

ECFS Neonatal Screening Working Group (NSWG)

Report for the Board, May 2015

It has been another very busy year for the NSWG.

We submitted a full application to the PTC6 funding stream of Horizon 2020. The bid scored above the funding threshold (12.5) but unfortunately just outside the automatic funding cut-off. We have been placed on a reserve list and we are waiting to hear if funds become available to support the project

The CF EVE project aims to evaluate the effectiveness of NBS and optimise this public health strategy. We have developed an innovative methodology to achieve this goal. The aims of CF EVE are closely aligned to those of the NSWG and we will continue to explore alternative funding opportunities.

Group members and Associates

We are now approaching 500 associated members on the NSWG database. Associated members come from a number of backgrounds, including physicians and scientists from across the globe. The ECFS rules are quite clear that full WG members must also be members of the ECFS. We are currently revising our membership list with this in mind, but will continue to send newsletters, reports and updates to associate members. Over 100 of our members are ECFS members and many have joined the ECFS as a result of our WG activities.

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) (New)
- Barry Linnane (Eire)
- Sarah Mayell (UK)
- Anne Munck (France)
- Dorota Sands (Poland)
- Olaf Sommerburg (Germany)

- Supported by Victoria Winters (UK)

Specific 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 equivocal diagnosis following newborn screening

After extension of the WG for a further 3 year period, we have an additional specific aim

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

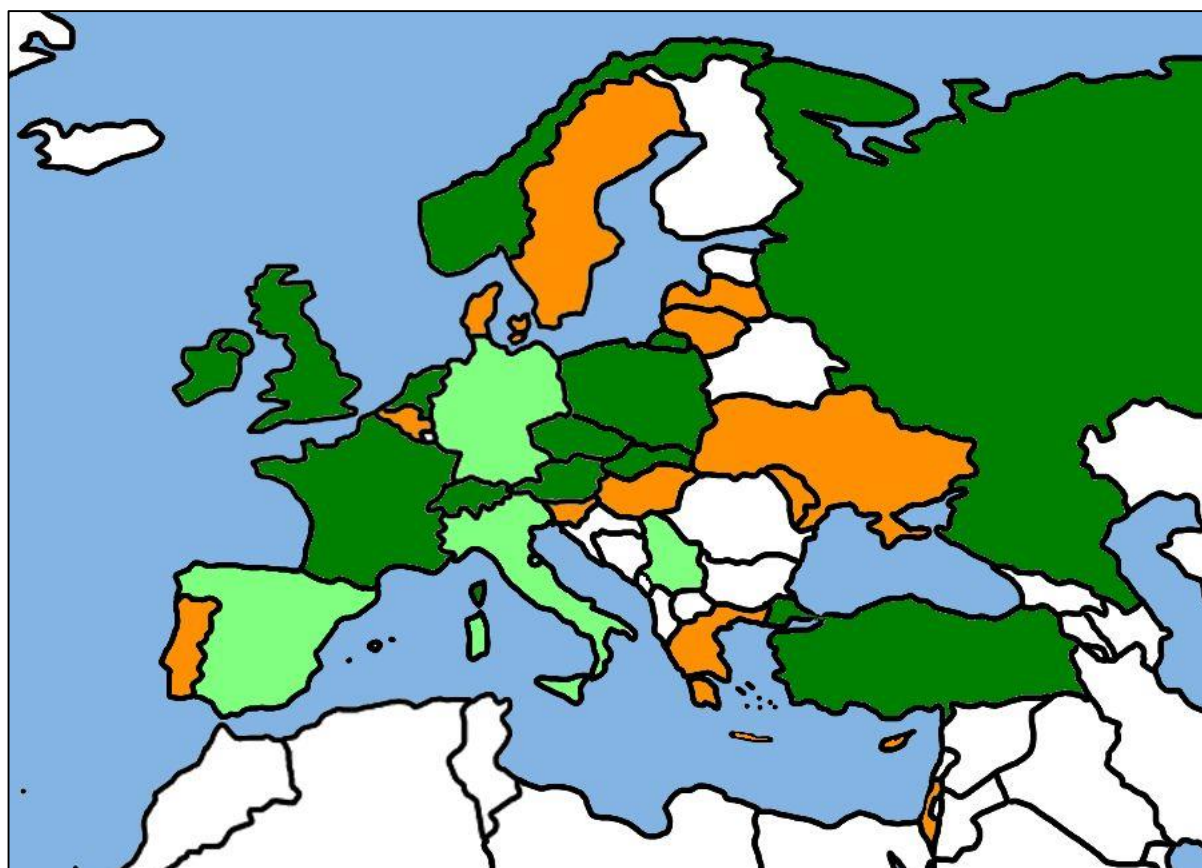
Progress report for each specific aim:

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 database. We now have 47 key country contacts in 36 countries.
- B. We have contributed to the update of the guideline produced by the Clinical Laboratory Standards Institute (Carlo Castellani, Olaf Sommerburg, Kevin Southern).
- C. Support at international and national meetings. Meetings within the past year have included;
 - i. ECFS NSWG Annual Meeting, Gothenburg June 2014
 - ii. Diagnostic Network Working Group Annual Meeting Warsaw February 2014
 - iii. The Latin America Meeting Gothenburg, June 2014
 - iv. The German CF Conference, Mainz, November 2014

Figure Status of newborn screening programmes across Europe



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

A more detailed report on the international status of NBS is available on request.

2. *To monitor performance and compare protocols*

We will address this specific aim through the following strategies

- A. The database is in constant use as a functional tool to interact with WG members
- B. The Standards of Care for NBS have been developed. These were produced by a Delphi consensus methodology and reflect the comments of all members of the Core Committee.
- C. The Working Group has been involved in the application of Horizon 2020 bid.

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. A £1.4 million application has been submitted to the UK HTA (in final stages of review).

4. *To determine the optimal management of infants with an equivocal 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 will have a major impact on the designation of these infants and provide 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 have organised a group, in partnership with the ECFS NSWG to establish a clearer global approach to diagnosis, especially following newborn screening. This group will meet in Phoenix at the NACFC 2015, with preparatory work expected for the summer.

Challenges achieved

1. An information network for associate members of the NSWG is now established
2. Newborn Screening for Cystic Fibrosis - Standards of Care - completed and published
3. CFSPID paper, completed and published
4. Data for the ECFS NSWG Survey 2013 collected – to be presented at the poster session at the ECFC
5. Ongoing regular WG newsletters
6. Full application to EU for the Horizon 2020 fund - Antenatal and newborn screening for cystic fibrosis (CF); Evaluating Validity and Effectiveness (CF-EVE)

Challenges on-going

1. In countries with NBS, supply annual progress reports for a database
2. To record the protocol undertaken in each country that has a regional programme
3. To record performance as determined by population screened and results (information being gathered by the 2013 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. Key workers will be encouraged to join the ECFS and become members of the Core Group
5. To liaise with national and European Registry Groups to collect longer term outcome data (some crossover with the Diagnostic Network on this project)
6. To develop and maintain resources to support implementation

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. The international Board is a group comprising membership from all continents and co-ordinated by the Working Group and the CFF.

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.