

# E-Rare-3 Call for Proposals 2018 for "Transnational research projects on hypothesis- driven use of multi-omic integrated approaches for discovery of disease causes and/or functional validation in the context of rare diseases"

---

## Preliminary Announcement

---

The ERA-Net "E-Rare" for research programmes on rare diseases has been extended to a third phase "E-Rare-3" (2014-2019) to further help in coordinating the research efforts of European countries in the field of rare diseases and implement the objectives of International Rare Disease Research Consortium (IRDiRC).

The following parties,

- Austrian Science Fund (FWF), Austria\*
- Research Foundation Flanders (FWO), Belgium, Flanders
- Fund for Scientific Research - FNRS (F.R.S.-FNRS), Belgium, French-speaking community
- Canadian Institutes of Health Research – Institute of Genetics (CIHR-IG), Canada
- Fonds de recherche du Québec-Santé (FRQS), Québec (Canada)
- Ministry of Education, Youth and Sports (MEYS), Czech Republic
- Academy of Finland (AKA), Finland
- French National Research Agency (ANR), France
- Federal Ministry of Education and Research (BMBF), Germany
- German Research Foundation (DFG), Germany
- General Secretariat for Research and Technology (GSRT), Greece
- National Research, Development and Innovation Office (NKFIH), Hungary
- Chief Scientist Office of the Ministry of Health (CSO-MOH), Israel
- Italian Ministry of Health (MoH-IT), Italy
- State Education Development Agency (VIAA), Latvia
- National Centre for Research and Development (NCBiR), Poland
- Executive Agency for Higher Education, Research, Development and Innovation Funding (UEFISCDI), Romania\*
- National Institute of Health Carlos III (ISCIII), Spain\*
- Swiss National Science Foundation (SNSF), Switzerland
- Netherlands Organization for Health Research and Development (ZonMw), The Netherlands
- The Scientific and Technological Research Council of Turkey (TUBITAK), Turkey

\*. *Decision still pending*

intend to open the tenth E-Rare joint call for funding multilateral research projects on rare diseases. The call is expected to be opened simultaneously by the parties in their respective countries.

## 1. AIM OF THE CALL

The aim of the call is to enable scientists in different countries to build an effective collaboration on a common interdisciplinary research project based on complementarities and sharing of expertise, with a clear translational research approach. Projects shall involve a group of rare diseases or a single rare disease following the European definition i.e. a disease affecting not more than five in 10 000 persons in the European Community, EC associated states and Canada.

## TOPICS OF THE CALL

**The research projects have to focus on hypothesis-driven use of multi-omic integrated approaches for discovery of disease causes and/or functional validation in the context of rare diseases.**

Transnational research proposals must cover at least one of the following areas, which are equal in relevance for this call:

- a. Combined multi-omics approaches (e.g. epigenomics, transcriptomics, metabolomics, proteomics, etc.) that complement genomics-based gene discovery strategies and that are driven by a lead hypothesis. These multi-omics approaches should extend beyond descriptive “-omics” data gathering, such as simple whole exome/genome sequencing for disease gene discovery. For transcriptomic and proteomic data, a strong rationale for physiological relevance of the collected sample/tissue/dataset must be available;
- b. Functional validation of clinical or biological inferences obtained from “-omics” results, e.g. by
  - developing new computational, statistical and experimental methods for analysis and interpretation of existing multi-omic datasets or for the identification of relevant biomarkers;
  - integrating the already obtained “-omics” results to generate and test new biological models;
- c. Application of “-omics” approaches to rare diseases for which the gene(s) is/are known to enable insight into disease pathophysiology. Emphasis will be given to approaches that transcend a single “-omics” approach to illuminate pathomechanism. Projects that generate “-omics” data with limited integration and interpretation will be considered lower priority;
- d. Development and application of concepts and methods for pathogenic read-outs of disease groups which can be used as “blue print” to discover new disease genes and inform pathomechanism. Projects on “simple” or “pure” gene hunts will be discouraged if they can be rationally performed at a single institution or by existing

international resource centers, with the exception of studies that inform fundamentally new genetic paradigms.

