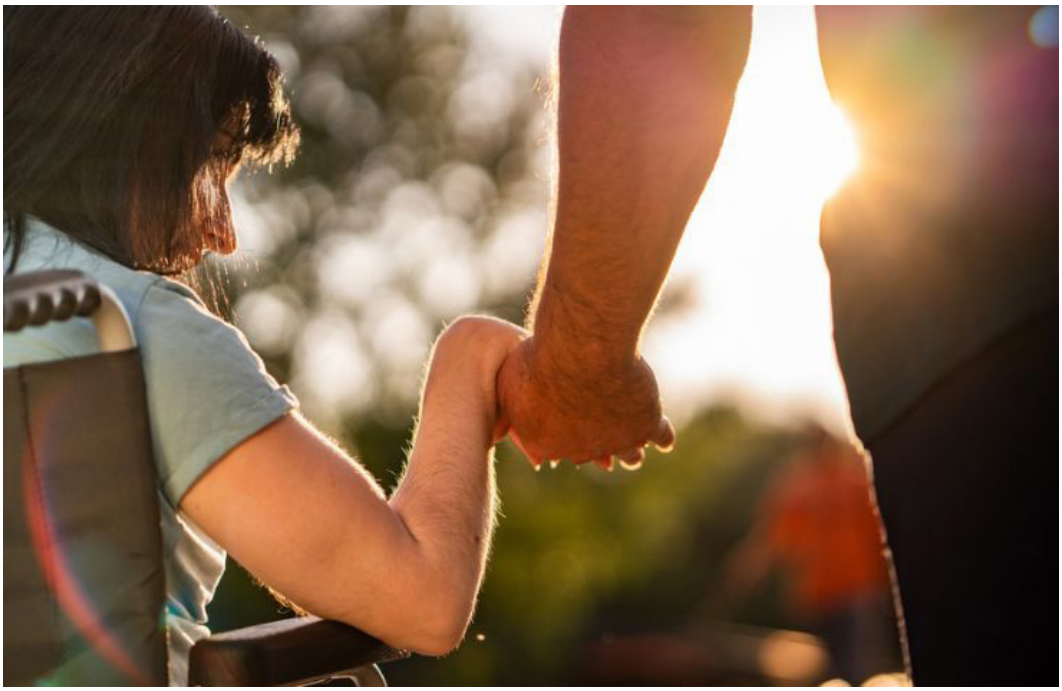

# Strength in numbers: How the Rare Barometer supports the rare disease community

*On Rare Disease Day, we talked with EURORDIS on how thousands of patient surveys can inform the future of diagnostics, care, and policies*

TO DATE, EXPERTS HAVE IDENTIFIED more than 6000 individual rare diseases, some of which are associated with only a handful of cases. Collectively, however, 300 million people worldwide live with a rare disease. And it impacts far more than that number—touching family members, friends, educators, and neighbors.

February 28 marks the internationally recognized Rare Disease Day. The day is organized by EURORDIS-Rare Diseases Europe,[1] a nonprofit alliance of more than 1000 rare disease organizations in 74 countries. Rare Disease Day is not just about raising awareness and amplifying the community's voice, it is a call to action to address unmet needs related to better diagnostics, care, policies, and expanded research. This year's campaign theme is "More than you can imagine." It celebrates the power of resilience and connection while also calling attention to the hidden struggles that people and families with rare disease face daily.

EURORDIS has been giving a voice to the rare community ever since it was founded in 1997. One of the most powerful ways it does this is through the Rare Barometer, which launched in 2016. The Rare Barometer is a program that conducts regular surveys to identify the perspectives and needs of the rare disease community.

"Our stories and voices are powerful, but there is also strength in data—and that's what the Rare Barometer brings to the table," says EURORDIS Social Research Director Jessie Dubief, who leads the program. "Not only does it ensure that our priorities are informed by the needs of our community, but it also ensures that every voice is heard."

As part of the Rare Barometer, Dubief and her team conduct both qualitative and quantitative surveys, released at least once a year, with thousands of respondents. The surveys collect evidence on everything from daily life, time to diagnosis, and mental health to impacts on family and other relationships. The surveys, translated into 25 languages, are critical for use in evidence-based advocacy efforts and help ensure that all member organizations can best represent the experiences and needs of patients and their families across Europe.

