

ECFS Neonatal Screening Working Group (NSWG)

Report for the Board, April 2017

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 and anyone can apply to be a member or 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.*

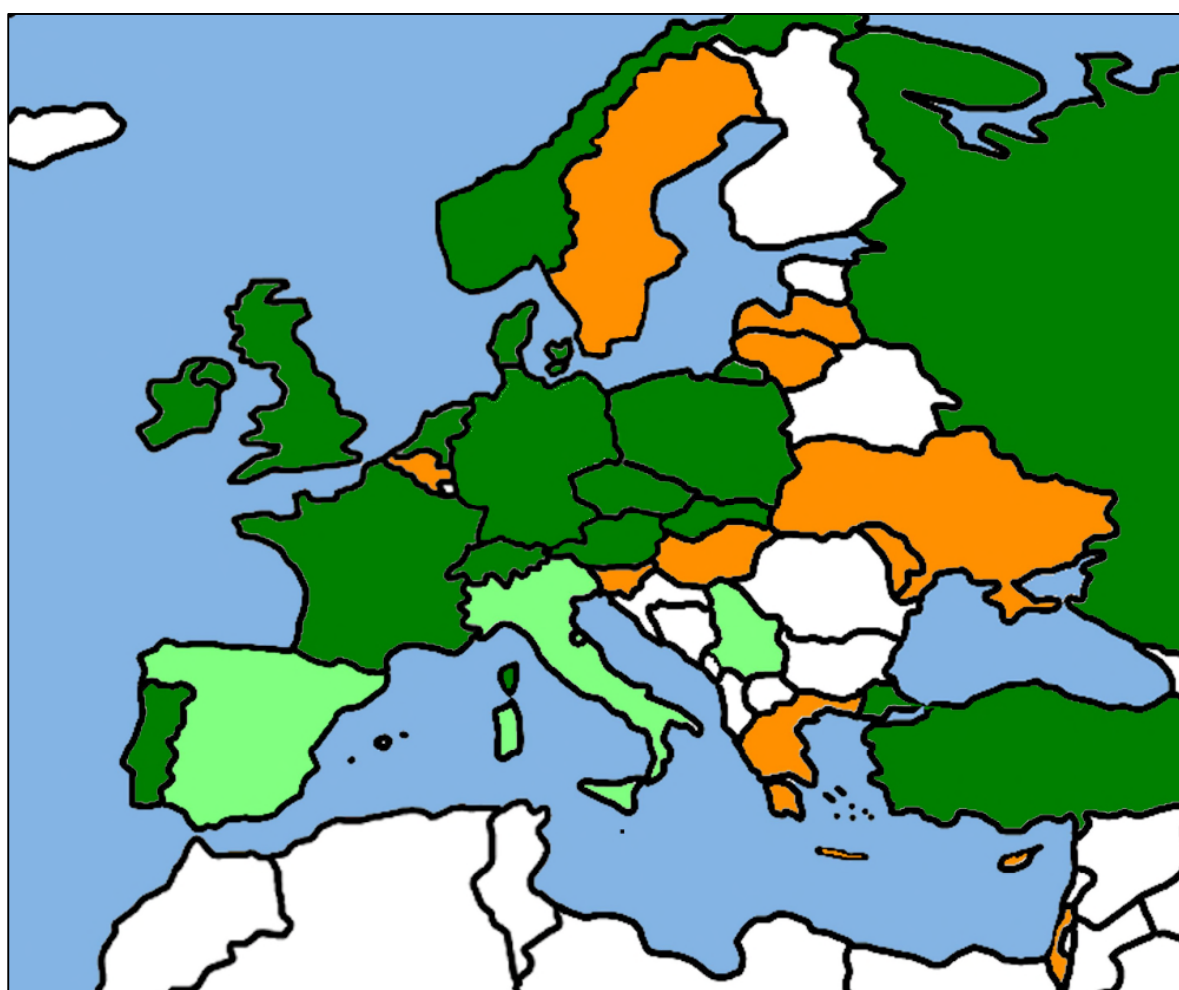
This is the primary aim of the WG and is being addressed by the following:

- 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, Basel, Switzerland - June 2016
 - ii. UK Special Interest Group Meeting, Birmingham, UK - November 2016
 - iii. Latvia, Riga (Baltic CF Meeting) - October 2016
 - iv. Harmonisation meeting at the NACFC, Orlando, USA – October 2016

- v. ECFS Steering Committee Meetings, Lisbon, Portugal - January 2017
- vi. Diagnostic Network Working Group Annual Meeting, Ljubljana, Slovenia - February 2017
- vii. Brussels, April 2017 (Belgian CF Meeting)
- viii. Lisbon, May 2017 (LEAD meeting)
- ix. ECFS NSWG Annual Meeting June 2017 Seville

Figure

CF Newborn Screening programmes across Europe at the end of 2016 (Germany started screening nationally at the end of 2016).



