ERNs and Research

– State of play from the European Commission perspective

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European Commission

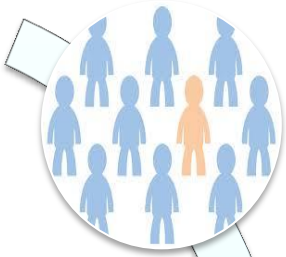
Research and Innovation

EU funded rare diseases research

Research priorities

Activities

Rare diseases



R&I funding

Over € 1 billion in more than 200 projects in FP7 and H2020 on: pathophysiology, natural history, delivered new diagnostics and therapies

Linking major EU and national initiatives

E-RARE: research funders collaboration: more than € 90 million in more than 100 projects
www.erare.eu

International coordination: IRDIRC

IRDIRC: Updated goals for 2027: diagnosis within 1 year, 1000 new therapies, methodologies to measure impact on patients: >50 international partners, policies and guidelines to implement goals
www.irdirc.org

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Highlights of projects involving ERN participants

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SolveRD

"Solving the unsolved rare diseases"

- To solve large numbers of rare diseases by sophisticated combined omics- approaches
- Pooling and re-analysis of 19.000 cases, WGS for 2000, multi-omics, matchmaking and functional analyses, clinical utility and cost-effectiveness aspects
- Coordinator Eberhard Karls Universitaet Tuebingen + 20 partners
- ERN-RND, ERN-ITHACA, ERN-Euro-NMD and ERN-GENTURIS form core ERNs, will reach out to all ERNs

www.solve-rd.eu

Diagnostic characterisation of rare diseases (SC1-PM-03–2017)

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ImmunAid

"Immunome project consortium for AutoInflammatory Disorders"

- Comprehensive -omics studies in Systemic auto-inflammatory diseases (SAID)
- Include more than 700 individuals (patient with monogenic SAID with undiagnosed SAID and healthy controls)
- Coordinator INSERM U932 “Immunity and cancer” in Curie Institute, Paris + 23 partners
- Several recruiting centres are member of the ERN Rare immunological and auto-inflammatory diseases (RITA)

Diagnostic characterisation of rare diseases (SC1-PM-03–2017)

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New therapies for rare diseases (SC1-PM-08–2017)

Project name and aim	Coordinating organisation	Link to ERN
CureCN: Developing adeno-associated virus vector-mediated Liver Gene Therapy for Crigler-Najjar Syndrome	ASSOCIATION GENETHON, FR	Metab ERN
RECOMB: Developing stem-cell based gene therapy for recombination deficient SCID	ACADEMISCH ZIEKENHUIS LEIDEN, NL	ERN Rita
TRACE: Adoptive T-cell transfer for treatment for refractory viral infection that occurs following allogeneic stem cell transplantation	LUDWIG-MAXIMILIANS-UNIVERSITAET MUENCHEN, DE	Eurobloodnet ERN PaedCan
UshTher: Conducting clinical trial of gene therapy with dual Adeno-Associated virus (AAV) vectors for retinitis pigmentosa in patients with Usher syndrome type IB	FONDAZIONE TELETHON, IT	ERN EYE

New therapies for rare diseases (SC1-PM-08–2017)

Project name and aim	Coordinating organisation	Link to ERN
HIT-CF: Bringing personalised disease modifying therapies to cystic fibrosis patients with ultra-rare CFTR mutations	UNIVERSITAIR MEDISCH CENTRUM UTRECHT, NL	ERN LUNG
MCDS-Therapy: Repurposing of carbamazepine for treatment of skeletal dysplasia	UNIVERSITY OF NEWCASTLE UPON TYNE, UK	ERN BOND
OligoGpivotalCF: Pivotal phase IIb clinical trial of inhaled alginate oligosaccharide (OligoG) for cystic fibrosis	ALGIPHARMA AS, NO	ERN LUNG
TUDCA-ALS: Safety and efficacy of tauroursodeoxycholic acid (TUDCA) as add-on treatment	HUMANITAS MIRASOLE SPA, IT	ERN-RND

More information available on Cordis Project Search: http://cordis.europa.eu/projects/home_en.html

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Topics referring to ERNs

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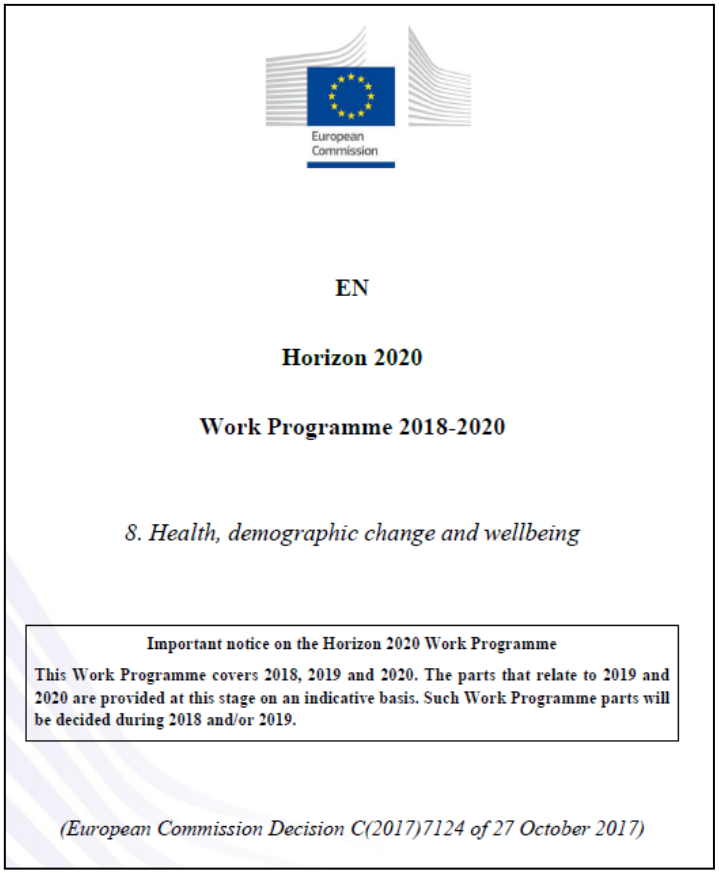
Horizon 2020 SC1 Work Programme 2018-2020*



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Horizon 2020

Work Programme 2018-2020

8. Health, demographic change and wellbeing

Important notice on the Horizon 2020 Work Programme

This Work Programme covers 2018, 2019 and 2020. The parts that relate to 2019 and 2020 are provided at this stage on an indicative basis. Such Work Programme parts will be decided during 2018 and/or 2019.

(European Commission Decision C(2017)7124 of 27 October 2017)

Rare Disease European Joint Programme Cofund SC1-BHC-04-2018

HTA research to support evidence-based healthcare SC1-BHC-26-2018

Innovation Procurement: Next generation sequencing (NGS) for routine diagnosis SC1-BHC-10-2019

+ see also other topics

*** parts related to 2019 and 2020 are indicative**

http://ec.europa.eu/research/participants/data/ref/h2020/wp/2018-2020/main/h2020-wp1820-health_en.pdf

Thank you

http://ec.europa.eu/programmes/horizon2020/

http://ec.europa.eu/research/health/index.cfm?pg=home

Funding opportunities:

http://ec.europa.eu/research/participants/portal/desktop/en/home.html