"EURORDIS is an alliance of patient associations, and as such we usually have a good understanding of the

*1. eurordis.org*

needs of the rare disease community," Dubief says. "But we wanted to go further and use robust methods to make sure that we are representing their needs. And this has proven useful, because we sometimes had surprises."

The surveys inform everything EURORDIS does as an alliance, and the research has resulted in real-world change. Its 2009 report *The Voice of 12,000 Patients: Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe*[2] was based on two surveys that laid the foundation for the Rare Barometer program. The results of the report were used to advocate for what were called "Centers of Expertise," now the European Reference Networks. They ensure people have access to high-quality care in multiple disciplines. This network opened the door for specialists in various locations to share their knowledge so that, Dubief says, "the expertise is traveling and not the patient, because many patients have to go from one doctor to another and sometimes in another country."

The 2024 Rare Barometer survey, released last May, centered on what is often called "the diagnostic odyssey"—the time from the first symptom to a confirmed diagnosis. In Europe, more than 10,000 patients representing 1675 diseases responded. While the average time to diagnosis was 4.7 years, it was 5.4 years for women and 10.4 years for adolescents (ages 10 to 20). One quarter of the participants reported having eight or more doctor visits before arriving at a diagnosis. Seventy percent of respondents were initially misdiagnosed or dismissed altogether. "Misdiagnoses make the diagnostic journey longer," says Dubief, who was not surprised by these numbers. "The statistic also shows that it's hard for health care professionals to really identify the disease."

Genetic testing can provide firm answers and shorten the diagnostic odyssey. The survey mentioned three obstacles people can face when trying to access genetic testing. For some, testing was not available in their country. For others, it was too expensive—but the obstacle that most impacted time to diagnosis, Dubief says, was health care professionals being reluctant to order the test or insufficiently informed about genetic testing.

Illumina works with health care systems around the world to educate clinicians on ordering tests and understanding results. Its partnership with EURORDIS underscores, from the patients' perspective, that these efforts are still urgently needed. "We are incredibly proud to partner with EURORDIS in our shared mission to support the rare disease community," says Shirlene Badger, PhD, global patient advocacy lead at Illumina. "The Rare Barometer shines a spotlight on the issues people face in accessing genomic testing, and gives us critical insight into how we can overcome these challenges. Together, we are making strides towards a future where no one faces their rare medical journey alone."

The most recent survey, published in February 2025, focused on disability recognition, social participation, and independent living.[3] It brought clarity and reinforcement to truths that Dubief and others had suspected. "We knew that most people with a rare disease probably lived with a disability," she says. "But we didn't know that it was eight in 10, and now that we have this figure—that's very different. It helps people understand the consequences and what should be done."

During a recent webinar on the latest Rare Barometer findings, one project manager in Italy explained how the survey information changed their strategy: They decided to forgo broad campaigns and pamphlet distribution in favor of more tailored outreach efforts to pediatricians and other health care professionals, to increase their awareness and reduce time to diagnosis.

For its surveys, the Rare Barometer relies on a panel of more than 20,000 people living with rare diseases, or their close family members, who have agreed to receive email invitations to participate. It's an engaged group that understands these data-collection efforts help show the strength of the rare disease community in numbers. And EURORDIS makes sure that the respondents feel seen and heard throughout the process. "For every survey, we always send the results to the participants, in their language, and tell them how we will be using the results," Dubief says. "We explain that EURORDIS uses findings from the Rare Barometer to advocate on behalf of the entire rare disease community." This practice of transparency has built a tremendous culture of trust among the participants—as well as some degree of hope. "It's important for them to see the end results of something they were involved in. The findings aren't just numbers on a spreadsheet; they represent the community's experiences and touch them directly." ◆

*To get involved in Rare Disease Day, support EURORDIS, or find activities to join, go to rarediseaseday.org.*

*2. eurordis.org/publications/the-voice-of-12000-patients*
*3. eurordis.org/rare-barometer-findings-daily-life-survey*