An interview with Research Policy and Initiatives Director Roseline Favresse at the European Organisation for Rare Diseases

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Abstract

Roseline Favresse is the Research Policy and Initiatives Director at the European Organisation for Rare Diseases (EURORDIS). We interviewed Roseline to learn more about the rare disease landscape within Europe and the exciting recent collaborations to push research and innovation for rare diseases forward into 2025 and beyond.

Roseline Favresse is the Research Policy and Initiatives Director at European Organisation for Rare Diseases (EURORDIS). She has extensive experience in supporting the rare disease community across Europe and has been instrumental in coordinating research projects and programmes to improve rare disease education and training. This includes the development of an online open academic course entitled "Diagnosing Rare Diseases: from the Clinic to Research and Back" to train people interested in diagnostic research and rare diseases. Roseline has specialised in setting up, developing, and managing capacity-building programmes in Europe and internationally. We are honoured to have had the opportunity to interview Roseline about the rare disease landscape in Europe and about exciting recent collaborations to advance research and innovation in rare diseases.

***Medical Writing* (MW):** For our readers unfamiliar with EURORDIS, could you please describe who you are, what you do, and your mission?

Roseline Favresse (RF): EURORDIS represents over 1000 rare disease patient organisations in 74 countries across Europe and beyond, with the goal of improving the lives of the roughly 30 million people living with a rare disease.[1] We bring together stakeholders, such as clinicians, researchers, patients, families, funders, and policymakers, to allow patient voices to be heard and to help shape policy. Our mission is to work across borders and all rare diseases, including rare cancers, to improve all aspects of patients' lives. We have a three-fold strategy: advocating for people with rare diseases by working with the EU Commission and the EU Parliament; providing people with rare diseases with the tools to self-advocate through training and mentoring programmes; and also partnering them with relevant stakeholders. Our work aims to empower patients and their families so that they are recognised as equal citizens with equal rights and to ensure that people with rare diseases receive timely diagnoses.

> Early diagnosis can be improved through newborn screening programmes.

MW: What are the most significant challenges/needs reported to you from families with rare diseases?

RF: The biggest challenge is the time it takes to obtain a diagnosis. A recent retrospective patient survey using Rare Barometer, a survey initiative of EURORDIS, collected the experiences and opinions of 10,453 people living with rare diseases and their close family members in 42 European countries.[2] The survey found that it takes, on average, 5 years to diagnose a rare disease. Other key findings were that 60% of patients were misdiagnosed with a physical condition, 60% were misdiagnosed with a psychological condition, 40% had not been referred to a specialist centre, and 25% had eight or more consultations with a healthcare professional before obtaining a diagnosis (Figure 1).[3] Women have a longer diagnosis journey than men (5.4 years vs. 3.7 years). Also, children and adolescents have a longer diagnosis journey, 8.8 years and 10.4 years, respectively, which may be because symptoms are attributed to the onset of puberty. Improvements must be made in appropriate and consistent coding of symptoms and data collection to advance research.

MW: Do these challenges differ across Europe?

RF: Early diagnosis can also be improved through newborn screening programmes, and 95% of respondents in a Rare Barometer survey were in favour of performing tests to diagnose a rare disease at birth.[4,5] While widely accepted across Europe, there remain discrepancies in the availability and number of conditions included in the screening tests.[6] Certain countries do not have a newborn screening programme, while Italy has the most comprehensive screening with 48 conditions.[7] Many ongoing pilot research programmes aim at improving these newborn screening programmes at the international and local levels.

MW: The European Economic and Social Committee has called for a European flagship initiative for health and to publish an Action Plan on rare diseases with achievable targets by 2023. Has Brexit affected the inclusion of the UK in these plans? If so, how?

