2026 ECFS WORKING GROUP ANNUAL REPORT

Please use this template for your annual Working Group report.
For more information, please read the following ECFS Working Group Terms of Reference document:

Group Info & Background

Name of Working Group:
ECFS-Diagnostic Network Working Group (DNWG)

Date of initial approval of Working Group:
2004 (M. Sinaasappel)

Names and emails of chair/coordinators:
Coordinator: Caroline Raynal

Vice coordinator name: Simon Gräber

Assistant name: Jasmin Berger

Long term aims of the Working Group (maximum 100 words) including estimated achievement

These are ongoing projects or objectives that may be amended or expanded if needed:
1/ To achieve pan-European cooperation on definitions of CF and clinical entities of the CF-spectrum.
2/ Standardization of existing electrophysiological techniques. Development, validation and standardization of new diagnostic technologies.
3/ Interpretation of disease liability of *CFTR*-variants and discussion of challenging cases within the group and outside
4/ Improvement/support for sweat test facilities in Europe and in LMIC
5/ Cooperation with other networks:
- ECFS-CTN and CFF TDN, standardization committee
- ECFS NSWG: neonatal screening and diagnosis are closely related
- European CF-Registry: expanding to CFSPID and CFTR-RD registries - supporting sweat test facilities in LMIC
- ECFS education committee

Outcomes already achieved (maximum 100 words):

1/ Link between ECFS-DNWG and CFF: co-authorship of manuscripts, US-speaker at annual DNWG meeting
2/ Sweat test Project:
  - Sweat test survey (Cirilli 2017)
  - Hands-on Training at ECFC: Belgrade 2018, Liverpool 2019, Vienna 2023, Glasgow 2024, Milan 2025

- ECFS Standards of care guidance for sweat testing. (Cirilli 2022; https://doi.org/10.1016/j.jcf.2022.01.004)
- e-learning tool on ECFS education platform and recording the hands-on session at ECFC 2023

3/ CFTR biomarkers: standardization and validation for NPD, ICM. Organoids and beta-adrenergic sweat test (ongoing)

4/ CFTR-Related Disorders-Project: Leader: Carlo Castellani - Publication of 4 papers in JCF (2022-2024): https://doi.org/10.1016/j.jcf.2022.09.011, https://doi.org/10.1016/j.jcf.2022.09.005, https://doi.org/10.1016/j.jcf.2024.03.008 and https://doi.org/10.1016/j.jcf.2024.03.008

Year Report

Report for this year (max 1000 words)

- short term aims and objectives for the year

1/ **Sweat test project.**
The third phase in the "Sweat test project" is to organize **hands-on sessions**, considering the latest published ECFS-recommendations. The aim is to support centres and help in problem-solving to achieve acceptable standards for good sweat testing practice as a diagnostic tool in their CF centre. This is an ongoing exercise, where we try to reach people from diverse countries who take care of sweat testing in their centre. These sessions are planned in cooperation with the ECFS-PR. This joint effort will further develop sweat testing standards in CF centres in LMIC that have already received initial support by an ECFS-PR program. After the successful sessions at previous annual ECFS conferences, we will have another sweat test hands-on session in Lisbon (3rd June 14:00-18:00).
An e-learning tool (filmed sessions in Vienna) on different sweat testing methods (conductivity, gauze/filter paper, Macroduct system) was developed for ECFS Education eLearning Programme.

2/ **New guidelines/recommendations on CFTR-related disorders (CFTR-RD)**: New stages: dissemination of our expertise
New insights in CFTR-disease liability led to the need of new recommendations on how to diagnose, treat and follow up people with a CFTR-RD. This project started in March 2020. CFTR-RD core group: Carlo Castellani, Kevin W Southern, Elke De Wachter, Nick Simmonds, Kris De Boeck, Isabelle Sermet-Gaudelus. We have published a series of four papers, covering key areas from diagnosis to management and then future directions/unmet needs. (see: publications) Last year, an e-learning tool on CFTR-related disorders was developed for the ECFS Education eLearning Programme. This year, we mainly focused on disseminating these recommendations at smaller events, at conference talks.

3/ **Beta-adrenergic sweat test**: A core group (I Sermet, T Gonska, B Tümmler, P Melotti) is working on the harmonisation of the beta-adrenergic sweat test. The goal is to develop a standardized, harmonized protocol, aiming to validate this diagnostic method and implement this at a later stage in other clinics to overcome some limitations of the sweat test. In 2024, at our annual meeting (8-10 Feb 2024), a hands-on session was organized focussing on the bubble sweat test. (see report Hannover meeting). Last year's annual DNWG-meeting (6-8 Feb 2025), included a hands-on session on the evaporimetry method, led by

Isabelle Sermet and team (Paris, FR) and Sophie Goghy and team (Brussels, BE). (see report Brussels meeting).

