2025

European
Reference
Network
for rare or low prevalence
complex diseases

Network
Connective Tissue
and Musculoskeletal
Diseases (ERN ReCONNET)

ERN ReCONNET Plenary and Board of Network On-Line Meeting 2024-2025

JANUARY 20 - 27, FEBRUARY 3-10, 2025

Day 1 _ ERN ReCONNET Plenary and Board of Network On-Line Meeting 2024-2025 Agenda

Date of the meeting: 20th January 2025 (15:00 – 17:00 CET)

15:00 – 15:10	Overview and goals of the meeting	M. Mosca M. Cutolo
15:10 – 15:20	ERN ReCONNET – Road to 2027	M. Mosca
15:20 – 15:30	Evaluation and other updates from the Coordination Team	D. Marinello
15:30 – 15:50	The Clinical Patient Management System	M. Mosca V. Smith
15:50 – 16:15	Education and Training	M. Cutolo W. Lautre (on behalf of E. Hachulla) T. Avcin
16:15 – 16:30	Research	R. Talarico G. Simonini
16:30 – 17:00	Discussion and closure of the meeting	M. Mosca M. Cutolo

Day 1- 20th January 2025

Overview and Goals of the Meeting

***Marta Mosca** – Network Coordinator ERN ReCONNET – Azienda Ospedaliero Universitaria Pisana, Italy.*
***Maurizio Cutolo** – Steering Committee – IRCCS AOU San Martino, Italy.*

The Network Coordinator M. Mosca and M. Cutolo welcomed participants and introduced the objectives of the 2024–2025 Plenary Meeting. The programme was designed to review the Network's achievements over the past year, present priorities for 2025, and foster discussion among healthcare professionals, ePAGs and network partners.

ERN ReCONNET – Road to 2027

***Marta Mosca** – Network Coordinator ERN ReCONNET – Azienda Ospedaliero Universitaria Pisana, Italy.*

Marta Mosca presented ERN ReCONNET's strategic roadmap towards 2027, highlighting the Network's evolution since 2017 and its vision for the coming years. Key priorities include strengthening governance, developing the TogethERN ReCONNET Registry, expanding education and research activities, enhancing cross-border collaboration through CPMS 2.0, improving quality of care, increasing patient involvement, and ensuring the long-term sustainability of ERNs within national healthcare systems.
The main areas of development that will be addressed in the years 2023-2027 are:

- **Governance:**
  - New ERN ReCONNET governance statutes
  - Identification of the National Hubs
  - Election of new disease coordinators
  - New member of the Coordination Team
- **Registries:**
  - Draft 10 disease-specific variables (TogethERN ReCONNET Level 3)
  - Draft general assessment variables (TogethERN ReCONNET Level 2)
  - Initial drafting of the legal documents
  - Manuscript mapping of coding systems
- **Education and training:**
  - Webinars
  - Design online accredited course (pilot project in 2025, accredited course on transition in 2026).
  - Planning of the 2025 ERN ReCONNET scientific congress (Prague)
  - ERN ReCONNET Exchange Program
- **CPMS:**
  - Participation in the development of the new CPMS
  - Preparatory tasks for the transition to the new CPMS
- **Promotion of early diagnosis and research:**
  - Red Flags for IgG4
  - Red flags development in other diseases (e.g. IIM, APS)
  - UCTD project
  - Definition of the project on Points to Consider on imaging in IgG4-related disease

  - ERN ReCONNET Good Practice Sharing Initiative 2024
- **Clinical Practice Guidelines and Clinical Decision-making tools:**
  - Red Flags IgG4 **paper** (doi: 10.1016/S2665-9913(24)00192-9)
  - Red Flags IIM development
  - RarERN Path – narrative medicine SLE
  - RarERN Path SSc **paper** (doi: 10.1177/23971983241269109)
  - ReCONNET, SLICC, SLEURO International Task Force on Rare Lupus manifestations
  - RP-QoL development
- **Transition**
  - Transition of care survey for healthcare professionals, draft survey for patients in collaboration with other ERNs
  - Paper on the results of the Transition of care survey for healthcare professionals (doi: 10.1093/rap/rkae149)
- **Quality Improvement:**
  - ERN Annual Monitoring data collection
  - Draft new HCP Performance System
- **Dissemination:**
  - Development of the new website and constant updates on the current website
  - New leaflet ERN ReCONNET
  - ERN ReCONNET @EULAR and @ACR
- **Collaboration with other entities and initiatives:**
  - Draft of the Memorandum of understanding with RITA
  - Collaboration with RITA on the transition survey
  - Publication of the results of the ERN transversal group survey on pregnancy in rare diseases.

Evaluation and other updates from the Coordination Team

***Diana Marinello** – Network Manager ERN ReCONNET – Azienda Ospedaliero Universitaria Pisana, Italy.*

Diana Marinello shared the results of the European Commission's five-year evaluation, in which ERN ReCONNET achieved full compliance with all network criteria, ranking among the highest-performing ERNs. Regarding the evaluation of the Full Members of ERN ReCONNET, 26 HCPs were evaluated, and six of them were audited (on-site evaluation). 25 out of 26 HCPs evaluated were rated as satisfactory, and only one HCP needed improvement.
A summary of the ERN continuous monitoring and annual performance indicators data collected in 2023 was also presented:

- 14,554 new confirmed cases of rCTDs patients seen by ERN ReCONNET
- 48% of HCPs reporting the use of ORPHACODEs in their centre
- 23 new training activities organised by ERN ReCONNET (353)
- 8920 new visitors and 31617 returning visitors to the ERN ReCONNET website
- 14 events where ERN ReCONNET was disseminated
- 89% of HCPs entered data directly into the SANTE data collection platform - 92% ERN survey

Finally, the new ERN ReCONNET Quality Improvement System is useful to collect the evaluation related activities: ERN continuous monitoring, HCP performance system, ERN and HCPs five-year evaluation, and internal evaluation.