Furthermore, additional elements must be taken care of in the application:

- Proposed projects should rest on an excellent lead hypothesis for the intended activities;
- Proposed projects that focus only on data sets from genomic approaches (e.g. exome/genome sequencing of a cohort) will have low priority;
- Proposed projects *have to* show multi-dimensional approaches and strong knowledge of interpretation of such data, ideally combining rigorous statistic methods with biological/experimental verification;
- A core set of “-omics” results should already be present and serve as a justification to perform other “-omics” experiments;
- The design of the study (sample collection, statistical power, interpretation, relevant models for hypothesis validation) must be well justified and should be part of the proposal;
- Appropriate bioinformatics and statistical skills should constitute, whenever justified, an integral part of the proposal;
- The new research data resulting from the project should be treated permissible according to the FAIR<sup>1</sup> principles, and deposited and shared, according to the national rules of the countries involved. It is strongly advised to make data accessible through RD-Connect (<http://rd-connect.eu/> - connecting databases, patient registries, biobanks and clinical bioinformatics data into a central resource for researchers worldwide) and through Elixir (<https://www.elixir-europe.org/platforms/data/elixir-deposition-databases> - compiling a list of resources for the deposition of experimental, biomolecular data). To make research data findable, accessible, interoperable and re-usable (FAIR), a data management strategy for the proposed full project is mandatory in the full proposal stage. Some countries involved in E-Rare JTC 2018 will also ask for a data management plan (DMP) at national level at the stage of full proposal or after granting of the project.

**The following approaches and topics are excluded from the scope of the call:**

- a. Approaches concerning rare infectious diseases or rare cancers;
- b. Approaches concerning rare adverse drug events/medical complications in treatments of common diseases;
- c. Interventional clinical trials.

Project proposals must clearly demonstrate the potential health impact as well as the added-value of transnational collaboration: gathering a critical mass of patients/biological material, sharing of resources (models, databases, diagnosis etc.), harmonization of data, sharing of specific know-how and/or innovative technologies, etc.

Each transnational collaborative project should represent the critical mass to achieve ambitious scientific goals. Consortia are encouraged to demonstrate engagement with industry for its active participation including areas of collaboration, sharing of resources, capabilities and expertise, in order to ensure an efficient transfer of pre-clinical results into clinical utility. Likewise, patient organizations are invited to participate where appropriate as their engagement has the potential to provide new insights that could lead to innovative discoveries, and ensures that research is relevant to patients' concerns.

---

<sup>1</sup> FAIR: Findable, Accessible, Interoperable, Reusable (for more information: see “The FAIR Guiding Principles for scientific data management and stewardship” (<https://www.nature.com/articles/sdata201618>))

It is expected that the inclusion of partner groups from participating Eastern European countries will contribute to strengthening the research capacity building in Europe as a whole.

## 2. GENERAL CONDITIONS FOR APPLICATION

Joint research proposals may be submitted by higher education institutions, non-university public research establishments, hospitals as well as commercial companies, in particular small and medium-size enterprises (SMEs), according to relevant national funding organisations' regulations for research funding.

Only transnational projects will be funded. Each consortium submitting a proposal must involve a **minimum of three eligible** and a **maximum of six eligible partners from at least three different countries** participating to the call (see list of parties/countries above). Not more than two eligible partners from the same country participating in the call will be accepted in one consortium.

Applicants are encouraged to **include partners from the participating Eastern European countries** (Czech Republic, Hungary, Latvia, Poland, Romania, Turkey). If they include such partners, the maximum number of partners can be increased to **eight** (see table below).

