delivered accross Europe: all newborns are screened, the test performed is harmless, you can be contacted to control a biological value, an elevated IRT level may be a biological characteristic without clinical impact, CF is a genetic disease and, if applicable what being a carrier means.

Information for parents of infants referred to a CF center

How ?

• Phone call (day before ST 30%, same day ST 30%)	74%
• Letter only	16%
• Phone call + letter	32%
• Health visitor	11%

By whom ?



Content ?

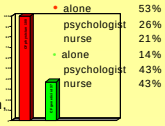
Information are mainly delivered with a reason of « borderline biological value to be controlled » (80%) or a technical problem with the biological sample (26%); 42% include the medical term « CF » and 11% try to avoid it. 74% explain the ST practical modalities. A phone number is at the family's disposal in 84%.

Information for parents of infants with CF

To whom ?

Both parents systematically	95%
Both parents and the baby	58%

By whom ?



Content ?

Consistent responses are delivered on CF genetic transmission, phenotype expression, treatment principles, CF multidisciplinary teams, improving prognosis and ongoing research, CF associations, bewareness of internet. A booklet is given to the parents in 63%.

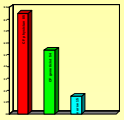
Information for parents of carriers

13 programmes include CFTR mutations analysis in their protocols

To whom ?

Both parents systematically 85%

By whom ?



Content ?

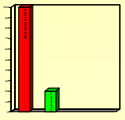
The baby carrier is safe, one of the parents has a CFTR mutation, DNA testing of both parents is suggested (70%), information should be forwarded to the child at adulthood (70%), a booklet is available (38%) and a letter is given (92%).

Information for parents of non-affected infants

To whom ?

Both parents or one out of two

By whom ?



Content ?

Information always emphasize the good health of the baby and the uselessness of further investigations, a letter is given in 63%.

Conclusion **The analysis of this survey suggests many consistent themes accross Europe. However, due to the large number of professionals involved, it is not possible to know precisely how information is given to parents without an ethically approved study. Our findings identified areas for improvment: 1) written information should be given to parents, at least for all parents on screening itself and for parents of infants with a positive CF diagnosis 2) guidelines and support for professionals in communicating with parents at the very initial step and further about initial raised IRT results.**