The new HCP performance system is required as mandatory by the EC and should be reported directly by each HCP in the Annual Monitoring and in the ERN Grant. The new tool aims to promote a continuous improvement system within ERN ReCONNET and as an opportunity for each centre to improve specific areas within ERN ReCONNET (e.g., contribution to the CPMS). The Coordination Team will support each HCP in this process. The tool would be designed in collaboration with the Steering Committee to make it more sustainable.

The Clinical Patient Management System

***Vanessa Smith** – WP4 Coordinator – Ghent University Hospital, The Netherlands.*
***Marta Mosca** – Network Coordinator ERN ReCONNET – Azienda Ospedaliero Universitaria Pisana, Italy.*

M. Mosca introduced Prof. Vanessa Smith as the beneficiary for the CPMS within the Grant 2023-2027. M. Mosca announced that a way to reimburse the activities of the discussion on the CPMS is being developed in order to recognise the time devoted to discussing cases within the new tool. M. Mosca emphasised that the CPMS is pivotal in achieving the fundamental ERN principle of "Share, Care and Cure".

Then, V. Smith updated the whole Network to the new CPMS 2.0. About CPMS 1.0., 47 panels and around 150 active users for ERN ReCONNET have been registered. The implementation of the new CPMS 2.0 has been meticulously planned, with 18 meetings of the business implementation group (BIG) held in 2024. V. Smith then explained the functionality of the new CPMS 2.0, showing how to access it via a simple Google search (https://cpms.ern-net.eu) and logging in using an EU login with two-step authentication. Videos on how to create an EU login directly from the CPMS 2.0 site, how to sign up to CPMS 2.0, a webinar for clinicians on how to use the system, and a brief presentation of the CPMS 2.0. are also available. Unlike the first CPMS version, the CPMS 2.0. does not use an SMS code for login; it has an EU login app for iPhone, which is available to download from the CPMS 2.0 site. The new system has been updated to allow the addition of a subject area with a subspecialty, such as interstitial lung disease, and to automatically search for experts to provide advice, such as those from ERN Lung. The new CPMS also facilitates meetings and video calls and organises recurring meetings in a GDPR-proof way. CPMS 2.0's key feature is the option to appoint an assistant within the healthcare provider to assist with uploads, panel management and consent taking. The oldest system will be completely closed by 31 January 2025.

V. Smith explained that the EC is pushing for the use of the new CPMS as it is the only system Ukraine can use. The EC strongly advises against contacting Ukrainian experts using the usual communication methods used within the network. Instead, the email address of the Ukraine Central Hub must be used (hub_ukraine@ohmatdyt.com.ua). This is because Ukraine is at war and does not keep informal patient consent forms. Only one hospital keeps them. Clinical cases must not be discussed without positive confirmation from the hub.

Education and Training

***Maurizio Cutolo** – Co-Chair WG Education & Training– IRCCS AOU San Martino, Italy.*
***Wiebke Lautré** – Project manager – CHU Bordeaux, France (on behalf of Eric Hachulla – Co-Chair WG Education & Training – CHU Lille, France).*
***Tadej Avcin** – Transition of Care Task Force – University Children's Hospital, UMC Ljubljana, Slovenia.*

Maurizio Cutolo reviewed the success of the 2024 webinar programme – n. 18 webinars were organised by ERN ReCONNET in 2024, covering almost all Disease Groups were covered, with the exception of Sjögren's and IgG4. Prof Cutolo concluded his presentation by presenting the webinar agenda for 2025. The number of proposals collected since the first call in July 2024 was 21 from all disease groups (last call on 9 January 2025). Patient-specific educational webinars decreased from 32% in 2024 to 14% in 2025, and clinician-specific webinars decreased from 41% in 2024 to 29% in 2025. Interestingly, patient and clinician educational webinars increased from 27% in 2024 to 57% in 2025. Wiebke Lautré updated participants on the ERN ReCONNET Exchange Programme, which was launched in two phases:

1. The call for host offers, which received 14 from seven different countries.
2. The call for participants, which received 45 applications.

Due to financial limitations, not all applicants were able to participate in 2025. However, an agreement was reached to send half in 2025 and the other half for the academic year 2025-26.

Regarding the accredited online training course, it will be organised into at least 10 learning modules (one for each disease group) and all modules should focus on the rare in rare, i.e. very rare diseases (RP, IgG4, EDS, MCTD and UCTD) would be presented in their entirety, while diseases that are slightly less rare (SLE, SS, SSc and APS) would present certain aspects of their disease that may be particularly rare or that may not be covered in other courses. This would avoid overlap with other courses, in particular the EULAR course.