Additional partners that secure their **own funding** may join consortia. However, their number is **limited to two**. The consortium coordinator must always be eligible to receive funding from the funding organisations participating in the call. Only groups that contribute substantially to at least one of the work packages are considered as partners. They must state clearly in the proposal if these funds are already secured or if not, how they plan to obtain funding in advance of the project start. It will be required to document the availability of their funds before October 1, 2018.

Number of partners requesting funding	Possible number of additional partners with own funding
3	2
4	
5	
6	
7 (only possible with inclusion of Eastern European partner)	1
8 (only possible with inclusion of Eastern European partner)	0

<sup>1</sup>Whilst applications will be **submitted jointly** by groups from several countries, individual groups will be funded by the individual E-Rare-3 funding organisation respective of the country from which applicants have applied. The applications are therefore subjected to **eligibility criteria of individual funding organisations**.

**Applicants are strongly advised to contact their national representative and confirm eligibility with their respective funding organisations in advance of submitting an application**

### 3. TIMETABLE

There will be a **two-stage submission procedure** for joint applications: pre-proposals and full proposals. The call is scheduled to open on **December 7, 2017**. The deadline for submitting the pre-proposals is scheduled for **February 6, 2018**. An independent international Scientific Evaluation Committee will carry out a scientific evaluation according to specific evaluation criteria. Based on this central evaluation, selected consortia will be invited to submit a full proposal by **mid May 2018** (deadline for full proposals: **June 19, 2018**).

For further information, please visit us on the website:  
<http://www.e-rare.eu>

For questions regarding the joint call please contact the  
Joint Call Secretariat:

Nika Ritsema  
Email: [ritsema@zonmw.nl](mailto:ritsema@zonmw.nl)  
Phone: +31 (0)70 349 54 85

Harald Moonen  
Email: [moonen@zonmw.nl](mailto:moonen@zonmw.nl)  
Phone: +31 (0) 70 349 53 49

Sonja van Weely  
Email: [weely@zonmw.nl](mailto:weely@zonmw.nl)

For questions regarding national eligibility criteria and requirements  
please contact the  
national contact person listed below

*The content of the call described in this pre-announcement may be subject to changes and is not legally binding to the funding organisations.*

## National/regional contact details

Country/Region	Institution	Website	National/regional contact
Austria	FWF	<a href="http://www.fwf.ac.at">www.fwf.ac.at</a>	Stephanie Resch Phone: +43 (1) 505 67 40-8201 Email: <a href="mailto:stephanie.resch@fwf.ac.at">stephanie.resch@fwf.ac.at</a>  Anita Stürtz Phone: +43 (1) 505 67 40-8206 E-mail: <a href="mailto:anita.stuertz@fwf.ac.at">anita.stuertz@fwf.ac.at</a>
Belgium/Flanders	FWO	<a href="http://www.fwo.be">www.fwo.be</a>	Alain Deleener Phone: +32 2 550 15 95 Email: <a href="mailto:eragnet@fwo.be">eragnet@fwo.be</a>  Toon Monbaliu Phone: +32 2 550 15 70 Email: <a href="mailto:eragnet@fwo.be">eragnet@fwo.be</a>
Belgium/French speaking community	FNRS	<a href="http://www.frs-fnrs.be/">www.frs-fnrs.be/</a>	Florence Quist Phone: +32 2 504 93 51 Email: <a href="mailto:florence.quist@frs-fnrs.be">florence.quist@frs-fnrs.be</a>  Joël Groeneveld Phone: +32 2 504 92 70 E-mail: <a href="mailto:joel.groeneveld@frs-fnrs.be">joel.groeneveld@frs-fnrs.be</a>
Canada	CIHR-IG	<a href="http://www.cihr-irsc.gc.ca">www.cihr-irsc.gc.ca</a>	Adrian Puga Phone: +1 613 952 5728 Email: <a href="mailto:adrian.puga@cihr-irsc.gc.ca">adrian.puga@cihr-irsc.gc.ca</a>
Canada (Québec)	FRQS	<a href="http://www.frqs.gouv.qc.ca">www.frqs.gouv.qc.ca</a>	Fonds de recherche du Québec-Santé (FRQS)  Maxime Beaudoin Phone: +1 514 873 2114, ext 1369 Email: <a href="mailto:maxime.beaudoin@frq.gouv.qc.ca">maxime.beaudoin@frq.gouv.qc.ca</a>  Anne-Cécile Desfaits Phone: +1 514 873 2114, ext 1368 <a href="mailto:annececile.desfaits@frq.gouv.qc.ca">annececile.desfaits@frq.gouv.qc.ca</a>
Czech Republic	MEYS	<a href="http://www.msmt.cz">www.msmt.cz</a>	Ministry of Education Youth and Sports Renata Hrubá Phone: +420 234 811 504 Mobile: +420 770 145 082 E-mail: <a href="mailto:renata.hruby@msmt.cz">renata.hruby@msmt.cz</a>
Finland	AKA	<a href="http://www.aka.fi">www.aka.fi</a>	Heikki Vilen Phone: +358 29 5335 135 Email: <a href="mailto:heikki.vilen@aka.fi">heikki.vilen@aka.fi</a>

