

Neonatal Screening Working Group (NSWG)

Board Report April 2013 (Amended November 2013).

It has been an exciting year for the NSWG after ratification as an official ECFS Working Group in September 2012. Since September the WG has had administrative support (temporary contract with the University of Liverpool for 3 years). This has provided great impetus to the group and has enabled the WG to address the specific aims listed below.

Group members

We now have over 350 members on the NSWG database. Members come from a number of backgrounds, including numerous physicians from across the globe. The group has been widely advertised through the WG newsletters and we have been actively seeking relevant CF NBS key contacts from around Europe and beyond. Membership is broad and includes scientists from screening laboratories that run newborn screening programmes. Two of the wider objectives of the WG are to encourage all members to join the ECFS and to broaden membership to professionals from outside of Europe, in particular countries from South America. We have already have contact with Uruguay and Argentina but are now trying to make contact with Chile and Brazil.

Core Committee

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

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

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

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 have now established a regular newsletter to discuss NBS and report on progress in specific countries. So far this has included Switzerland, Spain, Austria, Ireland, Norway, the Netherlands and France.
- B. Support at international and national meetings. Meetings within the past year have included;
 - i. Orlando, USA. Representation at US SIG meeting (10/10/12) – KS has reported on this within the second newsletter
 - ii. Budapest, Hungary. Presentation at the ISNS (5/11/12)
 - iii. Jerusalem, Israel, workshop at the DN meeting (14/2/13)
 - iv. Belek, Turkey, presentations at Turkish Respiratory Meeting (2-5/4/13)
 - v. Verona (ECFS Standards meeting), (12-13/4/13)
 - vi. ECFS NSWG Annual Meeting, Lisbon, Portugal. A satellite meeting at ECFC (12/6/13)
- C. We are establishing an information network through the database. We now have 37 key country contacts, with the database now holding over 350 members which is constantly growing and being updated
- D. We have identified key workers in most countries without NBS and aim to maintain a separate database of progress in those countries (updated every 3 months)
- E. We have improved the information available on the NSWG webpage on the ECFS website, including a repository for newsletters
- F. We aim to produce a pack for countries with emerging programmes with all the necessary information (both hard copy and virtual)

2. *To monitor performance and compare protocols*

We will address this specific aim through the following strategies

- A. The database is constantly in use as a functional tool to interact with WG members
- B. Key workers in each country with NBS have been identified. We are now in the process of developing and maintaining a separate database of performance, updated annually.
- C. Three questionnaires were developed with support from the Core Committee. Form A is a survey of countries without NBS, collecting data on plans and obstacles to NBS. Forms B & C are to collect data from countries and regions that are currently screening.
- D. The forms were distributed in February 2013. Supplementary forms have been sent out to further contacts in March and April. Forms were also sent to the US and Canada, with support from the CFF.
- E. Forms have been returned from 45 contacts
 - 1. 16 from countries without NBS
 - 2. 15 from countries with national programmes
 - 3. 14 from countries with regional programmes
 - 4. At least 10 more forms expected from regions and countries with CF NBS

- F. Preliminary results will be presented at the NSWG meeting in Lisbon.
- G. 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.

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

This specific 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. Lobbying organisations to highlight the issue of clinical trials in CF, in particular on the large number of infants recognised through NBS.
- C. To support the design and implementation of pragmatic clinical trials. A proposal has been submitted to the UK HTA (funding agency) for a large pragmatic trial examining anti-staphylococcal antibiotic prophylaxis (CF-START)

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

This aim is being addressed by the following:

- A. The survey of current practice in established NBS programmes (covered by the 2013 Survey and including US sites via the SIG), audited by the standards published in 2009
- B. We presented the preliminary findings in Lisbon
- C. Core statements are being produced to initiate a Delphi consensus process to establish clear guidance on the management of these infants (Shawcross, Southern and Mayell).
- D. We are working with the CFF Special Interest Group (NBS) to form a global consensus on terminology and management of these infants. Dr Parad spoke in Lisbon and will be a key contributor to this Delphi methodology.
- E. A statement on equivocal diagnosis is included in the ECFS Standards of Care document.

Challenges achieved

1. An information network for members of the NSWG has been achieved
2. In countries without NBS, we have identified a key worker
3. In countries with NBS, we have identified a key worker
4. Populating the WG webpage with up-to-date information
5. Regular WG newsletter

Challenges on-going

1. In countries without NBS, monitor progress (3 monthly to record plans for implementation)
2. In countries with NBS, supply annual progress reports for a database
3. To record the protocol undertaken in each country (2013 Survey)
4. 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
5. Key workers will be encouraged to join the ECFS and become members of the Core Group

6. To liaise with national and European Registry Groups to collect longer term outcome data (some crossover with the Diagnostic Network on this project)
7. To survey practice with respect to management of infants with an equivocal diagnosis (consensus process)
8. To develop and maintain resources to support implementation
9. To support a further consensus project to achieve Specific Aim 4