Tadej Avcin presented progress on the ERN ReCONNET Transition of Care accredited training programme. A date has been set for a Task Force meeting in Ljubljana in November 2025 to define the structure and content of the ERN ReCONNET training programmes. The draft framework and list of competencies will be discussed at this meeting, which will include virtual and face-to-face participants. Standards for ERN ReCONNET transition of care training are expected to be defined by the end of the year. It would be planned to organise an ERN ReCONNET course based on these standards in the future.

T. Avcin concluded by highlighting the existing inter-ERN collaboration within this project. There are some overlapping diseases between ERN RITA and ReCONNET, such as childhood-onset inflammatory connective tissue diseases, so it has been agreed with ERN RITA to collaborate in this area. ReCONNET will focus on connective tissue disorders while RITA will focus on Juvenile Idiopathic Arthritis (JIA) and other paediatric rheumatic diseases.

Research

Rosaria Talarico *– Scientific Coordinator ERN ReCONNET – Co-Chair WG Research & Quality of Care – Azienda Ospedaliero Universitaria Pisana, Italy.*
Ilaria Galetti *– Co-Chair WG Research & Quality of Care – Federation of European Scleroderma Associations (FESCA), GILS, Italy.*
Gabriele Simonini *– Transition of Care Task Force – Meyer Children's Hospital IRCCS, Firenze, Italy.*

S. Talarico, also on behalf of I. Galetti, both Chairs of the Research and Quality of Care Working Group, gave a brief summary of all the research activities carried out in the Network in recent months under the supervision of the working group:

1. Creation of clinical practice guidelines.
2. Patient's care pathways and organisation of care (RarERN Path methodology™)
3. ERN ReCONNET Supplement "Rare inside Rare" (14 papers)
4. VACCINATE Study, COVID-related initiatives
5. Economic burden of disease (Scuola Superiore Sant' Anna)
6. Study on cross-border procedures in ERN centres
7. Transition of Care
8. ERN ReCONNET Good Practice Sharing Initiative
9. Development of Red Flags, working on the early diagnosis of rare disease

Regarding the Red Flags activity, the paper published in Lancet Rheumatology on the identification of red flags for IgG4 was the first dissemination on this topic (doi: [10.1016/S2665-9913(24)00192-9](10.1016/S2665-9913(24)00192-9)). The Red Flags project is also ongoing in the IIM Disease Group. It has been discussed in the APS and MCTD Disease Groups, while a slightly different approach to Red Flags has been proposed in the SSc Disease Group, not for early diagnosis but for early identification of different organ involvement.

Another important publication achieved was represented by the patient care pathways model from the organisational point of view for systemic sclerosis (doi: [10.1177/23971983241269109](10.1177/23971983241269109)), in which the group of the Scuola Superiore Sant'Anna mapped the implementation of the survey for the health care providers.

S. Talarico also mentioned other important ongoing projects:

1. the development of a new tool to measure quality of life in relapsing polychondritis.
2. new points to consider for imaging in IgG4-RD
3. the development of diagnostic criteria in UCTD
4. the proposal of autoantibodies in juvenile SLE and follow-up in the adult phase.

She concluded by inviting all participants to the 2nd International Congress in Prague (9-11 April 2025) as a great opportunity to share many research activities related to connective tissue diseases (https://www.ern-reconnetcongress.com/).

The floor was then given to G. Simonini. He presented the research aspects of the Transitional Care Project.

G. Simonini introduced the Task Force as a multidisciplinary team composed of physicians, patient representatives and other key figures. He explained that the multidisciplinarity of the Task Force is essential to address the high number of unmet needs.

He explained that the transition process is divided into three, and eventually four, concurrent and sequential phases. The first phase involved mapping existing transition care processes through a survey. In the second phase, the best practice guideline would be developed. In the final phase, a data set for the registry would be defined to monitor the transition process as a whole. Another survey is being developed focused on patients and caregivers' needs and expectations. Thanks to this process, G. Simonini explained that four milestones should be achieved:

1) Mapping and harmonisation of terminology adopted in registries on the transition of paediatric patients to adulthood
2) Development of registry data sets for rCTDs paediatric patients
3) Development of informed consent and other documents dedicated to paediatric patients
4) Design of enrollment strategies targeted at patients, parents and paediatric centres

Discussion and closure of the meeting

The meeting concluded with an open discussion on educational resources, dissemination strategies and future collaborative activities. Participants reaffirmed the importance of sharing expertise across the Network and continuing to build on ERN ReCONNET's achievements in research, education and patient-centred care.

Day 2 _ ERN ReCONNET Plenary and Board of Network On-Line Meeting 2024-2025 Agenda

Date of the meeting: 27th January 2025 (14:00 – 16:00 CET)

14:00 – 14:05	Introduction to the meeting	M. Mosca M. Cutolo
14:05 – 14:20	Yearly update from the ePAG Advocates	S. Aguilera J. Andersen
14:20 – 14:35	The new Governance Statutes of ERN ReCONNET	M. Mosca D. Marinello
14:35 – 15:05	Registries	R. Talarico M. Schneider J. Fonseca G. Simonini
15:05 – 15:20	ERN ReCONNET Young	V. Smith S. Aguilera
15:20 – 15:35	Patient Partnership	J. Andersen G. Burmester
15:35 – 16:00	Discussion and closure of the meeting	M. Mosca M. Cutolo

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Day 2 – 27th January 2025

Introduction to the meeting

M. Mosca opened the second day of the Plenary Meeting by summarising the first day of the meeting, which had been very fruitful. She then described the agenda with the continuation of the presentations of the transversal activities of the working groups and then she introduced the first topic, the ePAG Advocates.