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

2. To monitor performance and compare protocols

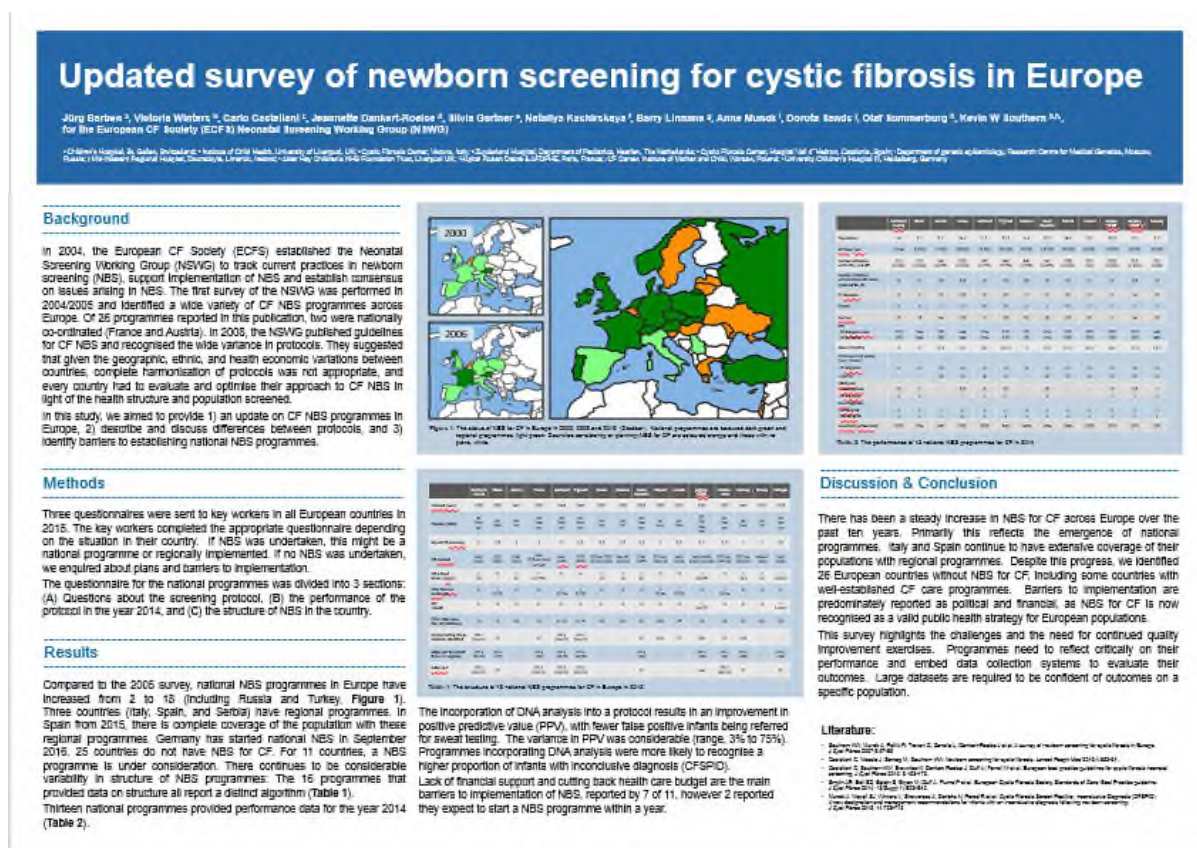
We will address this specific aim through the following strategies:

The paper “The expansion and performance of national newborn screening programmes for cystic fibrosis in Europe” Barben, J et al. was completed in August 2016 and published online in the Journal of Cystic Fibrosis at the end of December 2016.

Three questionnaires were sent to key workers in all European countries in 2015. The key workers completed the appropriate questionnaire depending on the situation in their country. If NBS was undertaken, it could be a national programme or regionally implemented. If no NBS was undertaken, we enquired about plans and barriers to the implementation of NBS.

The questionnaire for the national programmes was divided into 3 sections: (A) Questions about the screening protocol, (B) the performance of the protocol in the year 2014, and (C) the structure of NBS in the country.

Figure 2: The poster Updated survey of newborn screening for cystic fibrosis in Europe” presented at the NACFC, Orlando, USA by Jurg Barben



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). This trial will utilise NBS and the UK registry in an innovative manner, setting the template for future comparative effectiveness studies (see <http://www.cfstart.org.uk/>).

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 number 25630966). This has had a major impact on the designation of these infants and provides a more consistent approach to management. The NSWG worked with colleagues from across the globe 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.

Reports and preliminary statements were presented at the NACFC in Arizona, 2015 and a follow up meeting took place in Orlando 2016. The important papers from that exercise were published in the Journal of Pediatrics (PMID 28129812 and 28129811)

Challenges achieved

1. An information network for associate members of the NSWG is now established.
2. The following meetings supported over 12 months:
 - a. ECFS NSWG June 2016 – Basel, Switzerland
 - b. The Harmonisation Group Meeting – NACFC 2016, Orlando, USA??
 - c. UK Special Interest Group Meeting 2016 – Birmingham, UK
 - d. ECFS steering group meetings Lisbon, Portugal -Jan 2017
 - e. ECFS DNWG February 2017 – Ljubljana, Slovenia
3. The performance recorded for the 2015 European CF NBS Survey, has now been published as stated previously in this year's report
4. With the data from the ECFS NSWG Survey 2015, a poster and ePoster was presented at the NACFC 2016 – “Updated Survey of Newborn Screening for Cystic Fibrosis in Europe” by Jürg Barben.
5. A national clinical trial (CF START) has commenced.
6. CF NBS has gone from a regional programme to a national programme in Germany in the last quarter of 2016.

Challenges on-going

1. In countries with NBS, to supply annual progress reports for a database.
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. To support Belgium and Sweden to start National NBS.

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.

Appendix 1

References resulting from the Neonatal Screening Working Group

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