4/ **Widespread screening for CF in lower- and middle-income countries** (LMIC): Leaders of this project are Samia Hamouda (Tunis, Tunesia) and Anne Munck (Paris, Fr). The objective is to increase awareness of CF by policy makers in LMIC, to improve financial support for the implementation of standards of care in countries with a high CF incidence. This is an ongoing project.

5/ **Organisational structure for a 'Diagnostic FORUM'**:
The idea to have fixed meetings, with a selected group of experts from the DNWG to discuss "challenging CF diagnostic cases" was raised many years ago. Ideally, submission of a case is done some weeks prior to the meeting, resulting in a written consensus opinion report by the group of experts at the end of the meeting. ERN-Lung, supported by the European Commission, has developed the CPMS (Clinical Patient Management System) for its members (currently updated to CPMS 2.0). The aims of this secure web-based IT platform are to facilitate cross-border medical discussions and to support ERNs in the diagnosis and treatment of rare diseases. The CPMS platform contains all the ingredients we selected for an "optimal diagnostic forum". As some members of the ECFS-DNWG are also ERN-Lung members, access to CPMS is available to those. Close contact with the coordinator of ERN Lung (TOF Wagner), ERN-CF-Core network (Inez Bronsveld) and CPMS-responsible (Helge Hebestreit) have been established last year and CF-dedicated slots have been scheduled within the CPMS platform: on 28th May and 26th November 2025. We also had dedicated presentations on how to use the tool CPMS platform by Helge Hebestreit (DNWG meeting in Milan, 2025) and by Inez Bronsveld and Elke De Wachter (DNWG Annual meeting in Prague, 2026)
Involving coordinators or active members of other ECFS-working groups in the CF-case discussions on CPMS could be an opportunity to increase collaboration between ECFS and ERN, selecting specific experts in the field, with the aim to improve diagnosis and care for pwCF. We have invited experts to apply to join the forum in Milan (DNWG meeting, 2025).
The eventual aim is to make this available for the wider ECFS community.

- current number of members

142 members
All 72 participants of our annual DNWG-meeting (Brussels) have obtained or renewed their ECFS-membership. However, despite repeated reminders, not all members (those who did not attend the annual meeting) have responded. We should discuss with the board how to solve this issue and if DNWG members without ECFS membership should be withdrawn after repeated reminders.

- measures taken to encourage ECFS membership

- An ECFS-DNWG logo was developed in 2021 to increase visibility AND to emphasise the link with the ECFS
- During each official meeting (symposia and annual meetings) this issue is stressed. "Being part of the European CF Society is mandatory to become/remain a member of the DNWG".
- Before each announcement of a meeting e-mails are sent to all DNWG members to remind them that ECFS-membership is mandatory to remain part of the working group.
- Only CONFIRMED ECFS members received funding for accommodation during our annual DNWG-meeting in February for the past three years. This resulted in an important increase in ECFS-membership within the working group.

- outcomes/achievements (e.g. meetings, activities, website development, awards, publications etc).

1/ Meetings (between May 2025-May 2026)
- Sweat test hands-on session: 4th June 2025 - Milan, IT: 16 participants
- DNWG session (open meeting) at the 48th ECFS Conference (Milan): 6th June 2025 (Fri 12:30-14:30)
- 5th DNWG-brainstorm meeting at NACFC (Seattle): 24th September 2025. 16 participants
- 23rd Annual DNWG-meeting (Prague): 12th- 14th February 2026.
Programs and reports of the meetings are posted on the ECFS website and attached

2/ Website development
News regarding DNWG activities are posted on the website.

3/ Publications from the DNWG as a group:

CFTR-RD papers (4)

- Castellani C, De Boeck K, De Wachter E, Sermet-Gaudelus I, Simmonds NJ, Southern KW; ECFS Diagnostic Network Working Group. ECFS standards of care on CFTR-related disorders: Updated diagnostic criteria. J Cyst Fibros. 2022 Nov;21(6):908-921. doi: 10.1016/j.jcf.2022.09.011. Epub 2022 Oct 8. PMID: 36220763

- Sermet-Gaudelus I, Girodon E, Vermeulen F, Solomon GM, Melotti P, Graeber SY, Bronsveld I, Rowe SM, Wilschanski M, Tümmler B, Cutting GR, Gonska T. ECFS standards of care on CFTR-related disorders: Diagnostic criteria of CFTR dysfunction. J Cyst Fibros. 2022 Nov;21(6):922-936. doi:10.1016/j.jcf.2022.09.005. Epub 2022 Oct 5. PMID: 36207272.