Yearly update from the ePAG Advocates

***Jeanette Andersen** – ePAG Advocate – Lupus Europe, Denmark.*
***Silvia Aguilera** – ePAG Advocate – Asociación Española Síndrome Antifosfolipídico, Spain.*

Jeanette Andersen and Silvia Aguilera (co-chairs of the ERN ReCONNET ePAG) presented the renewed structure of the ePAG Advocates, introducing two ePAG Co-Chairs, Disease Coordinators and Supporting Patient Partners to strengthen patient representation across the Network.

J. Andersen clarified that the scope has not changed in the new organisation, but what is new is that some new patient representatives will be added to the Working Groups supporting the ePAGs. The ePAGs do not have specific projects, but work on the different projects going on in the Network (Working Groups and Disease Group projects, ERN ReCONNET Steering Committee and ePAG Steering Committee meetings), giving their important point of view on different aspects. They highlighted the essential contribution of ePAGs to Working Groups, Disease Groups and strategic projects, while emphasising the need for closer collaboration with clinicians and greater recognition of patients as equal partners in research, education and decision-making.

The new Governance Statutes of ERN ReCONNET

***Marta Mosca** – Network Coordinator ERN ReCONNET – Azienda Ospedaliero Universitaria Pisana, Italy.*
***Diana Marinello** – Network Manager ERN ReCONNET – Azienda Ospedaliero Universitaria Pisana, Italy.*

Marta Mosca and Diana Marinello presented the new Governance Statutes, designed to institutionalise the Network's organisational structure and align it with the evolution of the ERN system as a whole.

Major developments include the establishment of a new Strategic Board, the introduction of National Hubs to strengthen integration with national healthcare systems, updated membership and endorsement procedures, a new Quality Improvement and HCP Performance System, clearer conflict-of-interest policies and enhanced collaboration with Supporting Partners.

The revised governance aims to improve transparency, engagement and long-term sustainability of ERN ReCONNET.

Registries

***Rosaria Talarico** – Scientific Coordinator ERN ReCONNET – Azienda Ospedaliero Universitaria Pisana, Italy.*
***Matthias Schneider** – Co-Chair Registries and eHealth WG – Universitätsklinikum, Düsseldorf, Germany.*
***João Eurico Cabral da Fonseca** – Co-Chair Registries and eHealth WG – Centro Hospitalar de Lisboa Norte, Portugal.*
***Gabriele Simonini** – Transition of Care Task Force – Meyer Children's Hospital IRCCS, Firenze, Italy.*
***Gerieke Been** – Project Manager Rare Disease Databases and Omics Analysis – UMCG, Genomics Coordination Centre, MOLGENIS*

J.E. Fonseca outlined the objectives of the Registries and eHealth Working Group. Firstly, to improve CPMS virtual consultations and raise awareness within the ERN ReCONNET. Second, to support the TogethERN ReCONNET registry infrastructure and support the improvement of the CPMS. Another important task is the biobank survey.

Subsequently, the floor was given to M. Schneider, who spoke about coding systems and the importance of harmonising the way patients are documented in local systems. A survey was distributed among ERN ReCONNET HCPs to map the existing coding systems in use, yielding 45 responses. It was reported that only one centre used paper-based documentation for patient records. He concluded by stating that Coding Systems are possible to be changed by the Hospitals with the aim of standardising the system. A paper will be prepared in order to disseminate the results of the work.

S. Talarico provided an update on the development of the TogethERN Registry. She summarised the levels of the variables that will be included in TogethERN Registry:

- 1st level: common data set and mandatory to all the 24 ERN Registries. Maximum of 16 variables.
- 2nd level: general variables common to all the diseases of ERN ReCONNET. Maximum of 50 variables.
- 3rd level: disease-specific variables. Maximum of 50 variables.
- 4th level: for research activity (to be planned in the future).

She explained that the first level of common set of variables is the only mandatory level for all diseases in all ERNs (epidemiological data requested by the EC). The second level (general assessment variable) is cross-sectional for all ERN ReCONNET diseases and will be proposed to all disease groups after a DELPHI process to agree on the defined variables. With regard to disease-specific variables (Level 3), she explained that the situation varies among the different diseases of ERN ReCONNET. In fact, some diseases already have existing core sets (e.g. SLE), whereas other Disease Groups need to design a new core set (e.g. UCTD) or adapt an existing one (e.g. IIM).

In order to help all Disease Groups to clarify any possible doubts or needs regarding the variables, S. Talarico invited Disease Coordinators and ePAGs to attend the meeting scheduled for 21 February 2025 with Molgenis, explaining that 20 March 2025 has been set as the deadline for receiving variables from each Disease Group intending to provide a disease-specific core set of variables (third level) within the common ERN ReCONNET registry.

G. Simonini importantly gave an update on paediatric variables for the TogethERN ReCONNET Registry. In fact, a specific registry for paediatric patients has been created in parallel to the adult one. Four specific objectives have been developed to integrate the paediatric registry with its adult counterpart:

1. mapping and harmonisation of terminology used in registries on the transition of paediatric patients to adulthood (2024-2025).
2. development of registry data sets for paediatric rCTD patients (deadline 2026).
3. development of informed consent and other documents specific to paediatric patients (deadline 2026).
4. development of enrolment strategies targeted at patients, parents and paediatric centres (2024-2026).

