



Researchers and patients join forces to battle rare diseases

Researchers, clinicians and patients are collaborating in an EU-wide alliance to advance understanding of rare diseases and speed up the development of new treatments.

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Michela Onali, an Italian project manager now living in Sardinia, never expected to find herself at the forefront of the European fight against rare diseases. But the diagnosis of a rare genetic condition turned her world upside down.

“It took six years before I was diagnosed with GNE myopathy, a rare adult-onset muscle disease,” said Onali. “Once I got it, I felt a sense of urgency to understand more about my disease and to contribute in any way I could.”

Her disease, which affects about nine in one million people, causes progressive muscle weakness and currently has no cure. Her diagnosis led Onali to become a passionate mediator and advocate for rare disease patients.

In 2019, she joined an EU-funded research initiative called the European Joint Programme on Rare Diseases that gathered 130 partners, including research organisations, patients’ organisations, funding agencies and universities from 35 countries. Her role was to speak on behalf of other patients, as part of a patient organisation.

The scale of the joint programme is significant, in line with the challenge faced. Supported jointly by the EU and by national funding bodies, it is international in scope and impact, including partners from 27 EU countries, as well as from non-EU countries such as Canada, Israel, Switzerland, Turkey and the UK.

The researchers’ aim was to create a collaborative system for scientists, medical institutions and patients to work together on research into rare diseases.

United against disease

Daria Julkowska, assistant director at the French National Institute of Health and Medical Research, was responsible for the scientific coordination of the programme.

For her, the wide-ranging collaboration that the programme established was fundamental.

“The primary goal was to unite all stakeholders under one roof, to accelerate research on rare diseases,” she said.

The five-year collaborative effort, which ended in August 2024, gave researchers and patients across Europe access to a wealth of shared data and research tools. This transformed research on rare diseases, which, all together, affect more than 30 million people in the EU.

Most pharmaceutical companies traditionally do not focus on rare diseases because the market for each particular disease is small. More than 90% of the up to 7 000 registered rare diseases do not have an approved treatment option.

According to the European Medicines Agency, medical and scientific knowledge about rare diseases remains scarce and scattered. This makes the researchers’ work with patients even more important because, as Onali said, patients often become the biggest experts on their own disease.

New platform connects the dots

Over five years, the programme invested more than €100 million for research on rare diseases, of which around €55 million came from the EU and the rest from participating EU countries. It used the money to support several avenues of research.

“With the money we received, and through the funding agencies we partnered with, we funded studies on diagnosis, new therapies, the natural history of diseases, and even projects that focused on social sciences and humanities. The only areas we excluded were rare cancers and rare infectious diseases, to avoid duplication with other funding programmes,” Julkowska said.

The researchers also created a [Virtual Platform](#) to connect all existing data resources and tools related to rare diseases. The platform was developed as a single entry point, ensuring that users always access the most up-to-date information.

“Before, there was nothing connecting the different types of rare disease data, now we have a portal that will continue to grow,” Julkowska said.

Indeed, the research collaboration started in 2019 will be continued and expanded through an additional EU-funded research initiative called the new European Rare Diseases Research Alliance, which kicked off in September 2024.

New programme broadens scope and ambition

The new seven-year programme brings together nearly 180 partners – universities, research organisations, healthcare providers, industry and patient organisations – from 37 countries.

With this new alliance, the rare disease research community is expanding its efforts into practical work, overseeing clinical research of possible treatments.

Julkowska, who also coordinates the new programme, said it would put in place a permanent shared structure for rare disease research in Europe, further enhancing the rare diseases ecosystem, with public funders and now also some industry partners.

She also praised the level of commitment from everyone involved.

“A great example of this dedication was during the COVID-19 pandemic, when researchers, clinicians and patients alike continued to show up and push the projects forward, despite facing very difficult challenges in their personal and professional lives,” she said.

Meanwhile, Onali has also continued to work on promoting awareness of rare diseases. In 2023, she received the Black Pearl award for her work on GNE myopathy from EURORDIS, a pan-European association of rare disease patients

“It is very important to give visibility to these advocates who really work a lot to make sure to bring out the voice of patients,” she said.

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