NEONATAL SCREENING WORKING GROUP (NSWG)

Year of report: **2021**	
Name of Working Group: **Neonatal Screening Working Group (NSWG)**	Date of initial approval of working group: **2004**

Coordinator name: **Jürg Barben**

Vice coordinator name: **Kevin Southern**

Assistant name: **Daria Berger**

Long term aims of the working group (maximum 100 words) including estimated achievement date:

Original aims of the Working Group

- To support the implementation of newborn screening (NBS) for CF (ongoing process; implemented in Western Europe)
- To monitor performance and compare protocols to optimise effectiveness, whilst reducing negative impact (first survey published 2017)
- To encourage enrolment of all infants identified through NBS in clinical trials (ongoing process)
- To determine the optimal management of infants with an inconclusive diagnosis following newborn screening (ongoing process)
- Improving the processing of positive newborn screening results (ongoing process)

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 work stream will also examine mechanisms for processing results, information for parent/carers and factors that impact on timeliness.

Additional objectives for the NSWG from 2018 onwards

To determine key outcome measures to evaluate the performance of CF NBS

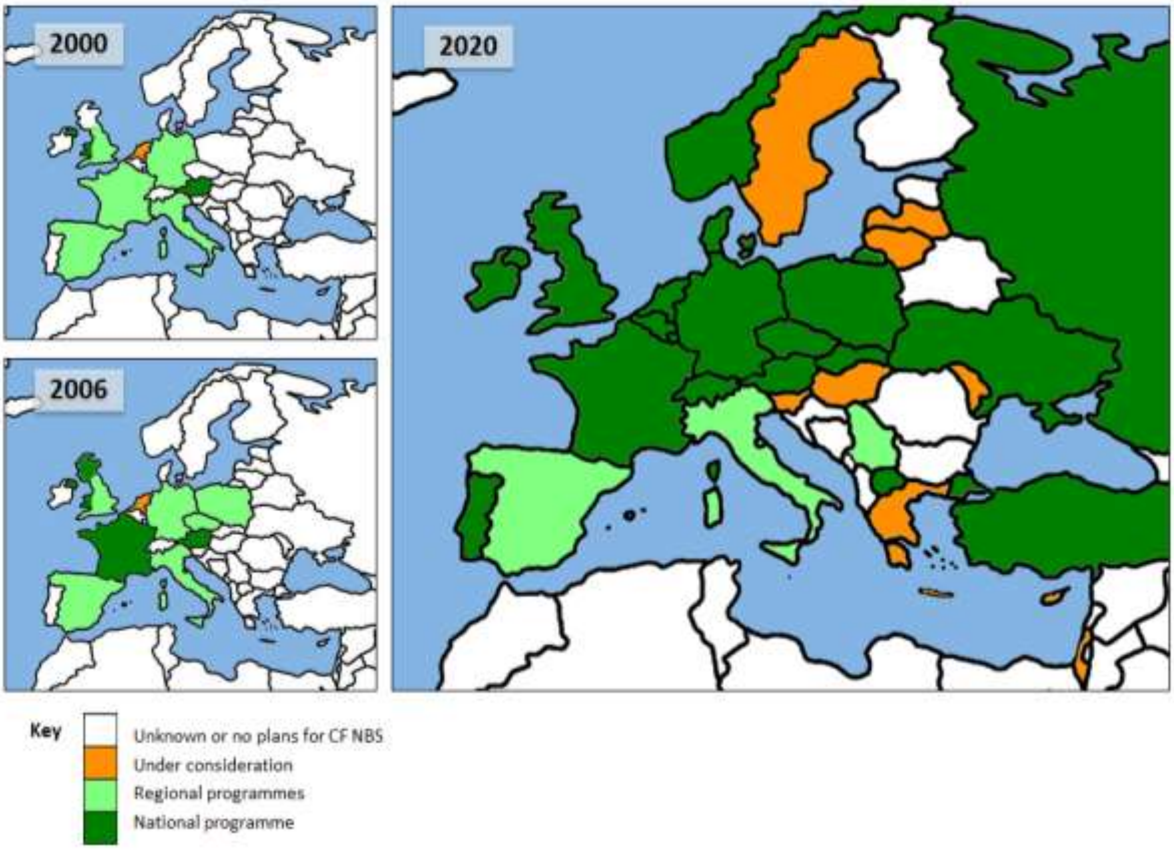
- To establish guidance and quality ranking on the collection of NBS outcome data
- To assess knowledge of CFSPID in Europe
- To provide resources to improve the evaluation and management of infants with CFSPID
- To work with the ECFS Registry group to clarify definition and recording of CFSPID outcomes

Outcomes already achieved (maximum 100 words):

- Implementation of newborn screening (NBS) for CF in most countries in Europe:

This is the primary aim of the NSWG. We are continuing to improve on our information network through the NSWG database. We have now over 50 **key workers** in 45 countries.

