

## Neonatal Screening Working Group (NSWG)

Year of report:  
**2024**

Date of initial approval of working group:  
**2004**

Coordinator name: **Jürg Barben**

Vice coordinator name: **Kevin Southern**

Assistant name: **vacant** (Daria Berger left our group in summer 2022 due to pregnancy, not currently filled)

Long term aims of the working group:

### *Original aims of the Working Group*

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

### *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

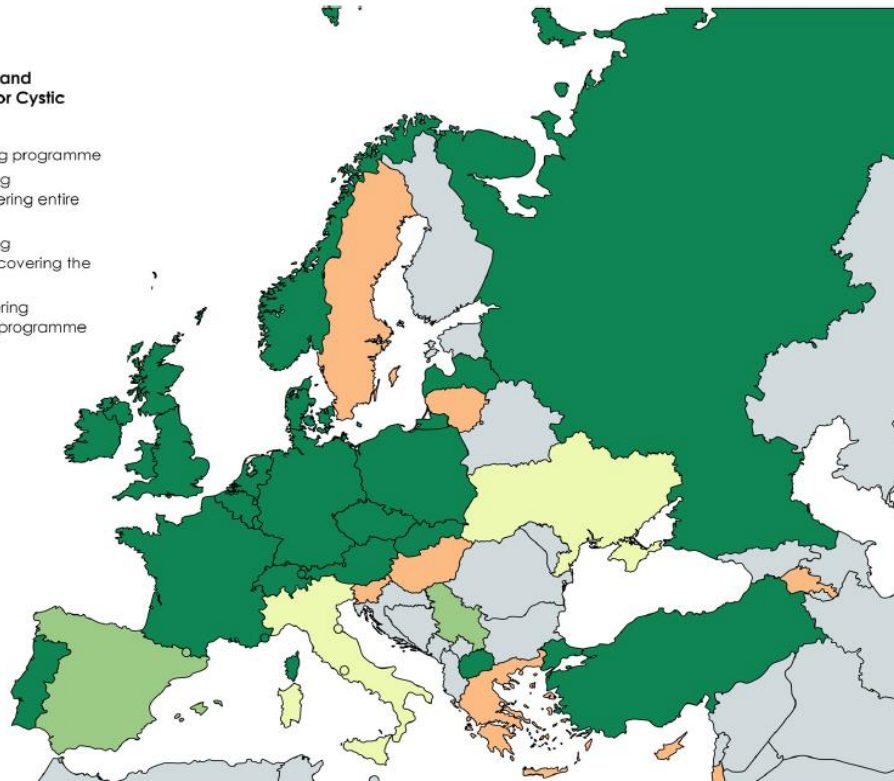
Outcomes already achieved:

- 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 2007](#), [second published 2017](#), [third published 2023](#))
- 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](#))
- CF Cases missed by NBS - Towards a Consistent Definition and Data Acquisition ([published 2023](#))

**Figure 1: CF Newborn Screening programmes across Europe, mid 2023**

**European Countries and Regions Screening for Cystic Fibrosis in 2022**

- National screening programme
- Regional screening programmes covering entire country
- Regional screening programmes not covering the entire country
- Countries considering starting a CF NBS programme in the future



### Report for this year

The core group of the NSWG now consists of 12 members: Jürg Barben (Coordinator, Switzerland), Kevin Southern (vice coordinator, UK), Carlo Castellani (founder of the group, Italy), Anne Munck (France), Olaf Sommerburg (Germany), Barry Linnane (Ireland), Dorota Sands (Poland), Silvia Gartner (Spain), Bülent Karadag (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. We have now updated the address of all our members and interested parties and now have more than 175 updated email addresses, of whom around 70% are ECFS members.

Since the last European CF Conference in Vienna in June 2023, the core group had three video conferences online. In addition, we had about 10 working meetings of the “Core Working Group” (AM, KS, CC, JB) to finalise the publication *CF Cases missed by NBS - Towards a Consistent Definition and Data Acquisition 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 implement the planned CFSPID-Registry and to build a new ECFS Working Group.

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 or may develop into a CFTR-RD, which was published early this year (*ECFS standards of care on CFTR-related disorders: Towards a comprehensive program for affected individuals*). In addition, in collaboration with the DNWG, we have published a letter entitled “Risk of false newborn screening after intra-uterine exposure to the new modulator therapy ETI”.

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

- To support the implementation of NBS programmes for CF in Eastern Europe
- To establish guidance and quality ranking on the collection of NBS outcome data with the focus on false negatives NBS results (children with CF not detected by NBS) ([project started late 2021, publication in 2023, new survey planned by mid-2024](#))
- To monitor performance and compare protocols to optimise effectiveness, whilst reducing negative impact ([next survey planned 2025/26](#))
- To assess and improve knowledge of CFSPID and to provide resources to improve the evaluation and management of infants with CFSPID in Europe ([ongoing](#))
- To find out by means of a survey how professionals deal with unclear diagnoses and inform parents ([international survey](#))
- To implement a CFSPID-Registry in collaboration with the ECFS Patient Registry Group ([Outcome parameter already defined by end of 2023, platform planned, financing initiated, launch planned for mid-2024, building a new ECFS Working Group, applied 2024](#))

### Aims for the coming year:

- To build up a new ECFS CFSPID Working Group to implement a CFSPID registry
- To establish guidance and quality ranking on the collection of NBS outcome data with the focus on false negatives NBS results
- To improve the evaluation, management and knowledge of infants with CFSPID
- Succession plan for the current coordinator (term finishes by 2025)

As the current Coordinator (Jürg Barben) will have completed his permitted term in 2025, a new Coordinator will be elected by the Core Committee by May 2025. The current deputy (Kevin Southern) will step down as he will be involved in a new ECFS Working Group (CFSPID). His deputy will be appointed at the next meeting in Glasgow in June (proposal: Anne Munck)

### Summary:

Last year, we analysed and published our updated survey with the new key outcome parameter on the performance of NBS programmes in Europe. In parallel, we have moved forward with a new project on how best to define and record false-negative NBS results with a publication in the IJNS. We have started a new survey early this year. We have also held several meetings with the ECFSPRG to define the parameters for a CFSPID registry and are in the process of implementing a CPSPID registry by building a new ECFS CFSPID Working Group.

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

In recent years, the NSWG has received 10,000 euros per year. We have used most of the money for the annual conference. Temporarily we had an assistant (Daria Berger) who also did the data collection and processing of the last NBS survey in Europe, which cost around 10,000 euros. We also have budgeted 10,000 Euro for the next European survey on the performance of the existing CF-NBS programmes planned in 2025/26 (mainly for collecting data and statistical analysis). We believe that the annual budget of 10,000 euros will cover the costs of the working group in future.

		Expenses		Income	
		DKK	Euro	DKK	Euro
2023	ECFS			74'500.00	10'000.00
	NSWG mtg Montpellier	3'523.85	473.00		
	VIE meeting speakers	14'016.87	1'881.46		
	VIE meeting expenses	20'662.60	2'773.50		
		38'203.32	5'127.96	74'500.00	10'000.00
	Result			36'296.68	4'872.04
	Opening Accumulated surplus				22'044
	Closing Accumulated surplus				26'916.04

Budget amount requested for next year:

In view of the next planned survey to monitor performance and compare protocols to optimise effectiveness in the coming year, we would be happy if we could make a carry-over of the ECFS funds (5'000 – 10'000 Euro) for one year. In addition, we would propose to the Board to carry-over 20,000 Euros for the development of the planned CFSPID registry.