Finally, the floor was given to G. Been, project manager at Molgenis, the IT company that has dealt with several registries of other ERNs (CRANIO, Genturis, ITHACA and SKIN) and also of TogethERN ReCONNET Registry. She gave an update on the registry's platform: what has been done, what is ongoing and what is to come.

The registry is based on Molgenis, a data platform built on FAIR principles to enable local data collection. Molgenis is completely open source and can be installed in any centre to collect data (http://molgenis.org). The platform allows data to be searched at European level and also linked to different European resources (currently only at metadata level).

Concerning work on TogethERN Registry already completed by Molgenis, G. Been explained that a pilot data entry tool was created, which allowed dummy data to be entered for certain patients (a workshop was held on this topic). Regarding ongoing activities, data elements are being created for specific disease groups. These data will then be examined to see how they can be more easily incorporated into Molgenis, i.e. by direct upload or by manually filling in forms. Finally, for the next activities there are the steps with missing actions: landing page; public dashboards; documents (central and local documents).

Importantly, she listed all the steps needed towards practical implementation of the registry:

1. Data processing agreement (to be signed by the UMCG and the registry's Coordinator)
2. Data sharing agreement (to be signed by all HCPs)
3. Informed consent for the patient (need to be drafted per country)
4. Ethical approval
5. Defining the Data Access Committee (DAC)
6. Finalize data elements to be collected (per disease group)

She explained that the ERN ReCONNET TogethERN Registry has already been included in the European Registry Directory (ERDRI.dor) and the Central Metadata Repository (ERDRI.mdr) based on the common data elements. Once the disease-specific elements are defined, the TogethERN Registry will be included in the other EU ERDRI levels. About Spider (https://eu-rd-platform.jrc.ec.europa.eu/spider/), it is the pseudonymisation tool that is used to create a pseudonym for the patients' sensitive data, i.e. name and date of birth, in the registry. The pseudonym is integrated into the registry and linked to the patient. There are still many concerns from the ERN community about Spider and in particular the terms of use and contracts behind it.

ERN ReCONNET Young

Silvia Aguilera *– Co-Chair ERN ReCONNET Young WG – Asociación Española Síndrome Antifosfolipídico, Spain*

Vanessa Smith *– Co-Chair ERN ReCONNET Young WG – Ghent University Hospital, The Netherlands*

The floor was then given to V. Smith and S. Aguilera, co-chairs of the ERN ReCONNET Young Working Group. V. Smith explained that the ERN ReCONNET Young Working Group allows two young clinicians from each Disease Group to join and contribute to the Network. The ERN ReCONNET Young Working Group is made up of young clinicians represented for each Disease Group within ERN ReCONNET, and they should bring information within the disease groups.

Over the past year, young clinicians have contributed to the promotion and testing of the new CPMS 2.0. They have also collaborated on several publications within the ERN ReCONNET (doi: 10.55563/clinexprheumatol/jpargp; doi: 10.1177/23971983241269109) and are involved in Working Groups and Disease Groups. V. Smith reported on some examples of active participation of young clinicians within the network: website update; Red Flags within the IIM Disease Group; management of digital ulcers in SSc; a systematic review of PJP prophylaxis across the ten diseases covered by ERN ReCONNET; collaboration on TogethERN ReCONNET Registry variables; coding systems and monitoring practices across ERNs.

M. Mosca commented that ERN is a place where young patients and clinicians can meet and work together. She invited young patients to the Working Group and asked participants to report activities to younger colleagues and ePAGs. She then encouraged her colleagues to invite young members of their clinics to join the ERN ReCONNET Young Working Group by collaborating and contributing.

ERN ReCONNET Patient Partnership Working Group

Jeanette Andersen *– Ad interim Co-Chair Patient Partnership WG– Lupus Europe, Denmark.*

Gerd Burmester *– Co-chair Patient Partnership WG – Charité Universitätsmedizin, Berlin, Germany.*

J. Andersen (Co-Chair of the Patient Partnership WG) started her talk by presenting the concept of Patient Partnership shared by the different ERNs.

The PP framework was published last year with a common direction across different ERNs. Of note, ERN ReCONNET has developed a specific Patient Partnership Strategy. To implement PP, the first step is to better understand how the patient-physician partnership works and how it can be improved. She reported on some suggestions such as improving common terminology and making the general rare disease community aware of how ERN ReCONNET works together. She reported on the idea of creating a survey (based on an ERN EYE survey) for both the Disease Groups and the Working Groups (both HCPs and ePAGS) to analyse what can be improved. Then she highlighted the importance of integrating patients into specific activities as mentioned in the ERN ReCONNET strategy document for patient involvement. Then she went through the proposed projects for 2025 as possible areas of collaborations:

- AI (patients using chatGPT and other sources to find medical information).
- Fatigue, as transversal for all disease groups
- Measuring the impact of patient (educated expert) input into clinical trials
- Diversity, equity and inclusion
- Lupus100 questions (most frequently asked questions from patients answered by physicians and patients), with the will to propose for other diseases.
- LupusGPT, as an example of how a chatbot can be built
- Consultation cards, a tool created to prepare patients and clinicians for the visit

J. Andersen reported that the Patient Partnership Working Group also had suggestions for the development of educational materials for patients and caregivers, which should be developed with three different levels of education. The Working Group also decided to develop webinars and short videos (three-to-five-minute videos) about the diseases and other cross-cutting topics such as CAR-T cell treatments and drugs, as well as topics such as psychological impact, fatigue, transition of care, AI and ChatGPT, all explained in lay language.