Figure 1: CF Newborn Screening programmes across Europe from 2000 until 2020



- New guidelines on the management of children with CRMS/CFSPID (started 2020, published 2021)
- Monitoring performance and compare protocols to optimise effectiveness, whilst reducing negative impact (first survey published 2017)
- Summary of current knowledge around newborn screening for CF (Publication of a book 2020)

Report for this year (max 1000 words)

- short term goals for the year
- current number of members
- measures taken to encourage ECFS membership
- outcomes/achievements (e.g. meetings, activities, website development, awards, publications etc).

The core group of the NSWG consists of 12 members: Jürg Barben (Coordinator, Switzerland), Kevin Southern (past coordinator, UK), Carlo Castellani (founder of the group, Italy), Anne Munck (France), Olaf Sommerburg (Germany), Barry Linnane (Ireland), Dorots Sands (Poland), Silvia Gartner (Spain), Sarah Mayell (UK), Nataliya

Kashirskaya (Russia), Karin de Winter-de Groot (Netherlands), and Marijke Proesmans (Belgium). All members of the core committee are members of the ECFS. In addition, the NSWG has a total of >50 key contacts in 45 countries and a database of nearly 500 email addresses.

Since the planned European CF Conference in Lyon in June 2020 could not take place due to the Corona pandemic, the annual meeting of the NSWG was also cancelled. Instead, several video conferences have taken place and the work has been done online. In the last year, there were a total of four online meetings of the core committee group and about 10 working meetings of the "Core Working Group" (Anne Munck, Kevin Southern, Carlo Castellani, Jürg Barben) to develop the *Updated guidance on the management of children with CRMS/CFSPID* and to define *key outcomes to evaluate performance of newborn screening programmes for cystic fibrosis* have taken place. Both projects were successfully completed and have already been published online in the *Journal of Cystic Fibrosis*. We also have updated the ECFS NSWG website.

In 2019, we were asked as a group to write and publish a series of review articles on NBS for CF in collaboration with the *International Journal of Newborn Screening* (IJNS). After consultation with the *Journal of Cystic Fibrosis*, who assured us that such a series of review articles would not fit into the current publication programme of the Journal, we started to collaborate with the IJNS and were able to attract well-known authors for this project (most members of the ECFS NSWG). Meanwhile all articles have been completed and published in the IJNS. We then turned it into a book "Newborns Screening for Cystic Fibrosis", which was published 2020.

The focus of the current work is

- To conduct another European survey on the performance of the CF newborn screening (NBS) programmes with the new defined outcome parameters (new survey started, finished by the end of 2021)
- To find out by means of a survey how professionals deal with unclear diagnoses and inform parents (international survey ongoing, finished by the end of 2021)
- To develop a strategy to detect false negatives (children with CF not detected by screening) as quickly and reliably as possible (started early this year, topic at ECFC online 2021, hopefully finished by the end of 2022)
- To set up a CFSPID-Registry (discussed at the next NSWG meeting, hopefully completed by the end of 2023)

Aims for the coming year (please state year) (max 50 words):

- To establish guidance and quality ranking on the collection of NBS outcome data
- To assess knowledge of CFSPID in Europe
- To provide resources to improve the evaluation and management of infants with CFSPID
- To work with the ECFS Registry group to clarify definition and recording of CFSPID outcomes and set up a CFSPID registry

Summary (maximum 100 words):

Despite the Covid pandemic, last year was a very productive year for the NSWG, even though we did not have assistant support. Since March 2021, we now have support from Daria Berger from the Institute for Social and Preventive Medicine (ISPM) in Berne (Switzerland) and we hope that this will last for as long as possible.

Breakdown of expenses (please include total amount received as well as expenditure and, if applicable, the outstanding balance (Euros)):

In 2020, only the new book "Newborn Screening for CF" was co-financed (all authors received a free book). Otherwise, we have not used any money from ECFS in the year 2020.

Budget amount requested for next year (please give the amount in Euros and the year):

Before 2019, the ECFS NSWG has received 10'000 Euro support per year to fund the activities of the assistant of Prof. Kevin Southern (Vicky Winters). As Vicky Winters has also ended her work with Prof Southern's step back in June 2019, we did not receive any money from ECFS because the position of the assistant was vacant. Since March 2021, the position is filled by Daria Berger (responsible for the CF newborn screening database in Switzerland), who already did the online survey to evaluate the performance of CF screening programs in Europe. So we hope to get the 10’000.- Euro again to finance Daria Berger’s work.