

ECFS WORKING GROUP REPORT

Year of report: 2025	
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: **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
- To monitor performance and compare protocols to optimise effectiveness, whilst reducing negative impact
- To encourage enrolment of all infants identified
- To determine the optimal management of children with CFSPID
- Improving the processing of positive NBS 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 (max 100 words):

- Implementation of NBS for CF in most countries in Europe (updated **Figure 1+2**), which is the primary aim of the NSWG.
- Monitoring performance and compare protocols to optimise effectiveness, whilst reducing negative impact ([1st survey published 2007](#), [2nd survey published 2013](#), [3rd survey 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](#))
- CF Cases Missed by NBS - Towards a Consistent Definition and Data Acquisition ([published 2023](#))
- Summary of current knowledge around NBS for CF ([book published 2020](#))

Figure 1: CF Newborn Screening programmes across Europe, updated February 2025

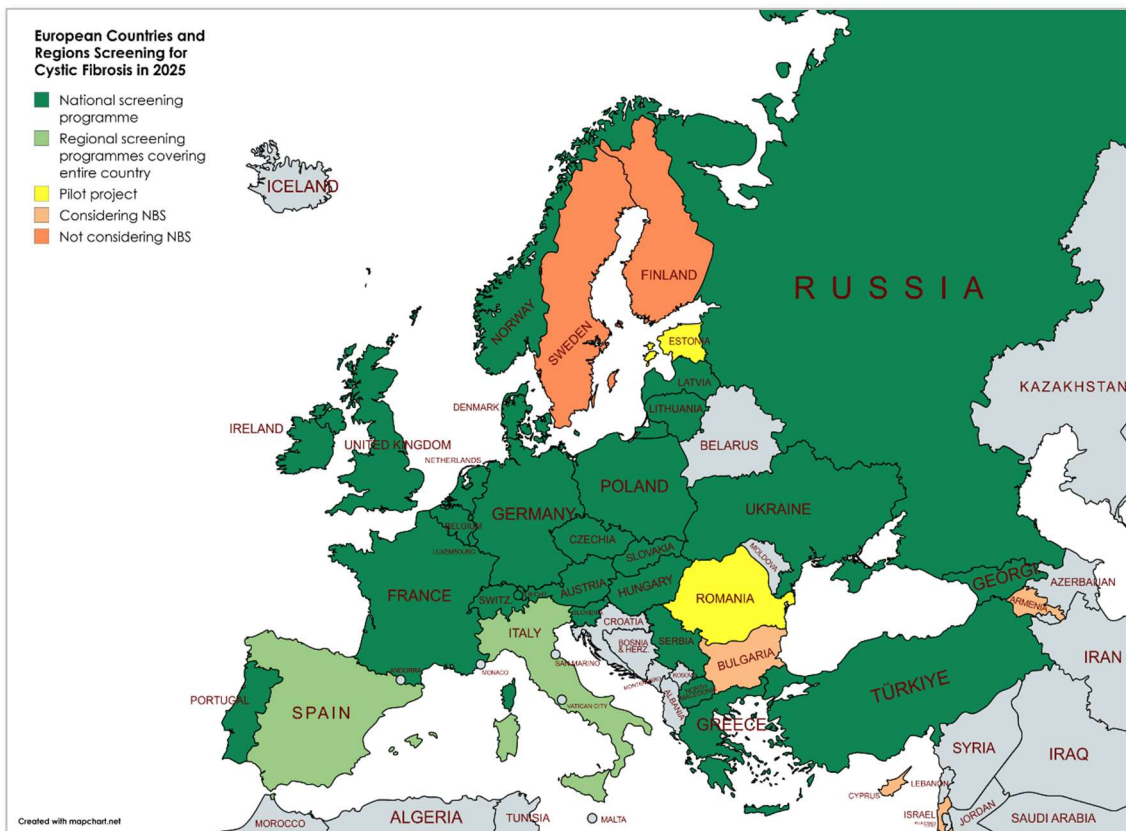


Figure 2: Screening algorithms in CF-NBS programmes, updated February 2025



Report for this year (max 1000 words)

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 55 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 nearly 200 updated email addresses; approx. 70% are ECFS members.

Since the last European CF Conference in Glasgow 2024, the core group had three video conferences online and one meeting in person at the ECFS DNWG Meeting in Brussels in February 2025. In addition, we had about 5 working meetings of the “Core Working Group” (AM, KS, CC, JB) to finalise the survey collecting *CF Cases missed by NBS*.

We also had several video conferences together with colleagues from the ECFS Patient Registry Group to define key registry parameters for the planned CFSPID-Registry. This resulted in the application for a CFPID registry group (within the ECFS registry), which is led by Andreas Jung and Kevin Southern in order to establish the CFSPID data registry within the framework of the ECFS CFPR. Specific aims of this group are:

1. To quantify the prevalence of CFSPID
2. To quantify the risk of an infant with a CFSPID designation converting to a CF diagnosis
3. To quantify the risk of an infant with a CFSPID designation developing a CFTR-related disorder
4. To identify how these infants/children are managed

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, pilot survey performed late 2024, new European survey started in February 2025](#))
- To implement a CFSPID-Registry in collaboration with the ECFSPRG ([Outcome parameter defined 2024, platform planned, financing initiated, building a new Working Group within the ECFSPRG 2024, launch for CFSPID-registry planned summer 2025](#))
- To monitor performance and compare protocols to optimise effectiveness, whilst reducing negative impact ([next European survey planned end 2025](#))
- To assess and improve knowledge of CFSPID and to provide resources to improve the evaluation and management of infants with CFSPID in Europe ([ongoing](#))

Aims for the coming year (max 50 words):

- To perform a new European survey monitoring performance and compare protocols to optimise effectiveness, whilst reducing negative impact
- To implement a CFSPID registry within the ECFSPRG
- To assess and discuss the new American CFF recommendation of introducing next generation sequencing into NBS for CF

As the current Coordinator (Jürg Barben) will have completed his permitted term in 2025, Olaf Sommerburg was elected as the new Coordinator by the Core Committee at the NSWG Meeting in Brussels on February 6, 2025. The current deputy (Kevin Southern) will step down as he will be involved as a co-coordinator for the new CFSPID Registry Group. The next NSWG deputy will be appointed at the June meeting in Milan.

Summary (max 100 words):

Last year, we focused on monitoring false-negative NBS results and started a survey in Europe (first results presented at the ECFC NSWG Meeting in Milan). We have also held several meetings with the ECFSPRG to define the parameters for a CFSPID registry and are in the process of implementing it. We also are in the preparing stage of a new European survey (4th survey) to monitor the performance and to compare protocols of European NBS programmes.

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 meeting at the ECFC in June (inviting speakers and cost for the location) and the meeting before the DNWG meeting in February. 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.

Neonatal screening - Record of Transactions		Expenses		Income	
		DKK	Euro	DKK	Euro
2024	ECFS			74.500,00	10.000,00
	Returned funds 3 Year review	120.120,00	16.123,49		
	GLA meeting speakers	23.957,60	3.215,79		
	GLA meeting expenses	14.844,90	1.992,60		
		158.922,50	21.331,88	74.500,00	10.000,00
	Result			- 84.422,50	- 11.331,88
	Opening Accumulated surplus				26.123
	Closing Accumulated surplus				14.791,12

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

In view of the next planned European survey to monitor performance and compare national protocols to optimise effectiveness end of the year 2025/26, we would be happy if we could make a carry-over of the ECFS funds (10'000 Euro) for one year.