She gave the floor to G. Burmester, co-chair of the Patient Partnership Working Group. He focused mainly on the emerging artificial intelligence tools, stating that this is a huge development moment for this type of IT tools (e.g. ChatGPT, Gemini, CoPilot, Lupus GPT, Lupus 100, etc.) and that health professionals can only cope with it by working together with patients. He added that these tools would enable clinicians to improve the quality of online information provided to patients, and that people who are just starting to experience the first signs of some disease and that have not yet been exposed to the reality of ERNs could be made aware of the network more quickly thanks to the correct use of these tools. Finally, he said that AI can help keep patients informed about the latest developments in therapeutic areas without language barriers.

Discussion and closure of the meeting

The final discussion focused on strengthening collaboration between clinicians and ePAG Advocates and on the implementation of the TogethERN Registry. Participants discussed harmonising registry data with existing disease-specific registries, ensuring interoperability rather than duplication, and using the registry to improve early diagnosis, monitor patient pathways and generate high-quality evidence for rare connective tissue and musculoskeletal diseases. The session concluded by reaffirming the importance of patient partnership, data harmonisation and collaborative research as key priorities for ERN ReCONNET.

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Day 3 _ ReCONNET Plenary and Board of Network On-Line Meeting 2024-2025 Agenda

Date of the meeting: 03rd February 2025 (14:00 – 16:30 CET)
Avenue: On-Line

14:00 – 14:05	Introduction to the meeting	M. Mosca M. Cutolo
14:00 – 14:30	APS Disease Group	S. Aguilera S.Sciascia *15 min presentation + 15 min discussion*
14:30 – 15:00	EDS Disease Group	M. Castori *15 min presentation + 15 min discussion*
15:00 – 15:30	IIM Disease Group	A. Meyer *15 min presentation + 15 min discussion*
15:30 – 16:00	IgG4 Disease Group	E. Della Torre *15 min presentation + 15 min discussion*
16:00 – 16:30	MCTD Disease Group	M. Ciupera *15 min presentation + 15 min discussion*
16:30	Closure of the meeting	M. Mosca M. Cutolo

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Day 3 – 03rd February 2025

Introduction to the meeting

M. Mosca began with a short introduction. The day would be dedicated to the Disease Groups of the ERN ReCONNET Network. Each group would have the opportunity to share all the activities carried out, the specific knowledge acquired through collaboration, and future projects. She ended the introduction by congratulating on the high number of participants, which means the great participation in ERN ReCONNET activities throughout the year.

APS Disease Group

***Silvia Aguilera** – ePAG Advocate of the Antiphospholipid Syndrome (APS) Disease Group - Asociación Española Síndrome Antifosfolipídico, Spain.*

***Savino Sciascia** – Disease Coordinator of the Antiphospholipid Syndrome (APS) Disease Group*

The APS Disease Group presented a broad portfolio of educational and research initiatives aimed at improving both clinical practice and patient information.

Key achievements included:

- the development of the APS 100 Questions project, inspired by the successful Lupus100 initiative,
- the translation of EULAR lay recommendations and educational webinars into multiple languages,
- the creation of a dedicated APS website to provide reliable information for patients and healthcare professionals.

On the research side, significant progress was reported on: the Red Flags for APS project, identifying early clinical indicators to facilitate diagnosis and on the development of ERN recommendations for the management of rare APS manifestations in collaboration with international partners, including ERKNet and the International Society on Thrombosis and Haemostasis.

EDS Disease Group

***Marco Castori** – Coordinator of the Ehlers-Danlos Syndrome (EDS) Disease Group – Fondazione IRCCS Casa Sollievo della Sofferenza, Foggia, Italy.*

The EDS Disease Group presented its educational programme for 2025, including two dedicated webinars and a comprehensive accredited online module covering diagnosis, clinical management and rehabilitation of non-vascular EDS and hypermobility spectrum disorders.

The group also reported ongoing work on disease-specific variables for the TogethERN ReCONNET Registry and introduced an important consensus initiative to refine the clinical definition and classification of generalised joint hypermobility and hypermobility spectrum disorders.

This work aims to address current diagnostic challenges and provide clinicians with more practical and evidence-based tools for patient assessment, while complementing ongoing international efforts to update EDS classification criteria.

IIM Disease Group

***Alain Meyer** – Coordinator of the Idiopathic Inflammatory Myopathies (IIM) Disease Group – Strasburg's University Hospital, France.*

The IIM Disease Group highlighted its strong activity across education, research and clinical collaboration.

Five educational webinars have already been delivered, with two additional webinars and a new accredited training module planned for 2025.

The group reported successful implementation of multidisciplinary discussions through CPMS and the active involvement of young clinicians in developing CPMS 2.0 and new clinical practice initiatives, including guidance on Pneumocystis jirovecii pneumonia prophylaxis.

Research activities include:

- the development of Red Flags for IIM,
- lay versions of clinical practice guidelines,
- the harmonisation of registry data with the MyoNet registry to support integration with the TogethERN ReCONNET Registry.

The Disease Group also announced dedicated scientific sessions at the 2nd ERN ReCONNET International Congress focusing on registry integration, unmet therapeutic needs and patient perspectives.

