

ECFS Neonatal Screening Working Group (NSWG)

Report for the Board, May 2016

Core Committee

The WG is co-ordinated by a Core Committee of volunteers,

- Kevin Southern (UK) (Co-ordinator)
- Jürg Barben (Switzerland)
- Carlo Castellani (Italy)
- Jeannette Dankert-Roelse (Netherlands)
- Silvia Gartner (Spain)
- Nataliya Kashirskaya (Russia)
- Barry Linnane (Eire)
- Sarah Mayell (UK)
- Anne Munck (France)
- Dorota Sands (Poland)
- Olaf Sommerburg (Germany)

Supported by Victoria Winters (UK)

All outputs from the NSWG are reviewed by the Core Committee.

Anyone can apply to be on the Committee.

Aims of Working Group:

1. To support the implementation of newborn screening (NBS) for CF
2. To monitor performance and compare protocols to optimise effectiveness, whilst reducing negative impact
3. To encourage enrolment of all infants identified through NBS in clinical trials
4. To determine the optimal management of infants with an inconclusive diagnosis following newborn screening
5. Improving the processing of positive newborn screening results

The focus of work stream 5 will be on communication, establishing best practice for the different protocols that exist and disseminating this good practice. The workstream will also examine mechanisms for processing results, information for parent/carers and factors that impact on timeliness.

Broader objectives

1. To work in an open and inclusive manner
2. To encourage membership of the ECFS
3. To encourage participation from countries outside the EU

Progress report for each specific aim:

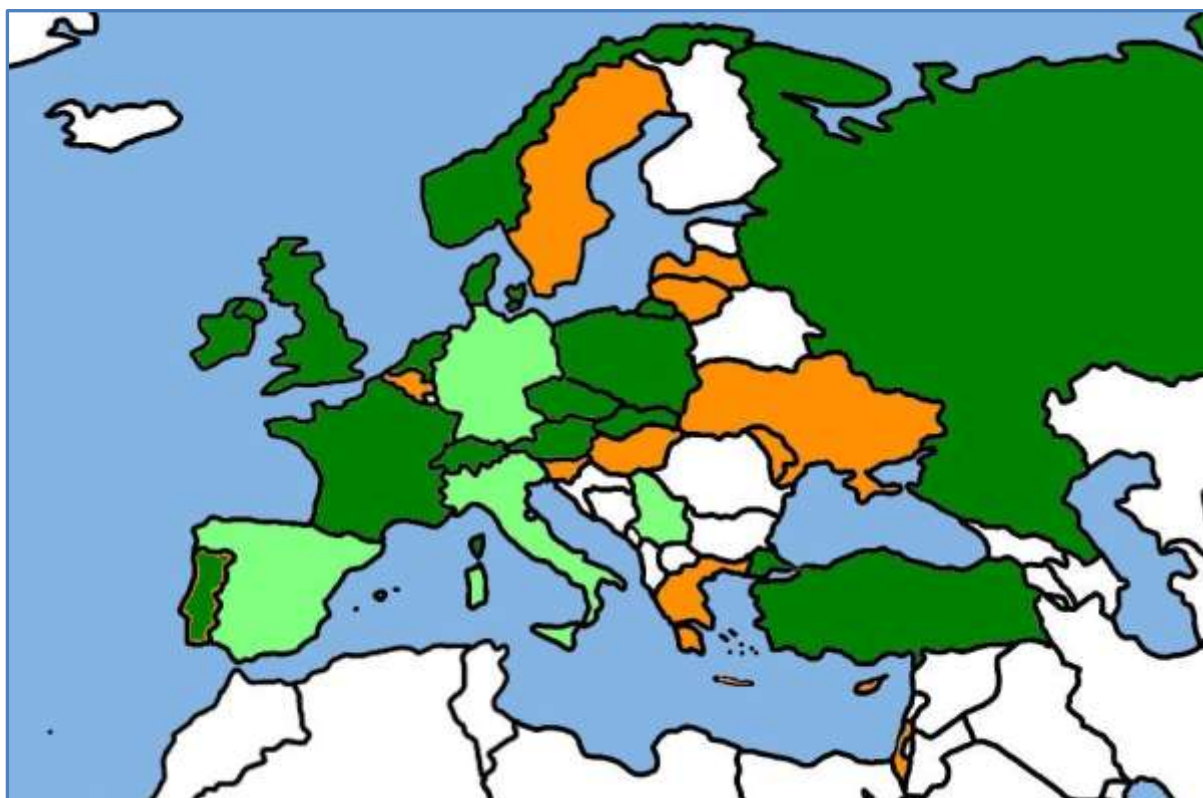
1. *To support the implementation of NBS for CF.*

This is the primary aim of the WG and is being addressed by the following:

- A. We are continuing to improve on our information network through the NSWG database. We now have 47 key country contacts in 36 countries.
- B. Support at international and national meetings. Meetings within the past year have included;
 - i. ECFS NSWG Annual Meeting, Brussels June 2015
 - ii. Diagnostic Network Working Group Annual Meeting, London February 2016
 - iii. UK Special Interest Group Meeting, Birmingham UK November 2015

Figure 1

CF Newborn Screening programmes across Europe 2016 (Denmark and Portugal have started screening nationally in the past few months).