Country/Region	Institution	Website	National/regional contact
France	ANR	<a href="http://www.agence-nationale-recherche.fr">www.agence-nationale-recherche.fr</a>	Ingrid Pfeifer Phone: + 33) (0)1 78 09 80 22 Daria Julkowska Phone: + 33) (0)1 78 09 80 78 Email: <a href="mailto:E-RareCalls@agencerecherche.fr">E-RareCalls@agencerecherche.fr</a>  Agence Nationale de la Recherche – ANR Health & Biology Department 50 Avenue Daumesnil 75012 Paris, France
Germany	BMBF/ PT-DLR	<a href="http://www.gesundheitsforschung-g-bmbf.de">www.gesundheitsforschung-g-bmbf.de</a>	Katarzyna Saedler Phone: +49 (0)228 3821 1947 Email: <a href="mailto:Katarzyna.Saedler@dlr.de">Katarzyna.Saedler@dlr.de</a>  Ralph Schuster Phone: +49 (0)228 3821 1233 Email: <a href="mailto:Ralph.Schuster@dlr.de">Ralph.Schuster@dlr.de</a>  Sabrina Legies Phone: +49 (0)228 3821 1212 Email: <a href="mailto:Sabrina.Legies@dlr.de">Sabrina.Legies@dlr.de</a>  Project Management Agency of the German Aerospace Centre (PT-DLR) -Health Research-
Germany	DFG	<a href="http://www.dfg.de">www.dfg.de</a>	Dr. Katja Großmann Email: <a href="mailto:katja.grossmann@dfg.de">katja.grossmann@dfg.de</a> Phone: +49 (0) 228 885 2565 Fax: +49 (0) 228) 885 2777 Kennedyallee 40 53175 Bonn
Greece	GSRT	<a href="http://www.gsrt.gr">www.gsrt.gr</a>	Sofia DIMITROPOULOU Phone: +30 2107 458 187 Email: <a href="mailto:s.dimitropoulou@gsrt.gr">s.dimitropoulou@gsrt.gr</a>  General Secretariat for Research & Technology International S&T Cooperation Directorate Division of Bilateral & Multilateral
Hungary	NKFIH	<a href="http://www.nkfi.gov.hu">www.nkfi.gov.hu</a>	National Research, Development and Innovation Office Department of Research and Development  Előd Nemerkenyi Phone: +36 1 8963987 Email: <a href="mailto:elod.nemerkenyi@nkfi.gov.hu">elod.nemerkenyi@nkfi.gov.hu</a>  Gábor Tóth Phone: +36 1 8961727 Email <a href="mailto:gabor.toth@nkfi.gov.hu">gabor.toth@nkfi.gov.hu</a>