IgG4 Disease Group

***Emanuele Della Torre** – Coordinator of the IgG4-related disease (IgG4-RD) Disease Group – IRCCS Ospedale San Raffaele, Milano, Italy.*

The IgG4-RD Disease Group reported remarkable progress over the past year, including the successful organisation of the International Symposium on IgG4-Related Diseases, held for the first time in Europe with ERN ReCONNET support.

The group also published the first structured Red Flags for IgG4-RD methodology in *The Lancet Rheumatology*, providing clinicians with an evidence-based tool to facilitate earlier diagnosis.

Additional achievements included the renewal of the Disease Group website and the launch of several collaborative projects, including:

- IgG4 recommendations on imaging,
- the development of patient-reported outcome measures and quality-of-life questionnaires in IgG4,
- multicentre studies on early disease manifestations of IgG4,
- implementation of IgG4 disease-specific variables within the TogethERN ReCONNET Registry.

MCTD Disease Group

***Magdalena Ciupera** – ePAG Advocate of the Mixed Connective Tissue Disease (MCTD) Disease Group – Let's stick together Association, Poland*

The MCTD Disease Group presented an extensive programme of activities combining research, education and patient advocacy. Recent achievements include scientific publications on clinical practice guidelines, quality of life and clinical case management, together with a forthcoming historical review marking 50 years of MCTD research.

The group has organised several educational webinars and approved two new patient-centred webinars for 2025, focusing on non-pharmacological management and improving communication between patients and clinicians. Ongoing research priorities include:

- the development of disease-specific variables for the TogethERN ReCONNET Registry,
- the Red Flags for MCTD project
- participation in the RarERN Path project to map patient care pathways across Europe.

The Disease Group also presented its accredited training module and previewed dedicated scientific sessions for the upcoming ERN ReCONNET International Congress addressing the challenges of diagnosis, classification and patient-centred management of MCTD and UCTD.

Closure of the meeting

The day concluded with a discussion on the value of sharing experiences across Disease Groups and strengthening collaborations both within and beyond ERN ReCONNET. Particular emphasis was placed on ensuring complementary registries rather than duplicating existing initiatives, expanding international partnerships, promoting the dissemination of Red Flags to support earlier diagnosis, and encouraging Disease Groups to learn from each other's successful approaches. These collaborative efforts continue to reinforce ERN ReCONNET's mission of improving care, research and education for people living with rare connective tissue and musculoskeletal diseases.

Day 4 _ ReCONNET Plenary and Board of Network On-Line Meeting 2024-2025 Agenda

Date of the meeting: 10th February 2025 (14:00 – 16:30 CET)

14:00 – 14:05	Introduction to the meeting	M. Mosca M. Cutolo
14:00 – 14:30	RP Disease Group	O. Sander *15 min presentation + 15 min discussion*
14:30 – 15:00	SLE Disease Group	J. Andersen *15 min presentation + 15 min discussion*
15:00 – 15:30	SS Disease Group	A. Vieira *15 min presentation + 15 min discussion*
15:30 – 16:00	SSc Disease Group	M. Matucci V. Smith S. Farrington *15 min presentation + 15 min discussion*
16:00 – 16:30	UCTD Disease Group	C. Pamfil *15 min presentation + 15 min discussion*
16:30	Closure of the meeting	M. Mosca M. Cutolo

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Day 4 – 10th February 2025

Introduction to the meeting

M. Mosca opened the fourth and last day of the Plenary Meeting, during which the Disease Groups presented all the different activities they had undertaken. She encouraged all participants to share any questions or problems they may have to find solutions together.

RP Disease Group

***Oliver Sander** – member of the Relapsing Polychondritis (RP) Disease Group – Universitätsklinikum, Düsseldorf, Germany*

The RP Disease Group showcased its strong scientific output despite working on one of the rarest diseases within ERN ReCONNET.

The group reported an extensive educational programme, including webinars for both patients and healthcare professionals and the development of an accredited e-learning module covering diagnosis, treatment, Red Flags and VEXAS syndrome.

A major achievement was the development of the first disease-specific Quality of Life questionnaire for Relapsing Polychondritis (QoL-RP), created through an international multilingual collaboration involving patients from 22 countries.

The project identified the aspects of daily life most affected by the disease and generated a disease-specific assessment tool that captures issues not addressed by generic quality-of-life instruments.

The Disease Group also reported progress in defining disease-specific variables for:

- the TogethERN ReCONNET Registry
- launching the Red Flags initiative and expanding its publication activity, contributing substantially to the international scientific literature on this ultra-rare disease.

SLE Disease Group

***Jeanette Andersen** – ePAG Advocate of the Systemic Lupus Erythematosus (SLE) Disease Group – Lupus Europe, Denmark.*

The SLE Disease Group presented several flagship initiatives developed in close collaboration with Lupus Europe, demonstrating how patient organisations and clinicians can jointly create innovative educational resources.

Among the most successful projects are Lupus100, now translated into numerous languages, and LupusGPT/EasyLupus, artificial intelligence-based tools providing reliable, evidence-based information drawn exclusively from validated scientific resources.

The group also presented consultation cards to improve patient-physician communication, results from the large Living with Lupus (Swiss Knife) Survey, and ongoing recommendations for the management of rare lupus manifestations developed together with international scientific societies.

