

# THE COORDINATION OF RARE DISEASES RESEARCH IN EUROPE:

## THE EUROPEAN JOINT PROGRAMME ON RARE DISEASES

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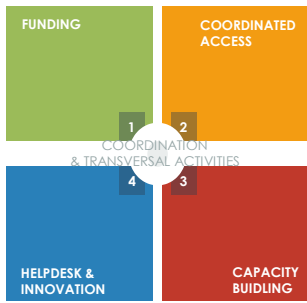


TEDDY NETWORK GENERAL ASSEMBLY  
ROME, 14 OF JANUARY 2018

# EUROPEAN JOINT PROGRAMME ON RARE DISEASES

## Background of preparatory work

- **March 2016** - First discussions started at E-Rare strategic workshop in Barcelona
- **August 2016** - The Commission sends official invitation to MS to nominate experts to the EJP RD working groups
- **October 2016** - First meeting of the MS EJP RD expert group
- **Dec 2016 – Feb 2017** - Preparation of the 1<sup>st</sup> Concept Draft of the EJP RD
- **April 2017** - 2<sup>nd</sup> meeting of the MS EJP RD experts group
- **Apr – June 2017** – nomination of new experts to future WGs
- **June 2017** – kick off meeting of the extended WGs of the EJP RD
- **June – Oct 2017** – preparation of the 2<sup>nd</sup> Concept Draft of the EJP RD → more than 70 experts involved + 24 ERN coordinators representing EU, associated and non-EU countries
- **12 Oct 2017** – 2<sup>nd</sup> EJP RD Concept Draft sent to the EC and WGs + ERN Coos
- **17 Oct 2017** – last meeting of the MS
- **End Oct 2017** – official publication of the WP 2018 – 2020 → kick off of the writing phase
- **Oct – Nov 2017** – call for Letters of Interest
- **11 Dec 2017** - Kick off of preparatory phase of EJP RD
- **31 Dec 2017** – final identification of contributors
- **30 of March 2018** – submission of the project

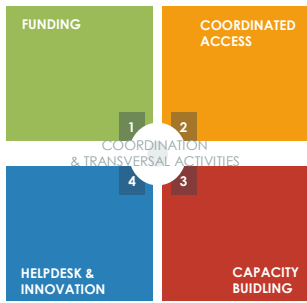


# EUROPEAN JOINT PROGRAMME ON RARE DISEASES

## OBJECTIVES

- **Main objective:** Create a research and innovation pipeline "from bench to bedside" ensuring rapid translation of research results into clinical applications and uptake in healthcare for the benefit of patients
- **Specific objective:** improve integration, efficacy, production and social impact of research on rare diseases through the development, demonstration and promotion of sharing of research and clinical data, materials, processes, knowledge and know-how, and an efficient model of financial support for research on rare diseases

- **Union contribution:** 55 M€
- **Total budget (min):** 78,5 M€ (→ expected > 110 M€)
- **Foreseen number of partners:** 70 – 100
- **Foreseen number of participating countries:** 29 including 23 EU MS, 4 associated (CH, IL, NO, TK) and 4 third countries (AU, CA, JP, USA)



# EJP RD EXPECTED IMPACT

- **Improve lives of rare disease patients by providing new and optimised treatment options and diagnostic tools for these diseases**
- **Decrease fragmentation of rare diseases expertise and research resources**
- **Increase the EU's capacity to innovate in the field of rare diseases**
- **Improve healthcare systems' capacity to take up research results**
- **Reinforce the EU's role as a global leader for rare diseases**

# RESULTS OF THE CALL FOR LETTERS OF INTENT

## 112 LOI received representing:

- 25 countries (AT, AU, BE, CA, CH, CZ, DE, DK, EE, ES, FR, HU, GR, IE, IL, IT, LT, LV, NL, PL, PT, SE, TK, UK, USA)
- 24 ERNs
- 31 « international » organisations (infrastructures, EURORDIS) + 3 charities