Legend;	
Dark Green	National Programmes
Light Green	Regional Programmes (variable coverage)
Orange	NBS considered or pilot study
White	No plans for NBS

The national programme in Ukraine has discontinued because of the unrest in that country.

2. *To monitor performance and compare protocols*

We will address this specific aim through the following strategies:

- A. This year the NSWG has been collecting data from 2014-2015 with a poster “Updated Survey of Newborn Screening for Cystic Fibrosis in Europe” submitted to the NACFC 2016.
- B. There will also be a paper to follow on from the 2005 Survey with the 2014-2015 results.
- C. Based on the previous application to H2020 bid, there has been a modified bid submitted to the CF trust.
- D. Harmonisation work at the NACFC 2015.

3. *To encourage enrolment of all infants identified in clinical trials*

This aim is being addressed by the following:

- A. Establish close links with emerging registries. Provide database information for the purpose of encouraging recruitment to clinical trials, working closely with the ECFS Clinical Trials Network.
- B. CF START – A UK trial to examine routine use of anti-staphylococcal antibiotic prophylaxis (£1.4 million HTA award).

4. *To determine the optimal management of infants with an inconclusive diagnosis following newborn screening*

The WG published the paper on infants with the designation, CF Screen Positive, Inconclusive Diagnosis (Munck et al. 2015 Pubmed ID 25630966). This has had a major impact on the designation of these infants and provides a more consistent approach to management.

The next stage of this programme is to establish more consistency across the globe and to that end the CFF organised a group, in partnership with the ECFS NSWG to establish a clearer global approach to diagnosis, especially following newborn screening.

The global harmonisation exercise started in the summer of 2015. As well as considering infants with an inconclusive diagnosis after NBS, the exercise has also considered atypical presentations later in life. It has therefore been a joint exercise with the Diagnostic Network WG.

Reports and preliminary statements were presented at the NACFC in Arizona. Some statements were modified and the core writing group have been developing manuscripts over the winter. A draft manuscript (like an executive summary) is currently being reviewed by the CFF and the CF Centre Directors in the US.

Challenges achieved

1. An information network for associate members of the NSWG is now established.
2. The following meetings supported:
 - a. ECFS NSWG June 2015 – Brussels
 - b. ECFS DNWG February 2016 – London
 - c. UK SIG Group Meeting 2015 – Birmingham
 - d. The Harmonisation Group Meeting – NACFC 2015, Phoenix, USA
3. Performance recorded for the 2015 European CF NBS Survey, including:
 - a. Number of infants diagnosed with CF through NBS
 - b. Number of infants with an equivocal diagnosis following NBS
 - c. Number of assessments/sweat tests (and results when available)
 - d. Number of false negative NBS tests (true, meconium ileus or equivocal)
 - e. Incidence of CF
4. With the data from the ECFS NSWG Survey 2015, an abstract/poster has been submitted to the NACFC 2016 – “Updated Survey of Newborn Screening for Cystic Fibrosis in Europe”
5. CF EVE application modified and submitted to the UK CF Trust for alternate funding (preliminary application March 2016)
6. This year CF NBS has started in Portugal and Denmark

Challenges on-going

1. In countries with NBS, supply annual progress reports for a database. Ukraine situation demonstrates the fragility of continuing public health strategies during periods of unrest.
2. To record the protocol undertaken in each country that has a regional programme
3. Key workers will be encouraged to join the ECFS and become members of the Core Group
4. To liaise with national and European Registry Groups to collect longer term outcome data (some crossover with the Diagnostic Network on this project)
5. To develop and maintain resources to support implementation
6. ECFS NSWG Updated Survey 2015 – paper to be published

The following specific outputs will be expected;

Output 1

An annual report to the ECFS outlining the progress of newborn screening for CF across Europe.

Output 2

Quality improvement will be a focus of the annual meeting arranged in Switzerland for the 2016 ECFC.

Output 3

Working with the Registry to provide a clear outcome field for NBS and for diagnostic designation.

Output 4

Report from the international board assessing diagnostic designation after newborn screening (harmonisation exercise).

Output 5

Information resources for parent/carers on:

- Newborn screening and subsequent sweat testing
- Carrier status identified following newborn screening

In addition, the WG will continue core activities of supporting developing programmes, organising meetings and producing newsletters.