Additional highlights included:

- the SLAKE educational platform,
- the successful RarERNPath project collecting more than 800 patient stories to improve care pathways,
- the launch of disease-specific e-learning modules,
- the continued work on registry variables
- the educational materials for neuropsychiatric lupus.

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SjD Disease Group

Ana Vieira *– ePAG Advocate of the Sjögren's Disease (SjD) Disease Group – Sjögren Europe, Spain*

The SjD Disease Group focused on addressing unmet needs in a disease whose systemic nature remains widely under-recognised.

New educational webinars will explore disease stratification, neurological manifestations and childhood-onset Sjögren's Disease.

The group is also working on a European survey to better understand disease management and patient-reported outcomes across Europe and initiated a multidisciplinary project on sexual health, one of the most overlooked aspects of the disease.

Progress was reported on integrating disease-specific variables from the HarmonicSS project into the TogethERN ReCONNET Registry, while the accredited training course will include dedicated modules on childhood Sjögren's, pregnancy, lymphoma and amyloidosis in Sjögren's.

Future priorities include:

- the development of a Sjögren's 100 Questions educational resource
- updating Sjögren's disease terminology across ERN ReCONNET materials
- increasing awareness that Sjögren's Disease is a complex systemic disease rather than simply a disorder of dryness.

SSc Disease Group

Vanessa Smith *– Coordinator of the SSc Disease Group – Ghent University Hospital, The Netherlands.*

Marco Matucci Cerinic *– Coordinator of the SSc Disease Group – IRCCS Ospedale San Raffaele di Milano, Italy.*

Sue Farrington *– ePAG Advocate of the SSc Disease Group – FESCA, Scleroderma & Raynaud's UK (SRUK), UK.*

The SSc Disease Group highlighted its longstanding commitment to multidisciplinary and patient-centred care.

One of its major achievements has been the development of the first ERN ReCONNET patient pathway (RarERN Path), created through extensive collaboration between healthcare professionals and patients to identify common needs and improve clinical management.

Other ongoing initiatives include:

- surveys on digital ulcers,
- patient education materials such as capillaroscopy infographics
- multilingual webinars
- accredited training modules covering neglected but clinically relevant topics including calcinosis, oral health, gastrointestinal involvement and soft tissue damage.

The group also described its monthly expert meetings for complex clinical cases and its efforts to integrate the existing EUSTAR registry with the TogethERN ReCONNET Registry, avoiding duplication while maximising the value of established European datasets.

Throughout the presentation, the importance of patient partnership was emphasised as a key driver for identifying unmet needs and improving quality of care.

UCTD Disease Group

Cristina Pamfil *– Coordinator of the Unclassified Connective Tissue Disease (UCTD) Disease Group – County Emergency Clinical Hospital Cluj, Romania.*

The UCTD Disease Group presented a comprehensive programme aimed at improving the diagnosis and long-term management of patients with undifferentiated connective tissue disease.

Ongoing work includes the development of disease-specific variables for the TogethERN ReCONNET Registry to better characterise clinical phenotypes, monitor disease progression, evaluate treatments and assess pregnancy outcomes.

The group is also preparing a consensus initiative to harmonise the definition and diagnosis of UCTD, addressing one of the major challenges in the field.

Additional projects include:

- the development of Red Flags to identify patients at higher risk of progression to defined connective tissue diseases,
- practical guidance to help patients prepare for medical consultations,
- accredited educational modules on diagnosis, treatment, pregnancy and vaccination,
- a series of educational webinars addressing key clinical topics.

Closure of the meeting

M. Mosca remarked that the four-day Plenary Meeting of ERN ReCONNET was extraordinary, during which the full spectrum of work and the progress achieved as a community could be observed. She asserted that ERN ReCONNET is implementing projects of outstanding quality, with clearly defined goals and increased awareness. She highlighted the introduction of the new CPMS, a pivotal component of ERN's efforts, underscoring its significance in the context of complex clinical cases. Furthermore, she referred to the educational projects, which encompass not only webinars but also a more structured educational project. It was explained that the 50 students selected for the accredited course are required to complete the entire programme. The objective is not to get a separate accreditation for each module, but rather an overall accreditation for the entire educational project.

Moreover, Prof. Mosca conveyed her profound gratitude for the patients' active participation in the educational project, emphasising its significance and unexpected nature. She acknowledged the team's efforts in implementing the governance changes, and expressed her gratitude to all the participants for their presence, commitment, and discussions, identifying the latter as significant opportunities for improvement.

Thereafter, the floor was given to M. Cutolo, who expressed his satisfaction with the active involvement of patients in the dissemination of their data during the Plenary Meeting. He emphasised that this exemplifies the ReCONNET Network's superior quality, given that it facilitates patient-physician interaction within the projects. Furthermore, M. Cutolo noted that many rare diseases initially presented a notable intrinsic complexity in their description and understanding. However, the collaboration of patients and healthcare professionals within the Network has led to a substantial enhancement in the knowledge about these diseases and a notable progress in their comprehension, particularly among the patients themselves. He emphasised that the significance of persevering, collaborating, and producing genuinely original results for ERN ReCONNET was fully acknowledged. He expressed his deep gratitude to M. Mosca for her tireless efforts in assuming the role of moderator and coordinator, and acknowledged the contributions of the entire Coordination Team. He concluded by extending an invitation to the *2nd International ERN ReCONNET Congress*, which is to be held in Prague in April.