## Share of interest between pillars:

- P1: 27 institutions from 20 countries (AT, BE, CA, CH, CZ, DE, EE, ES, FR, HU, GR, IE, IL, IT, LT, LT, PL, PT, SE, TK) + EURORDIS + charities
- P2: 71 institutions from 18 countries (AT, AU, BE, CH, DE, DK, EE, ES, FR, HU, IT, LV, NL, PL, PT, SE, TK, UK) + ERNs + international (infrastructures)
- P3: 51 institutions from 18 countries (AT, AU, CH, CZ, DE, EE, ES, FR, HU, IE, IT, LT, LV, NL, PL, PT, TK, USA) + international (EURORDIS; infrastructures) + ERNs
- P4: 32 institutions from 10 countries (BE, CH, ES, FR, IT, NL, TK, USA) + ERNs + charities + international (EURORDIS; infrastructures)

# EJP RD STRUCTURE

COORDINATION  
& TRANSVERSAL ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

COMMUNICATION

SUSTAINABILITY

HELPDESK OFFICE – FAST TRACK TO SUPPORT & INFORMATION

1

FUNDING

2

COORDINATED  
ACCESS

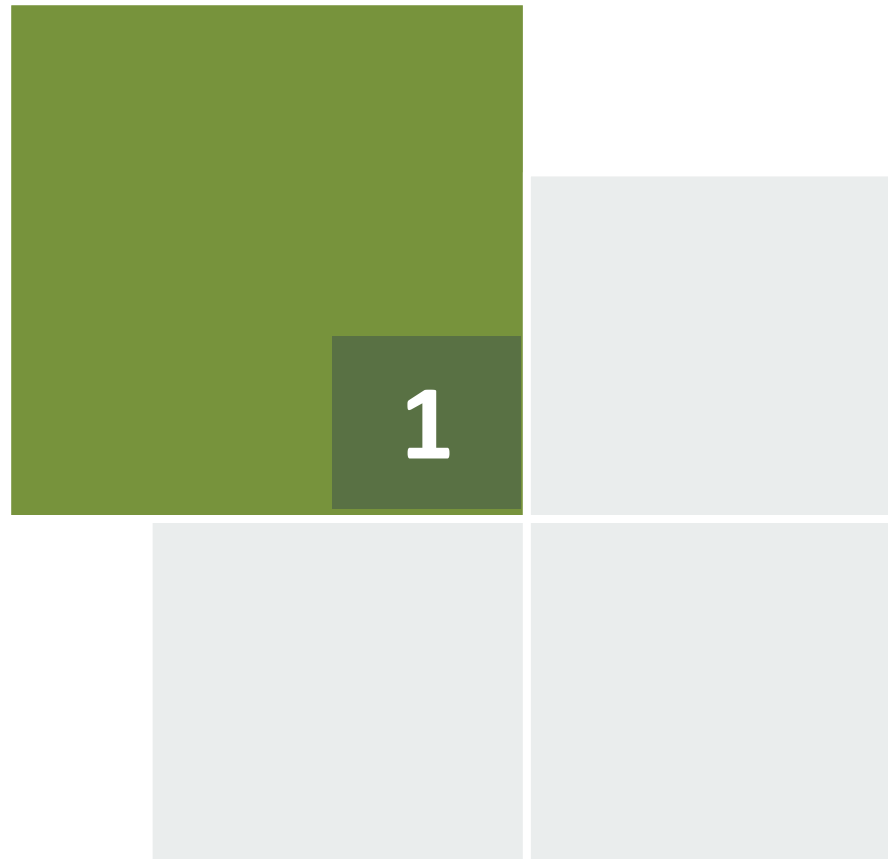
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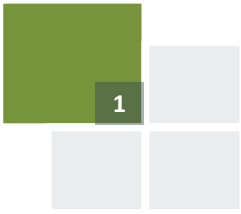
CAPACITY  
BUILDING

4

INNOVATION

# PILLAR 1: RESEARCH COLLABORATIVE FUNDING



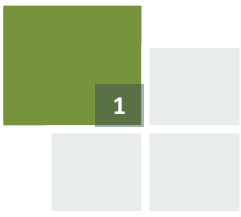


# BACKGROUND & JUSTIFICATION

- **The need for funding of transnational cooperative research on Rare Diseases (RD) for reducing fragmentation of research and finding synergies across different actors in this field is evident**
- **Only few European countries fund research on RD through specific dedicated programs**
- **Complementary to EC funding, the E-Rare funding program has successfully implemented funding for transnational research projects for RD since 2007**

