New Horizon 2020 projects to develop diagnostic tools and therapies for Rare Diseases

11 new collaborative research and innovation projects kicked off as the result of the Horizon 2020 calls for proposals in 2017 focussing on New therapies for rare diseases (SC1-PM-08-2017) and Diagnostic characterisation of rare diseases (SC1-PM-03-2017).

Over €77 million of EU contribution was committed in total to these projects involving 123 organisations including universities, hospitals, foundations, SMEs and patient organisations across the EU. Many of these projects link with the recently established European Reference Networks.

According to the EU definition, a disease is considered rare when it affects 1 or less people in every 2,000. Rare diseases affect an estimated 30 million people in the EU. A large number of rare diseases lack accurate diagnostic means and most of rare diseases do not have an effective therapy.

The funded projects will address these needs, carrying out research to new develop 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 billion has been committed in collaborative research through the Seventh Framework Programme (FP7) and Horizon 2020 in more than 200 projects related to rare diseases.

Overview of the new projects

Project acronym and aim	EU funding	Coordinating organisation
CARAMBA Aim: working on SLAMF7-CAR T cells prepared by Sleeping Beauty gene-transfer for immunotherapy of multiple myeloma, a rare hematologic disease	6,097,875.00 €	UNIVERSITAETSKLINIKUM WUERZBURG - KLINIKUM DER BAYERISCHEN JULIUS-MAXIMILIANSUNIVERSITAT, Germany
CLINGLIO	6,155,125.00 €	LIPOPHARMA THERAPEUTICS SL, Spain

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Aim: conducting a clinical phase IIB trial with Minerval in patients with newly-diagnosed malignant glioma		
CureCN Aim: developing adeno-associated virus vector-mediated Liver Gene Therapy for Crigler-Najjar Syndrome	6,249,103.75 €	ASSOCIATION GENETHON, France
HIT-CF Aim: bringing personalised disease modifying therapies to cystic fibrosis patients with ultra-rare CFTR (Cystic Fibrosis Transmembrane Conductance Regulator) mutations, who could otherwise never get access to such treatment	8,753,615.00 €	UNIVERSITAIR MEDISCH CENTRUM UTRECHT, Netherlands
MCDS-Therapy Aim: repurposing of carbamazepine for treatment of skeletal dysplasia	5,697,390.50 €	UNIVERSITY OF NEWCASTLE UPON TYNE, United Kingdom
OligoGpivotalCF Aim: conducting a pivotal phase IIb clinical trial of inhaled alginate oligosaccharide (OligoG) for cystic fibrosis	6,013,748.75 €	ALGIPHARMA AS, Norway
RECOMB Aim: developing stem-cell based gene therapy for recombination deficient SCID (RECOMB)	5,990,460.00 €	ACADEMISCH ZIEKENHUIS LEIDEN, Netherlands
Solve-RD Aim: diagnose a large numbers of rare diseases for which a molecular cause is not known yet by sophisticated combined Omics approaches	15,361,621.25 €	EBERHARD KARLS UNIVERSITAET TUEBINGEN, Germany

TRACE Aim: 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	6,000,000.00 €	LUDWIG-MAXIMILIANS-UNIVERSITAET MUENCHEN, Germany
TUDCA-ALS Aim: working on safety and efficacy of tauroursodeoxycholic acid (TUDCA) as add-on treatment in patients affected by amyotrophic lateral sclerosis	5,596,928.00 €	HUMANITAS MIRASOLE SPA, Italy
UshTher Aim: conducting clinical trial of gene therapy with dual Adeno-Associated virus (AAV) vectors for retinitis pigmentosa in patients with Usher syndrome type IB	5,998,515.75 €	FONDAZIONE TELETHON, Italy

More information on funded projects is available on **Cordis**.