

Missions for the European Framework Programme 9

BETTER LIVES FOR PEOPLE WITH RARE DISEASES

Less than one year from symptoms to solutions by 2027

A proposal by [Fondazione Telethon, Italy](#)

With 30 million EU citizens affected, most of which are children, rare diseases are a huge health and social burden for patients and families. Their multiplicity and complexity must be tackled by research, technology, economics and social sciences. Europe can play a crucial role in the challenge launched by the global IRDiRC consortium to shorten the time to access available diagnoses and treatments.



KEY CRITERIA

1. BOLD, INSPIRATIONAL, WITH WIDE RELEVANCE

More than 7,000 rare diseases are known to exist. Although each of them affects less than 5 in 10,000 people in the EU, altogether 30 million people in Europe live with a rare disease. Most of them are pediatric and severely affect the lives of children and parents alike.

Diversity and complexity are major hurdles that leave too many rare diseases without solutions, be they a diagnosis, a standard of care, or a therapy. Even when any of these are available, gaining access to them is a slow process due to development, clinical, or economic limitations.

Nevertheless, research on rare diseases unravels fundamental biological pathways with significant spillover benefits over common diseases; rare diseases have proven the ideal test bed for the most advanced and innovative therapies, such as gene and cell therapy and genome editing.

However, rare diseases are neglected by major investments and available treatments are scant. The few advanced therapies that have reached the market lack a viable and sustainable economic model. In addition, the difficulty of living with a rare disease is exacerbated by isolation.

Rarity of diseases and patients calls for a wide international action so that the EU dimension is the most appropriate level to tackle this mission, in cooperation with even wider global efforts.

2. A CLEAR DIRECTION: TARGETED, MEASURABLE AND TIME-BOUND

Ameliorating lives with a rare disease by shortening the time required for receiving a diagnosis or to access treatments down to one year from coming to medical attention is a measurable goal aligned with the vision for year 2027 set by the International Rare Diseases Research Consortium (IRDiRC), with the significant input of European members.

Increasing the number of therapies for rare diseases will widen the number of people who will benefit from this enterprise and contribute to the IRDiRC goal of 1,000 new therapies by 2027.

3. AMBITIOUS BUT REALISTIC RESEARCH & INNOVATION ACTIONS

As one of the preeminent arenas in the global effort to fight rare diseases, Europe will play a crucial role in addressing the ambitious goals envisioned together with IRDiRC.

Numerous European academic teams and industrial biotech/pharma companies have advanced knowledge, built a solid array of competences and achieved therapeutic breakthroughs in the fight against rare diseases.

The 24 European Reference Networks accrue more than 900 healthcare units in 26 EU countries working on a wide range of rare diseases.

This articulated scenario makes the ambitious research and innovation actions envisaged for rare diseases realistic ones.

4. CROSS-DISCIPLINARY, CROSS-SECTORAL AND CROSS-ACTOR INNOVATION

Research and development efforts are needed to study the diseases causes and mechanisms, to test therapeutic approaches in the laboratory, to translate them to the clinic, and to engage biotech and pharma companies in the development up to market availability of new medicines. The diversity of diseases

under the “rare” category (from neurological to rare cancer, from genetic to immunology) requires the engagement of a broad spectrum of researchers in biomedicine and bioinformatics.

The regulatory path for clinical trial approval and marketing authorization will need continued refinement to meet the challenge of ultra-rare patients and the complexity of advanced therapies. Advanced sequencing and omics technologies will provide biomarkers and diagnostic means to solve undiagnosed diseases and for natural history studies. Health technology assessments will be needed to make therapies and diagnostic tools sustainable.

Ethical issues regarding storage and accessibility of sensitive personal data will need to be addressed. Finally and most importantly, patients and families will become active players in all steps of the innovation path through active participation and continued education. The European community of patients is wide, well organized and already empowered to participate in research and innovation in a real co-creation process.

5. MULTIPLE, BOTTOM-UP SOLUTIONS

A wide range of approaches will address the rare disease mission. Fundamental research will feed the clinical pipeline, while the outcomes of clinical trials will produce insights and new questions, spurring further basic research. Systems biology and bioinformatics will address new biological pathways and undiagnosed diseases.

Clinical studies will need new approaches to tackle the limited number of patients. Easily accessible and reliable telemedicine solutions will be needed.

New economic models and health policies will be required to ensure a fair access to the available therapies to all patients across Europe, no matter where the closest reference center is located. Moreover, public-private partnerships will apply innovative development models.