EU SUPPORT FOR RARE DISEASES RESEARCH

Rare diseases

Rare Diseases comprise from 6000 to 8000 life threatening or chronically debilitating diseases which affect less than five persons per 10,000 citizens in the European Union. It is estimated that rare diseases affect altogether more than 30 million people in the EU.

Many rare diseases manifest in childhood, result in a shortened lifespan and lead to a dependency on care throughout the patients’ lives causing significant human suffering to the patients and their families. Patients affected by rare diseases often spend years enduring a “diagnostic odyssey” before receiving the correct diagnosis. Most rare diseases lack effective treatments representing an enormous unmet medical need. Rare diseases patients need highly specialised health care and social services resulting in high costs for society and families.

EU funded research on rare diseases

EU funded projects respond to the challenges related to rare diseases carrying out research to develop new diagnostic tools and therapies for rare diseases. The European Commission has made major investments in research and innovation in rare diseases for more than two decades throughout the EU Framework Programmes for Research and Innovation.

Altogether more than €1.4 billion has been committed in research and innovation through the Seventh Framework Programme (FP7) and Horizon 2020 in more than 200 projects related to rare diseases. Funded projects cover nearly all fields of medicine, e.g. molecular genetics, metabolic diseases, neurology, neuromuscular and musculoskeletal disorders, cardiovascular, haematological disorders, immunology, cancer, infectious diseases, nephrology, urology, ophthalmology and dermatology. The EU funding facilitates the formation of multidisciplinary teams from universities, research organisations, healthcare providers, SMEs, industry and patient organisations from across Europe and beyond.

The European Joint Programme on Rare Diseases EJP RD launched in January 2019 aims at establishing a research and innovation pipeline ensuring rapid translation of research results into clinical applications and uptake in healthcare. EJP RD involves research funders, universities, research organisations, research
infrastructures, hospitals and patient organisations from 35 countries including 27 EU Member States, seven Associated Countries and Canada.

The International Rare Diseases Research Consortium IRDiRC aims to accelerate and coordinate rare diseases research efforts around the world involving more than 50 organisations investing in rare diseases research including the European Commission. IRDiRC has the ambitious long term vision to enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention.

**Project examples**

**Solve-RD** project has two main objectives: 1) to solve large numbers of rare diseases, for which a molecular cause is not known yet, by sophisticated combined “-Omics approaches”, and 2) to improve diagnostics of rare diseases patients through a “genetic knowledge web”.

**ImmunAID** has the overall objective to deliver a method for rapid and accurate diagnosis across all the spectrum of systemic autoinflammatory diseases (SAID), in order to improve clinical management of SAID patients.

**CARAMBA** will conduct a phase I/II clinical trial of immunotherapy with patient-derived T cells that are engineered to express a synthetic chimeric antigen receptor (CAR) specific for the myeloma antigen SLAMF7, for potential treatment of multiple myeloma.

**CLINGLIO** project aims to conduct a clinical phase IIB trial with Minerval in patients with newly-diagnosed malignant glioma.

**CureCN** project has the objective of developing a curative gene therapy for Crigler-Najjar Syndrome based on liver gene transfer with and adeno-associated virus (AAV) vector expressing the UGT1A1 transgene.

**HIT-CF** project aims at bringing personalised disease modifying therapies to cystic fibrosis patients with ultra-rare CFTR mutations by conducting a randomised, double-blind, placebo-controlled, repeated-crossover, thre-armed platform trial.

**MCDS-Therapy** has the objective of advancing the repurposing of carbamazepine for the treatment of metaphyseal chondrodysplasia, type Schmid (MCDS) through a multicentre and multinational clinical trial (Phase1, Phase2/3).

**OligoGpivotalCF** project aims at conducting a pivotal phase IIb clinical trial of inhaled alginate oligosaccharide (OligoG) for cystic fibrosis.

**RECOMB** project aims at developing stem-cell based gene therapy for the most common major category of Severe Combined Immunodeficiency (SCID), the recombination deficient SCID (RECOMB).
**TRACE** project has the objective of bringing adoptive T-cell transfer into clinical routine as a life-saving, curative and safe treatment for refractory viral infection that occurs following allogeneic stem cell transplantation.

**TUDCA-ALS** project is working on safety and efficacy of tauroursodeoxycholic acid (TUDCA) as add-on treatment in patients affected by amyotrophic lateral sclerosis.

**UshTher** project aims at conducting a clinical trial of gene therapy with dual Adeno-Associated virus (AAV) vectors for retinitis pigmentosa in patients with Usher syndrome type IB.

**E-RARE-3** ERA-NET brings together funding organisations from 14 EU Member States and Switzerland, Israel, Turkey and Canada. E-Rare-3 together with its two predecessors have provided funding over to €100 million to more than 100 collaborative projects on rare diseases research through joint calls for proposals since 2006.

**EURENOMICS** project has provided new hope for the 2 million people in Europe who suffer from rare kidney diseases thanks to ground-breaking research that has led to diagnostic tests and a new type of treatment.

**RD-CONNECT** supported a platform linking up databases, biobanks, registries and other valuable resources in support of research into rare diseases.

**NEUROMICS** project successfully applied omics-technologies for studying rare neurodegenerative and neuromuscular diseases, which resulted in the identification of over 100 new disease genes and several putative biomarkers for disease onset and progression. These achievements will greatly improve the diagnosis and monitoring of these diseases and will help developing novel therapeutic approaches.

**Alpha-Man** project contributed to the early phases of the clinical development of an enzyme replacement therapy for rare disease called alpha-mannosidosis. In spring 2018 an EU-wide marketing authorisation was granted for the medicinal product Lamzede for the treatment of patients with non-neurological manifestations of mild to moderate alpha-mannosidosis. The holder of the Marketing Authorisation is Chiesi Farmaceutici S.p.A.

More information on funded research and innovation projects is available on [Cordis website](https://cordis.europa.eu).

### Other EU initiatives in the field

**European Reference Networks** (ERN) established under the Directive on patients’ rights in cross-border healthcare aim to tackle complex or rare diseases and conditions that require highly specialised treatment and a concentration of knowledge and resources. ERNs offer the potential to give patients and doctors across Europe access to the best expertise and timely exchange of life-saving knowledge, without having to travel to another country. [EU protects](https://europa.eu/eu/protection/) video highlights how the ERN EpiCARE connected experts to treat epilepsy.

**RD-Action** is a Joint Action funded by the EU Health Programme (2015-2018) supporting the development of national and European policies in the field of rare diseases including the [Orphanet](https://www.orphanet.eu), the biggest global repository of information on rare diseases.