




# Rare respiratory diseases are ready for primetime: from Rare Disease Day to the European Reference Networks

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The ERN-LUNG network is currently made up of 60 centres in 12 countries and is focused on rare respiratory diseases <http://ow.ly/CG0d3089xDC>

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Rare diseases are life-threatening or chronically debilitating conditions which are of such low prevalence (fewer than 1 in 2000 people) that special combined efforts are needed to address them [1]. Launched in 2008 by Rare Diseases Europe (EURORDIS), a non-governmental patient-driven alliance representing 733 rare disease patient organisations in 64 countries, Rare Disease Day (figure 1) takes place on the last day of February each year. The main objective of Rare Disease Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives [2]. In 2017, Rare Disease Day will emphasise that research brings hope to people living with a rare disease [2]. Indeed, due to the rarity and diversity of rare diseases, research needs to be international to ensure that experts, researchers and clinicians are connected, that clinical trials are multinational, and that patients can benefit from the pooling of resources across borders [2].

Supporting education, research and care related to rare diseases is very timely in Europe, with the announcement on December 15 2016 that the European Commission's Board of Member States had approved 23 European Reference Networks (ERNs), including one for rare respiratory diseases (ERN-LUNG; table 1) [3]. Furthermore, in order to share knowledge and expertise more efficiently, the European Union Committee of Experts on Rare Diseases (EUCERD) has adopted recommendations on quality criteria for centres of expertise in rare diseases in member states [4]. Centres of expertise should allow the concentration of both expertise and patient numbers in one place to tackle rare or complex diseases and conditions more effectively [3, 4]. Developing centres of expertise and ERNs in the field of rare diseases was proposed in the European Council recommendation for action in the field of rare diseases and more recently in the cross-border healthcare directive as a means of organising care for the thousands of heterogeneous rare conditions affecting scattered patient populations across Europe [4].

The ERN-LUNG is a network of European healthcare providers dedicated to ensuring and promoting excellence in care and research for the benefit of patients affected by rare respiratory diseases.

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FIGURE 1 The logo of Rare Disease Day ([www.rarediseaseday.org](http://www.rarediseaseday.org)).

ERN-LUNG's vision is to be a European knowledge hub for such diseases and to decrease morbidity and mortality from them in people of all ages. It is a non-profit, international, professional, patient-centric and scientific network and it is committed Europe-wide and globally to the prevention, diagnosis and treatment of rare respiratory diseases through patient care and advocacy, education and research. In keeping with these goals, ERN-LUNG will interact with both national and international organisations which have similar goals.

ERN-LUNG is currently made up of 60 centres in 12 countries and is organised into nine core networks representing the diversity of diseases and conditions affecting the respiratory system (figure 2, table 2). The current core networks are interstitial lung diseases [5–21], cystic fibrosis [22–25], pulmonary hypertension

TABLE 1 Outcome of the 2016 call [3] to establish European Reference Networks (ERNs), as approved by the European Commission's Board of Member States on December 15 2016

| Network                     | Focus   |
|-----------------------------|---|
| <b>ERN BOND</b>             | ERN on rare bone disorders  |
| <b>ERN CRANIO</b>           | ERN on rare craniofacial anomalies and ENT disorders                    |
| <b>Endo-ERN</b>             | ERN on rare endocrine conditions  |
| <b>ERN EpiCARE</b>          | ERN on rare and complex epilepsies                                      |
| <b>ERKNet</b>               | European rare kidney diseases reference network                         |
| <b>ERN RND</b>              | ERN on rare neurological diseases                                       |
| <b>ERNICA</b>               | ERN on rare inherited and congenital anomalies                          |
| <b>ERN-LUNG</b>             | ERN on rare respiratory diseases  |
| <b>ERN Skin</b>             | ERN on rare and undiagnosed skin disorders                              |
| <b>ERN EURACAN</b>          | ERN on rare adult cancers (solid tumours)                               |
| <b>ERN EuroBloodNet</b>     | ERN on rare haematological diseases                                     |
| <b>ERN EURO-NMD</b>         | ERN for rare neuromuscular diseases                                     |
| <b>ERN EYE</b>              | ERN on rare eye diseases  |
| <b>ERN GENTURIS</b>         | ERN on genetic tumour risk syndromes                                    |
| <b>ERN GUARD-HEART</b>      | ERN on uncommon and rare diseases of the heart                          |
| <b>ERN ITHACA</b>           | ERN on rare congenital malformations and rare intellectual disabilities |
| <b>MetabERN</b>             | ERN for rare hereditary metabolic disorders                             |
| <b>ERN PaedCan</b>          | ERN for paediatric cancer (haemato-oncology)                            |
| <b>ERN RARE-LIVER</b>       | ERN on rare hepatological diseases                                      |
| <b>ERN ReCONNET</b>         | Rare connective tissue and musculoskeletal diseases network             |
| <b>ERN RITA</b>             | Rare immunodeficiency, autoinflammatory and autoimmune diseases network |
| <b>ERN TRANSPLANT-CHILD</b> | ERN on transplantation in children                                      |
| <b>VASCERN</b>              | ERN on rare multisystemic vascular diseases                             |

ENT: ear, nose and throat.

