Press release

New global Rare Barometer survey on the journey to diagnosis for people living with a rare disease

***21 March 2022, Paris* – The EURORDIS survey programme Rare Barometer has launched an international survey on the journey to diagnosis for people living with a rare disease, with the support of Rare Diseases International (RDI). The survey is open to people living with a rare disease and their family members from any country in the world, and is translated in 26 languages.**

Rare disease patients report that obtaining a diagnosis can be a long and difficult journey, with enormous consequences for patients and their families. Early diagnosis is one of the first keys to better management of symptoms and the course of the disease before it has an irreversible effect on the patient's health. This opens the door to specialists and health services that can ensure better rare disease care pathways.

> **Sandra Courbier**, **EURORDIS Social Research Director - Rare Barometer Programme Lead** commented: "We want to understand the journey people living with a rare disease are going through when seeking a diagnosis for their rare disease, by measuring the time needed to obtain a diagnosis, or the consequences of being undiagnosed or misdiagnosed, as well as the role of new technologies."

Great advances in science and technology, such as newborn screening and advanced therapies, have provided new ways to diagnose a disease, but this needs to be harmonised and strengthened across Europe, as set out in our call for a European Action Plan for rare diseases.

> "There have been a number of national or disease-focused surveys on the journey to diagnosis for people living with a rare disease. This global survey will supplement existing facts and figures and provide a unique opportunity to compare the differences between countries and regions to obtain meaningful data per disease or group of diseases at an international level and identify best practices," added **Courbier**. "As such, by identifying personal and external factors influencing the process of obtaining a diagnosis from a patient perspective, we will contribute to developing policy recommendations for shortening and improving the patient journey to diagnosis."

The survey will help support and generate new knowledge about the diagnosis journey from the perspective of people living with a rare disease worldwide. Based on a mixed method approach, a combination of qualitative and quantitative methods, it will provide an opportunity for rare disease patients to have a strong voice, as they have been involved in every step of the survey development. Prior to the launch, patient representatives participated in an online panel to

exchange views and share their experiences of obtaining a diagnosis, which was followed by eight individual interviews. The findings have been used to guide the questionnaire, with the support of an expert committee composed of relevant experts in the field of diagnosis, including policy experts, patient advocacy organisations, sociologists, and corporate partners.

About Rare Barometer, EURORDIS survey programme

The Rare Barometer programme brings together close to 15,000 rare disease patients, family members and carers who share their experiences and opinions on the issues that matter to the rare disease community.

The Rare Barometer survey software enables high-quality, secure data collection and analysis. The Programme was created to systematically collect patients' opinions on transversal topics and introduce them into the policy and decision-making process, transforming patients' and families' opinions and experiences into figures and facts that can be shared with a wider public and policymakers.

About EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of 984 rare disease patient organisations from 74 countries that work together to improve the lives of the 30 million people living with a rare disease in Europe. By connecting patients, families, and patient groups, as well as by bringing together all stakeholders and mobilising the rare disease community, EURORDIS strengthens the patient voice and shapes research, policies and patient services.

About Rare Diseases International

Rare Diseases International (RDI) is the global alliance of Persons Living with a Rare Disease and families across the globe. RDI brings together national, regional and international organisations active in over 100 countries. As an alliance, RDI works for greater equity for all Persons Living with a Rare Disease and families across the globe.

About rare diseases

The European Union considers a disease as rare when it affects less than 1 in 2,000 citizens. Over 6,000 different rare diseases have been identified to date affecting an estimated 30 million people in Europe and 300 million worldwide. Due to the low prevalence of each disease, medical expertise is rare, knowledge is scarce, care offerings inadequate and research limited. Despite their great overall number, rare disease patients are the orphans of health systems, often denied diagnosis, treatment and the benefits of research.

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