- Simmonds NJ, Southern KW, De Wachter E, De Boeck K, Bodewes F, Mainz JG, Middleton PG, Schwarz C, Vloeberghs V, Wilschanski M, Bourrat E, Chalmers JD, Ooi CY, Debray D, Downey DG, Eschenhagen P, Girodon E, Hickman G, Koitschev A, Nazareth D, Nick JA, Peckham D, VanDevanter D, Raynal C, Scheers I, Waller MD, Sermet-Gaudelus I, Castellani C; ECFS Diagnostic Network Working Group.. et al ECFS standards of care on CFTR-related disorders: Identification and care of the disorders. J Cyst Fibros. 2024 Mar 19:S1569-1993(24)00037-7. doi: 10.1016/j.jcf.2024.03.008. Epub ahead of print. PMID: 38508949.

- De Wachter E, De Boeck K, Sermet-Gaudelus I, Simmonds NJ, Munck A, Naehrlich L, Barben J, Boyd C, Veen SJ, Carr SB, Fajac I, Farrell PM, Girodon E, Gonska T, Grody WW, Jain M, Jung A, Kerem E, Raraigh KS, van Koningsbruggen-Rietschel S, Waller MD, Southern KW, Castellani C; ECFS Diagnostic Network Working Group.l. ECFS standards of care on CFTR-related disorders: Towards a comprehensive program for affected individuals. J Cyst Fibros. 2024 Feb 21:S1569-1993(24)00011-0. doi:10.1016/j.jcf.2024.01.012. Epub ahead of print. PMID: 38388234.

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

1/ **Planned meetings (2026-2027):**

- Sweat test hands-on session (3rd June 2026)
- DNWG meeting (5th June 2026)
- DNWG-brainstorming meeting - NACFC 2026
- Annual ECFS-DNWG meeting …Feb 2027 - *dates and venue TBC*

2/ Continue the work on CFTR-RD project – particularly around dissemination, registries and the other points discussed in Paper 4 (De Wachter JCF 2024)

3/ Sweat test project:

- Sweat test hands-on sessions.
- Repeat the sweat test survey after the efforts to improve sweat test facilities in Europe.
- Multicentre project aiming to establish reference values for sweat chloride according to age (category)

4/ Get more actively involved in ERN-CPMS

5/ Investing in beta-adrenergic sweat test as an additional, accessible diagnostic tool (CFTR-functional test)

6/ Update of the guidelines for molecular genetic diagnosis of cystic fibrosis and CFTR-RD (last update in 2009)

Summary (maximum 100 words):

DNWG is an active group of enthusiastic CF specialists with special interest in diagnostic issues. It consists of CF physicians, geneticists, clinical biochemists, basic scientists, and other MDT members, making a very complementary group. This enables us to have diverse and strong expertise. We have made significant progress but there are still many important scientific and clinical issues to be resolved, which the DNWG has the capability and expertise to achieve. These include: the interpretation of *CFTR* variants and their disease liability; the CF spectrum and the interpretation of biomarkers for CFTR activity; and CFTR activity and its translation to the individual patient.

Finances

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

Annual DNWG meeting in Prague:
72 attendees
Sponsorship for ECFS-DNWG:
Elitech, Dvysr, 3GENES, ITA Interact, Filière Muco-CFTR (FR), Vertex Pharmaceuticals and ECFS

Expenses: 22 902,18 euro (hotel 2 nights, meeting rooms, YI-award (one still not reimbursed), catering, dinner, social event, travel cost invited speaker)

Total balance of the Annual meeting: balanced budget

Attached a detailed overview of the DNWG-budget (excel file)

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

We would like to receive **the annual 10.000 euro** to support the annual DNWG-meeting 2027, YI-award and travel costs for an invited speaker and to set-up future hands-on sweat test sessions (covering partly the rent of the room)

Remark: The number of participants of the annual meeting has importantly increased over the years. Life is getting more expensive (increased costs for hotel stay, meals). Despite all efforts to find sufficient funding from different domains (industry, university, patient's associations) we certainly won't be able to cover the expenses of housing, catering, restaurant etc for all participants of our upcoming annual February meetings. In that situation, we thought we might ask a registration fee, covering most of the expenses for next year's annual DNWG meeting.

For the start-up and running costs of the new project (Nest steps of the sweat test project on establishing reference values for sweat chloride according to age) an additional budget (hand-over) is more than welcome.

The deadline for submission to the ECFS Office is 1st May.

The report will then be presented to the board prior to the summer ECFS board meeting and then a decision will be made at the next board meeting for approval before being distributed.