RF: From a EURORDIS perspective, we still work with UK groups as before. However, there has been a direct impact on the five or six European reference networks coordinated from the UK, which have a wealth of clinical

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experience. Some of the coordinators from reference networks have relocated to countries within the EU. While they can still collaborate with EURORDIS, the UK centres are no longer assured partners of the European network, which is not ideal. The situation for research is not as bad as anticipated. After some anxious months and negotiations, the UK remains associated with Horizon Europe,[8] the EU's funding programme for research and innovation.

MW: The European Rare Diseases Research Alliance (ERDERA) was launched in September 2024 to address research and funding gaps in rare diseases. Could you briefly explain its origins, mission, and key goals?

RF: ERDERA is an alliance between the European Union and member states, with around 180 partners from 37 European countries and beyond.[9] This is a 7-year initiative, with a budget of €380 million funded by Horizon Europe, member states, and public and private partners. It is the largest co-funded partnership for rare diseases in research and innovation.

The objective is to support patient-driven research aligning with International Rare Disease Research Consortium (IRDiRC), established by the European Commission and the US NIH in 2011. The ultimate aims are to reduce the time to diagnosis to 6 months once patients have seen a medical specialist; have 1000 new therapies approved to offer treatment to the currently 95% rare diseases with no therapeutic option available; and improve evaluation and understanding of the impact of rare diseases on patients, families, and health care systems to inform policy decisions.

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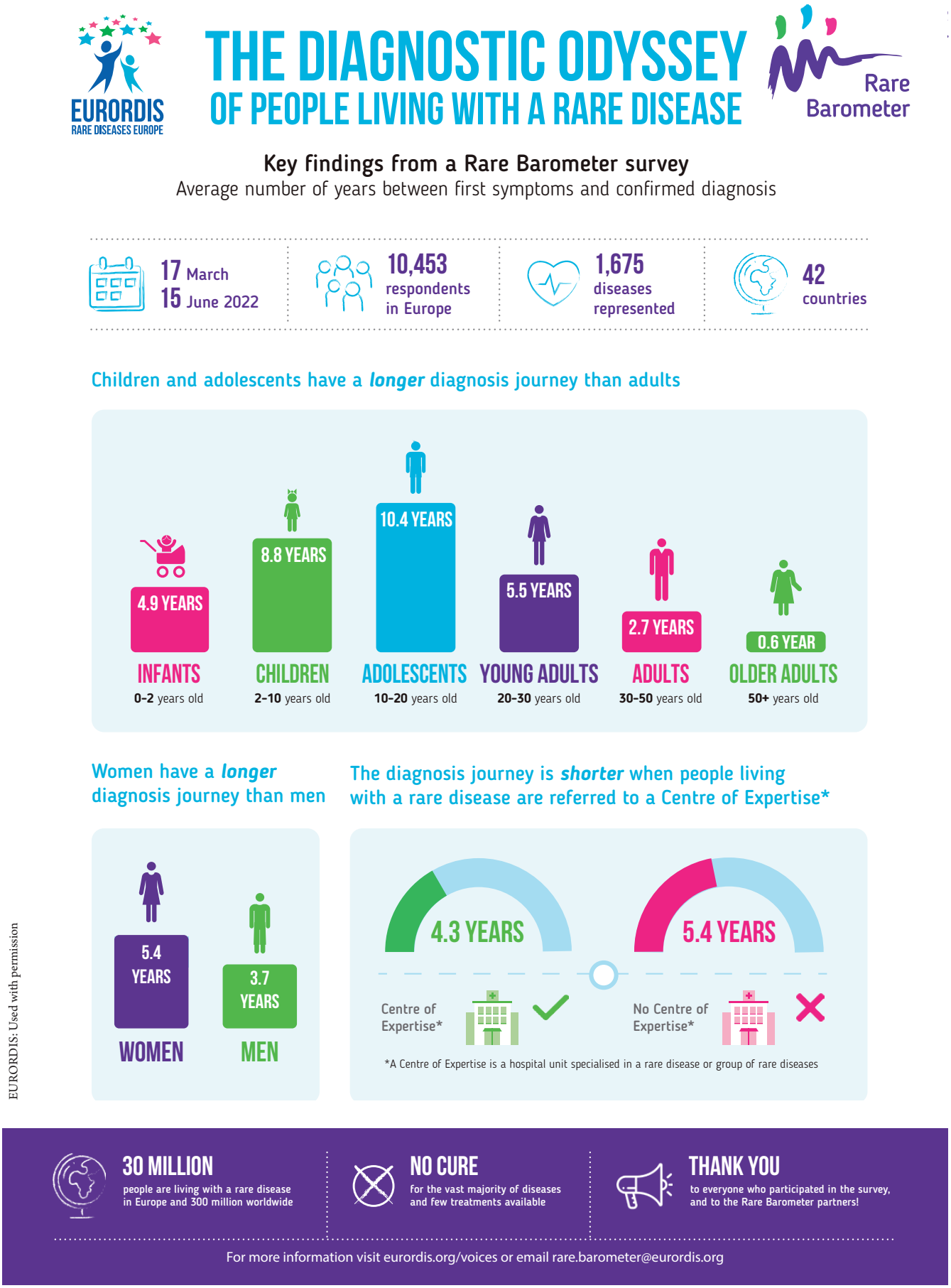
There are 25 packages with activities ranging from funding research into rare diseases to improving education and data collection, integration, analysis, and sharing at a global scale. ERDERA will develop training programmes for patients and young researchers and establish a Master's degree in rare diseases. At the moment, rare diseases are missing from the medical school curriculum in many countries, and there is a need to educate the next generation of physicians, paediatricians, and primary care practitioners.