Country/Region	Institution	Website	National/regional contact
Israel	CSO-MOH	<a href="http://www.health.gov.il">www.health.gov.il</a>	Irit Allon Phone: +972-2-5082167 Email: <a href="mailto:Irit.allon@moh.health.gov.il">Irit.allon@moh.health.gov.il</a>
Italy	MoH-It	<a href="http://www.ministerosalute.it/">www.ministerosalute.it/</a>	Dr. Giselda Scalera Phone: +39 065994 2596 Email: <a href="mailto:g.scalera@sanita.it">g.scalera@sanita.it</a> research.EU.dgric@sanita.it  Head Office 5 (Health Research IRCCS), Directorate General for Research and Innovation I Healthcare Ministry of Health, Viale Giorgio Ribotta, 5 -00144 Rome, Italy
Latvia	VIAA	<a href="http://www.viaa.gov.lv">www.viaa.gov.lv</a>	Dr Maija Bundule Phone: +371- 67785423 E-Mail: <a href="mailto:Maija.Bundule@viaa.gov.lv">Maija.Bundule@viaa.gov.lv</a>  Dr Uldis Berkis Phone: +371 67785487; +371 29472349 Email: <a href="mailto:Uldis.Berkis@viaa.gov.lv">Uldis.Berkis@viaa.gov.lv</a>
Poland	NCBiR	<a href="http://www.en.ncbir.pl/">www.en.ncbir.pl/</a>	Marcin Chmielewski Section for Research Projects BIOMED, Nowogrodzka Str. 47a, 00-695 Warsaw, Poland, Phone: +48 22 39 07 109 Email: <a href="mailto:marcin.chmielewski@ncbr.gov.pl">marcin.chmielewski@ncbr.gov.pl</a>
Romania	UEFISCDI	<a href="http://uefiscdi.gov.ro/">http://uefiscdi.gov.ro/</a>	Executive Agency for Higher Education, Research, Development and Innovation Funding  Mihaela Manole Phone: +40213023863 Email: <a href="mailto:mihaela.manole@uefiscdi.ro">mihaela.manole@uefiscdi.ro</a>  Nicoleta Dumitrache Phone: +40213023886 Email: <a href="mailto:nicoleta.dumitrache@uefiscdi.ro">nicoleta.dumitrache@uefiscdi.ro</a>
Spain	ISCIII	<a href="http://www.isciii.es">www.isciii.es</a>	María Druet SG de Programas Internacionales de Investigación Phone: +34 9182 22530 E-mail: <a href="mailto:mdruet@isciii.es">mdruet@isciii.es</a>
Switzerland	SNSF	<a href="http://www.snf.ch">www.snf.ch</a>	Raphael Banz Division of Biology and Medicine Swiss National Science Foundation Phone: +41 31 308 21 82 Email: <a href="mailto:raphael.banz@snf.ch">raphael.banz@snf.ch</a>

Country/Region	Institution	Website	National/regional contact
The Netherlands	ZonMw	<a href="http://www.zonmw.nl">www.zonmw.nl</a>	<p>Nika Ritsema Phone: +31 (0)70 349 54 85 Email: <a href="mailto:ritsema@zonmw.nl">ritsema@zonmw.nl</a></p> <p>Harald Moonen Phone: +31-(0)70 349 53 49 Email: <a href="mailto:moonen@zonmw.nl">moonen@zonmw.nl</a></p> <p>Sonja van Weely Email: <a href="mailto:weely@zonmw.nl">weely@zonmw.nl</a></p> <p>The Netherlands Organization for Health Research and Development (ZonMw)</p>
Turkey	TUBITAK	<a href="http://www.tubitak.gov.tr">www.tubitak.gov.tr</a>	<p>Jale Şahin Phone: +90- 312- 298 17 96 Email: <a href="mailto:e-rare@tubitak.gov.tr">e-rare@tubitak.gov.tr</a></p> <p>The Scientific and Technological Research Council of Turkey (TUBITAK) International Cooperation Department Division of Bilateral and Multilateral Relations</p>