

Neonatal Screening Working Group (NSWG)

Coordinator: **Jürg Barben**
Vice coordinator: **Kevin Southern**
Assistant: **vacant** (Daria Berger left our group in summer 2022 due to pregnancy)

Date of initial approval of working group:
2004

Long term aims of the working group:

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 2007 by Southern et al., second 2017 by Barben et al., current survey in preparation for publication](#))
- 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](#))

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, building up a CFSPID registry

Outcomes already achieved:

- Implementation of NBS for CF in most countries in Europe (see Figure 1), which is the primary aim of the NSWG. We are continuing to improve on our information network through the NSWG database.
- Monitoring performance and compare protocols to optimise effectiveness, whilst reducing negative impact ([first survey published 2017, second survey published 2022](#))
- New guidelines on the management of children with CRMS/CFSPID ([published 2021](#))
- Defining key outcomes to evaluate performance of NBS programmes for CF ([published 2021](#))
- Summary of current knowledge around newborn screening for CF ([book published 2020](#))

Figure 1: CF Newborn Screening programmes across Europe, updated end 2022

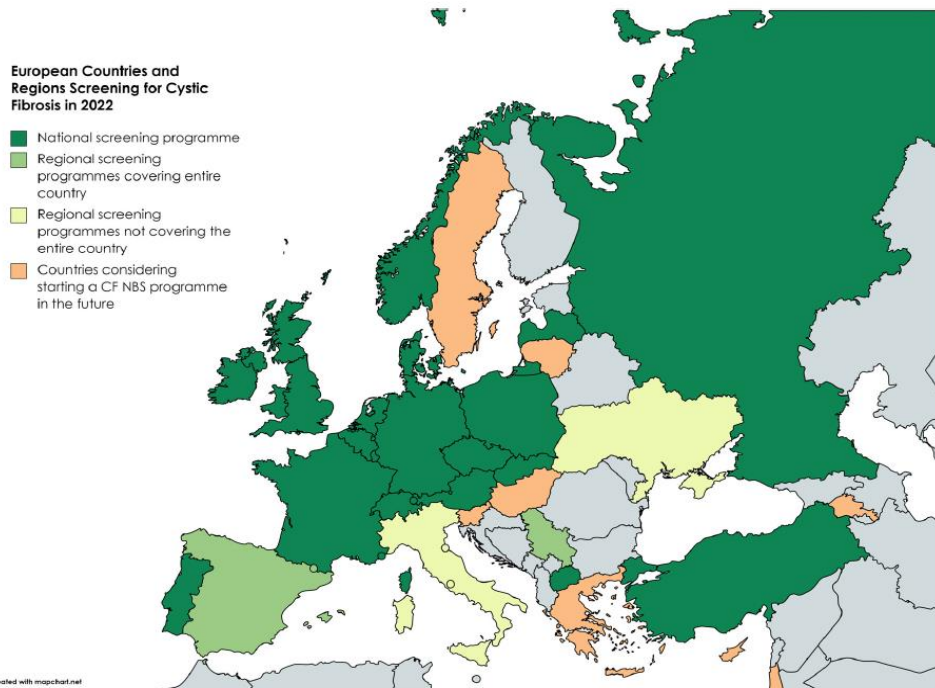


Fig. 1. The status of NBS for CF in Europe 2022. National programmes are coloured dark green and regional programmes, light green. Countries considering or planning NBS for CF are coloured light orange and those with no plans, light grey.

Report for this year

Sarah Mayell (UK) has left the core group in 2022, Bülent Kardag from Turkey has replaced her. The core group of the NSWG now 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), Bülent Kardag (Turkey), Nataliya Kashirskaya (Russia), Karin de Winter-de Groot (Netherlands), and Marijke Proesmans (Belgium). All members of the core committee are members of the ECFS.

The NSWG has a total of >50 key contacts in 45 countries and a database of nearly 500 email addresses.

Since the last European CF Conference in Rotterdam in June 2022, the core group had two video conference online and one meeting in person at the ECFS DNWG Meeting in Montpellier in February 2023. In addition, we had about 10 working meetings of the “Core Working Group” (AM, KS, CC, JB) to finalise the publication of the updated *European Survey on the performance of newborn screening programmes for CF in Europe* with the new defined outcome parameters in 2021.

We also had several video conferences together with colleagues from the ECFS Patient Registry Group (lead by Andreas Jung) to define key registry parameters for the planned CFSPID-Registry.

Another important piece of work was the collaboration with the DNWG to better define the diagnosis of CFTR related disorder (CFTR-RD) and to describe which CFSPID children will develop into a CFTR-RD.

The focus of the current work is

- To develop an optimal strategy to detect false negatives (children with CF not detected by NBS) as quickly and reliably as possible (project started late 2021, topic at the ECFS NSWG meeting in Vienna, hopefully finished by mid 2023 to start a new survey in Europe)
- To implement a CFSPID-Registry in collaboration with the ECFS Patient Registry Group (we already had many video conferences and we hope to implement this registry within the next two years)
- To find out by means of a survey how professionals deal with unclear diagnoses and inform parents (international survey)

Aims for the coming year:

- To establish guidance and quality ranking on the collection of NBS outcome data with the focus on false negatives NBS results
- To work with the ECFS Registry group to implement a CFSPID registry (maybe in combination with a CFTR-RD-Registry)
- To provide resources to improve the evaluation and management of infants with CFSPID (ongoing)
- To assess and improve knowledge of CFSPID in Europe (ongoing)

Summary:

Last year, we analysed and published our updated survey with the new key outcome parameter on the performance of newborn screening programmes in Europe. In parallel, we have moved forward with a new project on how best to define and record false-negative NBS results and plan to conduct a survey later this year. We have also held several meetings with the ECFS PRG to define the parameters for a CFSPID registry and are in the process of implementing it.

Unfortunately, the new NSWG assistant left us early in the summer due to pregnancy and we are looking for a new assistant.

Breakdown of expenses (total amount received as well as expenditure and outstanding balance):

In 2021-22, the European survey on the performance of the CF NBS was funded with 10'000 Euro. Currently, 22,000 Euro are on the account (10,000 planned for the development of the CFSPID registry)

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

For the implementation of a CFSPID registry, 10,000 to 20,000 euros are necessary after consultation with Andreas Jung (Chair of the ECFS PRG).