

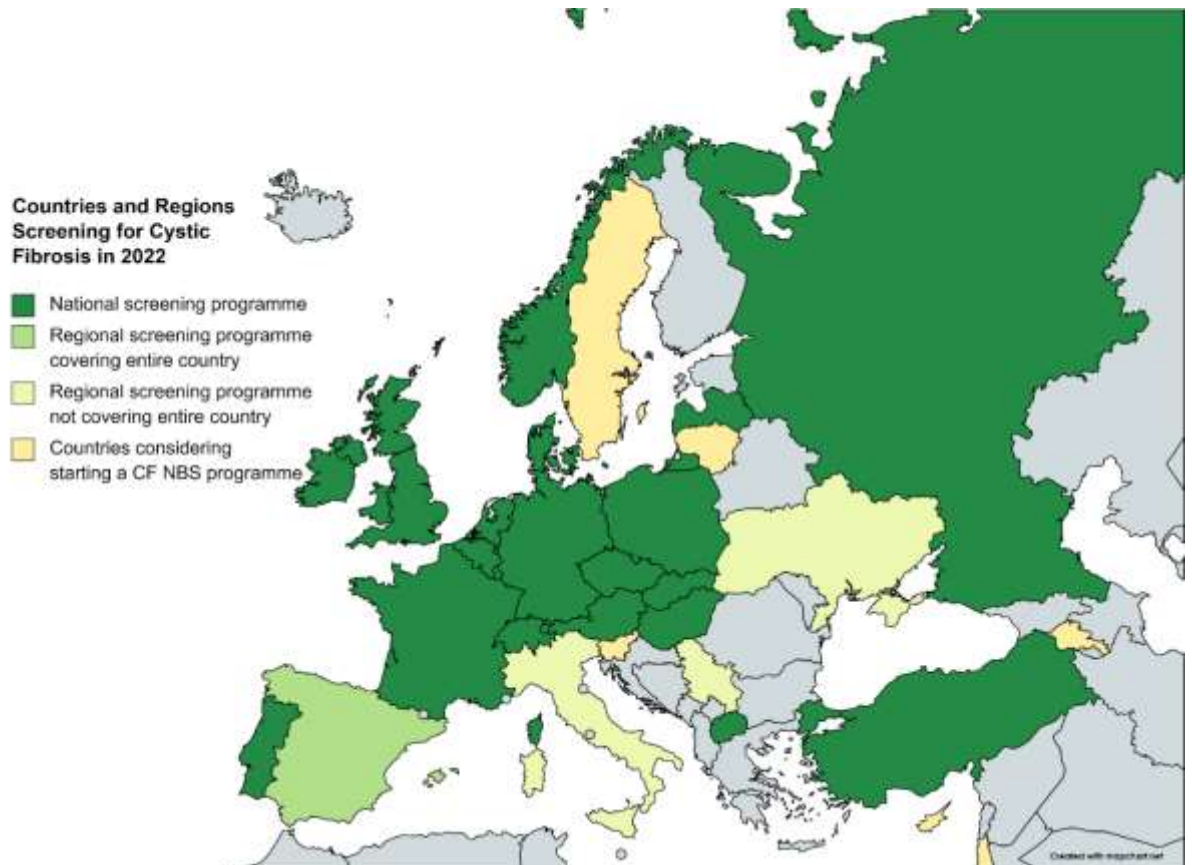
Year of report: <b>2022</b>	
Name of Working Group: <b>Neonatal Screening Working Group (NSWG)</b>	Date of initial approval of working group: <b>2004</b>
Contact details of coordinator, vice coordinator and assistant (if applicable) including ECFS membership numbers: Coordinator name: <b>Jürg Barben</b> ( <a href="mailto:juerg.barben@kispisg.ch">juerg.barben@kispisg.ch</a> ) ECFS Membership number: <b>4095</b> Vice coordinator name: <b>Kevin Southern</b> ( <a href="mailto:K.W.Southern@liverpool.ac.uk">K.W.Southern@liverpool.ac.uk</a> ) ECFS Membership number <b>439</b> Assistant name: <b>Daria Berger</b> ( <a href="mailto:daria.berger@ispm.unibe.ch">daria.berger@ispm.unibe.ch</a> ) ECFS Membership number: not yet member	
Long term aims of the working group (maximum 100 words) including estimated achievement date: <p><b>Original aims of the Working Group</b></p> <ul style="list-style-type: none"> <li>▪ To support the implementation of newborn screening (NBS) for CF (<a href="#">ongoing process; implemented in Western Europe</a>)</li> <li>▪ To monitor performance and compare protocols to optimise effectiveness, whilst reducing negative impact (<a href="#">first survey published 2007 by Southern et al., second 2017 by Barben et al., current survey in preparation for publication</a>)</li> <li>▪ To encourage enrolment of all infants identified through NBS in clinical trials (<a href="#">ongoing process</a>)</li> <li>▪ To determine the optimal management of infants with an inconclusive diagnosis following newborn screening (<a href="#">ongoing process</a>)</li> <li>▪ Improving the processing of positive newborn screening results (<a href="#">ongoing process</a>)</li> </ul> <p>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.</p> <p><b>Additional objectives for the NSWG from 2018 onwards</b></p> <p>To determine key outcome measures to evaluate the performance of CF NBS</p> <ul style="list-style-type: none"> <li>▪ To establish guidance and quality ranking on the collection of NBS outcome data</li> <li>▪ To assess knowledge of CFSPID in Europe</li> <li>▪ To provide resources to improve the evaluation and management of infants with CFSPID</li> <li>▪ To work with the ECFS Registry group to clarify definition and recording of CFSPID outcomes</li> </ul>	

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, updated 2022**



- 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, current survey for publication](#))
- Summary of current knowledge around newborn screening for CF ([Publication of a book 2020](#))

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 and perform an updated *European Survey on the performance of newborn screening programmes for cystic fibrosis in Europe* with the new defined outcome parameters (published 2021).

#### **The focus of the current work is**

- To evaluate and publish the European survey on the performance of the CF newborn screening (NBS) programmes with the new defined outcome parameters ([new survey started, evaluation and publication is on the way](#))
- To find out by means of a survey how professionals deal with unclear diagnoses and inform parents ([international survey, publication is on the way](#))
- To develop a strategy to detect false negatives (children with CF not detected by screening) as quickly and reliably as possible ([project started 2021, topic at the ECFC online 2021, hopefully finished by the end of 2022](#))
- To set up a CFSPID-Registry in collaboration with the ECFS Registry Group (lead by Andreas Jung; [we already had many Zoom-conferences and we will further discuss it at the next NSWG meeting, hopefully implemented in the next few years](#))

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

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

Summary (maximum 100 words):

Despite the covid-19 pandemic and the lack of opportunities to meet in person, there has been a lively exchange via Zoom in the group. Fortunately, last year the new assistant (Daria Berger) was able to start work and was very active in the new European survey on the performance of the CF NBS. We hope to publish our last survey on the performance of the CF NBS this summer.

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

In 2021, the European survey on the performance of the CF NBS was funded with 10'000.- Euro.

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



The statistical analysis and publication (Daria Berger, ISPM Berne) will certainly cost another 5,000 euros. For the establishment of a CFSPID registry, 10,000 to 20,000 euros are necessary after consultation with Dr. Andreas Jung (Zoom-Meeting on April 7, 2022).