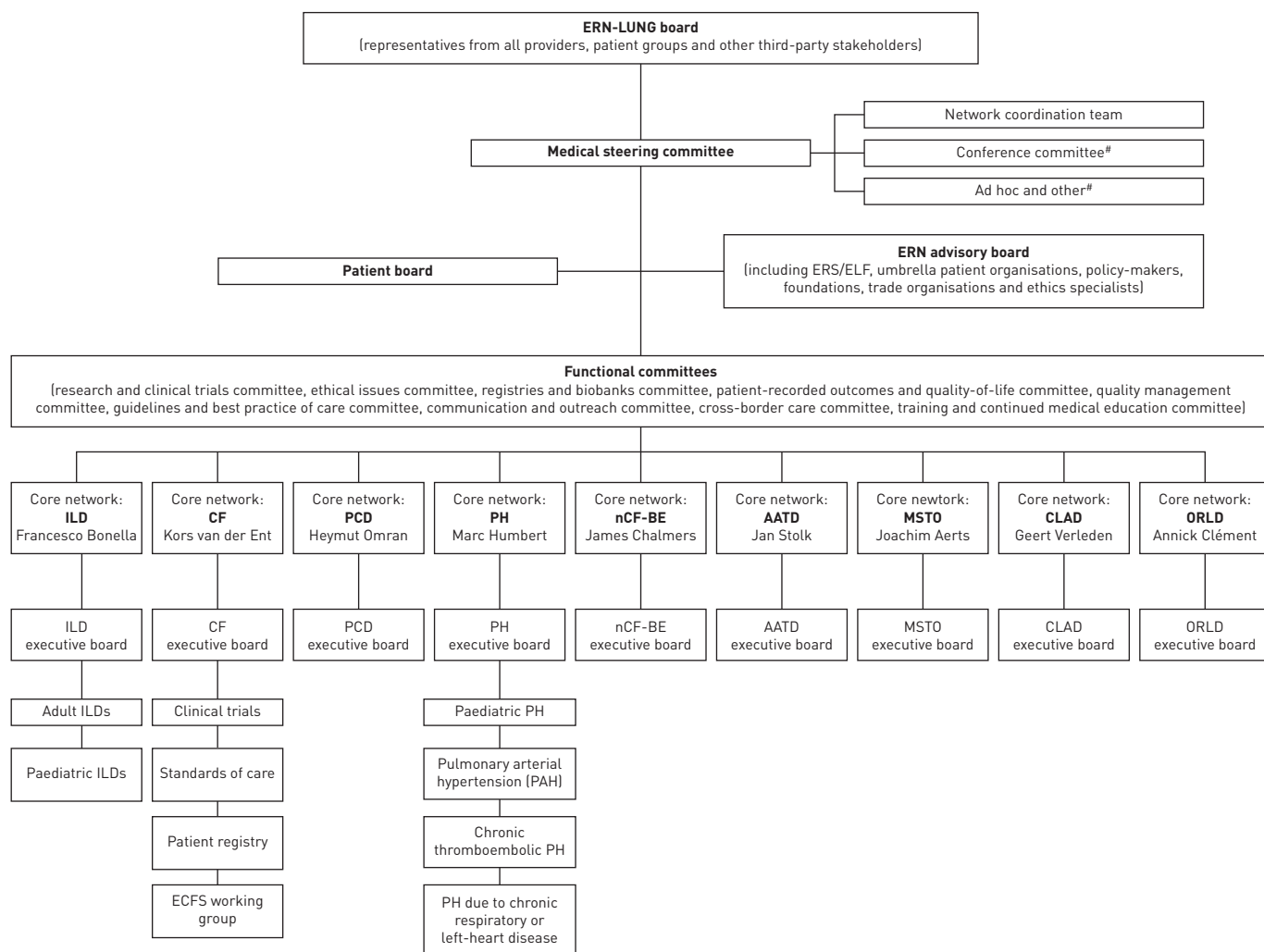


FIGURE 2 The structure of the European Reference Network on rare respiratory diseases (ERN-LUNG). ERS: *European Respiratory Society*; ELF: *European Lung Foundation*; ILD: interstitial lung disease; CF: cystic fibrosis; PCD: primary ciliary dyskinesia; PH: pulmonary hypertension; PAH: pulmonary arterial hypertension; nCF-BE: nonCF-bronchiectasis; AATD:  $\alpha_1$ -antitrypsin deficiency; MSTO: mesothelioma; CLAD: chronic lung allograft dysfunction; ORLD: other rare lung diseases. #: if installed by the Medical Steering Committee