There are services provided to the rare disease ecosystem with the goal of accelerating the translation of research into clinical development, diagnostics, and treatments. The clinical research network activities will coordinate the regulatory, outcome assessments, and data aspects in preparation for clinical trials.

There is a subcomponent in ERDERA to support national mirror groups, focusing on local issues, as well as countries currently under-represented in Horizon Europe (e.g. Portugal, Lithuania, Croatia) to ensure



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The Diagnostic Odyssey of People Living with a Rare Disease

equity of research development and access to services for rare diseases.

MW: What do you predict will be the most exciting news in 2025 for the rare disease community?

RF: We have been advocating for an EU Action Plan for rare diseases for many years, which is gaining momentum and is being endorsed by members of the EU Parliament. Also, internationally, Rare Diseases International has launched a campaign for a World Health Assembly Resolution on Rare Diseases in 2025.[10] Gaining a commitment from the WHO will improve awareness activities at a country level, setting clear targets and deadlines regarding improving diagnosis, access to care and treatment, and research development.

MW: For medical writers working in the rare disease field, what is the one quality or piece of knowledge you would like them to have to support the rare disease community in their work?

RF: Rare diseases are diverse; an individual's needs will differ from one condition to another in terms of severity and prognosis. Medical writers need the quality of empathy. For all the positive stories of advocacy we hear about, we should also look at what is behind their stories. It is essential to communicate about the ordinary and not the extraordinary. Even positive experiences will not have been without significant challenges. Also, bear in mind that not everyone has the capacity to do advocacy, which may be due to their disease, their motivation, or a variety of personal reasons. We should recognise that a lot of people with a rare disease struggle with their quality of life. For some, it is impossible to embark on researching a cure for their condition, especially as they often do not have a clear diagnosis. Living with a rare disease can be overwhelming for the average family, as not everybody relates to their daily living challenges. Individuals are frequently alienated by the healthcare environment, where their needs have been persistently overlooked. A lot has changed in a decade, though, with regard to advocacy and awareness.

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Disclosures and conflicts of interest

The author declares no conflicts of interest.

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Author information

Heather L. Mason has 14 years of freelance expertise across diverse therapeutic areas. Her passion for rare diseases emerged from professional experience and personal insight, driving her commitment to patient advocacy and inclusive healthcare communications. She is dedicated to translating complex medical information into accessible content that serves the scientific community and patient populations.