Press Release

Major survey reveals lengthy diagnostic delays for rare disease patients

***14 May 2024, Paris* – EURORDIS-Rare Diseases Europe has released findings from an extensive Rare Barometer survey highlighting the prolonged and inequitable diagnostic journeys faced by rare disease patients right across Europe.**

Conducted between 17 March and 15 June 2022, the landmark Rare Barometer survey encompassed responses from over 10,000 patients from 42 countries, representing 1,675 rare diseases.

Speaking about the survey's findings, **Jessie Dubief**, Social Research Director at EURORDIS and corresponding author of the Rare Barometer study, said:

> "The survey's findings reveal an average diagnostic journey of nearly five years, with marked variations depending on demographic and geographic factors. Notably, women and young patients face longer diagnostic times, underscoring a pressing need for tailored approaches to improve diagnosis rates, reduce waiting times, and tackle systemic inequities."

Responding to the survey's findings, **Virginie Bros-Facer**, Chief Executive Officer of EURORDIS, said:

> "We urge EU and national policymakers to prioritise enhancing diagnostic services, as our findings show unacceptable delays that compromise patient care and quality of life. Every day without a diagnosis is a day lost for patients and their families.
>
> "We must close the gap in diagnostic services to ensure all patients, regardless of their location, condition, gender, or age, have timely access to accurate diagnoses which in turn can open the door to subsequent care pathways.
>
> "Furthermore, we advocate for improved support for undiagnosed conditions and equitable access to diagnostic technologies across Europe. The policymakers of the EU's next five-year legislative term are uniquely well-positioned to lead substantial improvements in these areas, with their extensive regulatory capabilities and resources, and the commitment to building a European Health Union. They must support national efforts to streamline and enhance diagnostic protocols and address the specific needs of the undiagnosed community."

The key findings

1. **The average rare disease patient waits half a decade for a diagnosis**

The survey illuminates that the average time from the first onset of symptoms to a confirmed diagnosis for rare disease patients stretches to **4.7** years (more than four years and eight months).

Additionally, **25%** of those surveyed reported received eight or more consultations with a healthcare professional before obtaining a confirmation of their diagnosis.

This prolonged diagnostic journey not only delays necessary treatments but also prolongs patient suffering and uncertainty, exacerbating the challenges faced by those living with rare diseases.

2. **The majority of rare disease patients experience misdiagnosis**

The survey shows that a staggering **60%** of surveyed individuals were initially misdiagnosed with a different physical disease, while **60%** faced misdiagnosis with a psychological condition or had their symptoms dismissed altogether.

These misdiagnoses can lead to inappropriate treatments that may worsen the patient's condition or leave the actual disease unmanaged, highlighting a critical area for improvement in medical training and awareness of rare diseases.

3. **Gender and age are major determinants of diagnostic journey lengths**

The survey reveals that significant disparities in the duration of diagnostic journeys exist based on gender and age, with women and children notably experiencing longer waits for accurate diagnoses compared to men and adults.

Women rare disease patients wait an average of **5.4** years to receive a diagnosis, compared to **3.7** years for men.

Adolescents (10-20 years old) endure the longest diagnostic wait at **10.4** years, while older adults (over 50 years old) face the shortest wait at **0.6** years.

On average, infants (0-2 years old) spend **4.9** years waiting for a diagnosis, children (2-10 years old) wait **8.8** years, young adults (20-30 years old) wait **5.5** years, and adults (30-50 years old) wait **2.7** years.

These findings relating to the factors of gender and age underscore the necessity for targeted educational programs and diagnostic protocols that address and mitigate these inequalities.

4. **Centres of Expertise clearly speed up diagnostic journeys**

40% of patients surveyed reported that they were *not* referred to a Centre of Expertise, yet those referred to such centres experienced significantly shorter diagnostic journeys. Those referred to a

Centre of Expertise average a **4.3**-year wait for diagnosis, compared to a **5.4**-year wait for those not referred – a difference of **1.1** years.

This difference underscores the need for more such centres and increased referrals, which could greatly improve diagnostic processes for rare disease patients across Europe.

The next steps

Today, EURORDIS and the rare disease community are convening both in Brussels and online for the *12th European Conference on Rare Diseases and Orphan Products (ECRD 2024)*. A key focus will be improving diagnostic pathways across Europe.

Discussions at ECRD 2024 will also address policy action calls in the EU for the next five-year term following the European Parliament elections and the appointment of the European Commission, as highlighted in EURORDIS' *#ActRare2024* campaign. This campaign focuses on eight critical policy areas, including promoting earlier, more accurate diagnosis.

In the area of diagnosis, EURORDIS has outlined the **following policy recommendations** for the EU's incoming policymakers:

- **Promote Equal Access:** The EU should ensure comprehensive screening and diagnostic services are equally accessible to all European residents.
- **Strengthen Networks**: The EU should enhance collaboration within European networks of specialised healthcare providers, including supporting European Reference Networks (ERNs) for rare diseases and facilitating cross-ERN expert panels to guide from diagnosis to high-quality care.
- **Support Advanced Diagnostics:** The EU should promote research and the equitable implementation of advanced diagnostic technologies at all healthcare stages, including newborn screening and genome sequencing.
- **Enhance Data Systems:** The EU should prioritise improving the interoperability and standardisation of data systems for effective diagnostics, focusing on transnational collaboration for complex and rare diseases.
- **Recognise Undiagnosed Patients:** The EU should identify undiagnosed rare disease patients as a vulnerable group and develop a comprehensive European approach to ensure they receive optimal care, regardless of diagnosis status.
- **Assist in Decision-Making:** The EU should support Member States in crafting prevention strategies, including uniform newborn screening programmes across the EU, supported by an EU-level multistakeholder expert group.
- **Improve Training:** The EU should help Member States establish training for primary and emergency care personnel to enhance patient triage and referrals to specialist centres.

Read the results in full

- **Read the full paper**
- **Download the factsheet**

About EURORDIS-Rare Diseases Europe

EURORDIS-Rare Diseases Europe is a unique, non-profit alliance of over 1,000 rare disease 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 people, families, and rare disease 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 services.

About Rare Barometer

Rare Barometer is the survey programme run independently by EURORDIS-Rare Diseases Europe and is a not-for-profit initiative. It conducts regular studies to identify the perspectives and needs of the rare disease community in order to be their voice within European and International initiatives and policy developments. Rare Barometer brings together more than 20,000 people living with a rare disease or family members to make the voice of the rare disease community stronger.

Contact

Julien Poulain
Communications Manager
EURORDIS-Rare Diseases Europe
Julien.poulain@eurordis.org
+33 6 42 98 14 32