[26–31], primary ciliary dyskinesia [32–37], non-cystic fibrosis bronchiectasis [38–40],  $\alpha_1$ -antitrypsin deficiency [41], mesothelioma [42], chronic lung allograft dysfunction [43–45] and “other rare lung diseases” (e.g. pulmonary malformations, congenital central hypoventilation syndrome etc) [46–49]. In addition to these thematic subgroups, ERN-LUNG is also organised into functional committees whose aims are to tackle overarching topics affecting all of the current and future core networks, including research and clinical trials, ethical issues, registries and biobanks, patient-reported outcomes and quality-of-life, quality management, guidelines and best practice of care, communication and outreach, cross-border care, and training and continued medical education.

The ERN-LUNG’s current set-up will evolve with the inclusion of more members and partners after it is formally established. The creation of a ninth core network on “other rare lung diseases” is intended to serve as an intermediate solution to the question of the formal creation of additional core networks over the next 5 years, covering groups that could not formally become core networks in the initial application stage. The strategic plan for ERN-LUNG is set out in a multi-annual work plan submitted in response to the call for Framework Partnership Agreements for the period 2017–2021 (Chafea: HP-ERN-2016). The plan sets out an evaluation strategy that will be followed to review the ERN-LUNG’s achievements within the first 5 years of its establishment.

The ERN awarding ceremony will take place on March 9–10, in Vilnius, Lithuania, in the presence of ERN coordinators and patients’ representatives. A few weeks later, the ERN-LUNG kick-off meeting will take place on April 4–5 in Frankfurt with all the healthcare providers and patients’ representatives attending.

TABLE 2 Disposition of the medical steering committee of the European Reference Network on rare respiratory diseases (ERN-LUNG)

| Committee position               | Candidate                                |
|----------------------------------|--|
| Patient representative           | H. de Keyser, Brussels, Belgium          |
| Patient representative           | G. Meszaros, Budapest, Hungary           |
| Network coordinator              | T.O.F. Wagner, Frankfurt, Germany        |
| Network vice coordinator         | M. Humbert, Paris, France                |
| ILD core network coordinator     | F. Bonella, Essen, Germany               |
| CF core network coordinator      | K. van der Ent, Utrecht, The Netherlands |
| PCD core network coordinator     | H. Omran, Münster, Germany               |
| PH core network coordinator      | M. Humbert, Paris, France                |
| PH core network vice coordinator | N. Galiè, Bologna, Italy                 |
| nCF-BE core network coordinator  | J. Chalmers, Dundee, UK                  |
| AATD core network coordinator    | J. Stolk, Leiden, The Netherlands        |
| MSTO core network coordinator    | J. Aerts, Rotterdam, The Netherlands     |
| CLAD core network coordinator    | G. Verleden, Leuven, Belgium             |
| ORLD core network coordinator    | A. Clément, Paris, France                |
| Network coordination team        | A. Pfalz, Frankfurt, Germany             |

ILD: interstitial lung disease; CF: cystic fibrosis; PCD: primary ciliary dyskinesia; PH: pulmonary hypertension; nonCF-BE: nonCF-bronchiectasis; AATD:  $\alpha_1$ -antitrypsin deficiency; MSTO: mesothelioma; CLAD: chronic lung allograft dysfunction; ORLD: other rare lung diseases.

The *European Respiratory Society* has always been very supportive of rare pulmonary disease research, education and care, and the society's publications regularly disseminate novel data and guidelines in these fields [50, 51]. Of note, the society will endorse the 7th International Meeting on Pulmonary Rare Diseases and Orphan Drugs, which will take place in Milan, Italy on February 24–25. This conference, chaired by Dr Sergio Harari, Chief Editor of the *European Respiratory Review*, is dedicated entirely to rare pulmonary diseases and rare variants of more common pulmonary conditions. The proceedings from this conference will be published in the *European Respiratory Review* and updates from the ERN-LUNG will feature regularly in the *European Respiratory Journal*. Rare respiratory diseases are indeed ready for primetime!

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