**Pillar 1 Chairs:**  
**Ralph Schuster (DLR, DE)**  
**Sonja van Weely (ZonMw, NL)**





# PROPOSED STRUCTURE

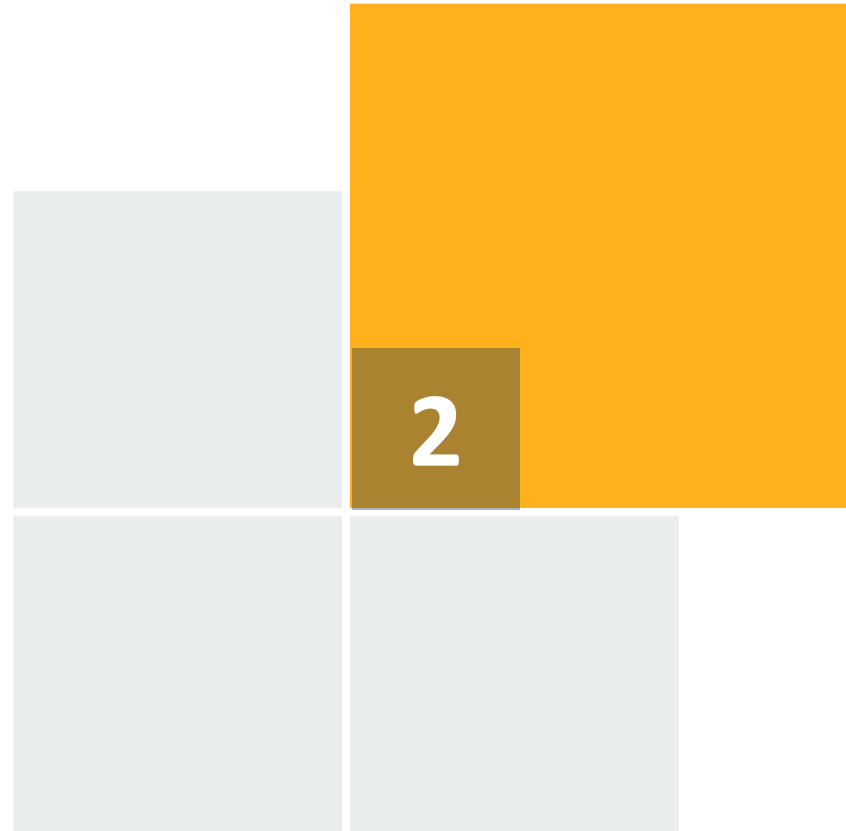
**WP1: Joint Transnational Calls for collaborative research projects**

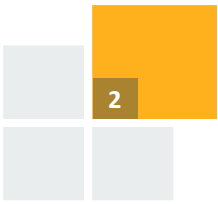
**WP2: Networking to share knowledge on rare diseases**

**WP3: Rare disease research challenges**

**WP4: Monitoring of granted projects**

# PILLAR 2: COORDINATED ACCESS TO DATA AND SERVICES





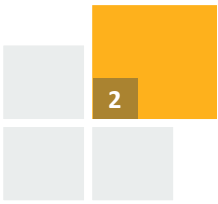
# BACKGROUND & JUSTIFICATION

- Existing fragmentation of efforts, data and resources, too many redundancies between teams and projects
- Difficulties to re-use results of funded research that is often lost because of lack of sustainability or of anticipation
- Existing inefficiency and inefficacy into the whole research pipeline
- Need for optimisation of tools and resources already existing due to lack of awareness or lack of access
- Need to achieve the ambitious IRDiRC diagnosis & therapies goals

**Pillar 2 Chairs:**

**Ana Rath (INSERM, FR)**

**Olaf Riess (Univ Tübingen, DE)**



# PROPOSED STRUCTURE

**WP1: Next-generation platform for data and resources for RD research**

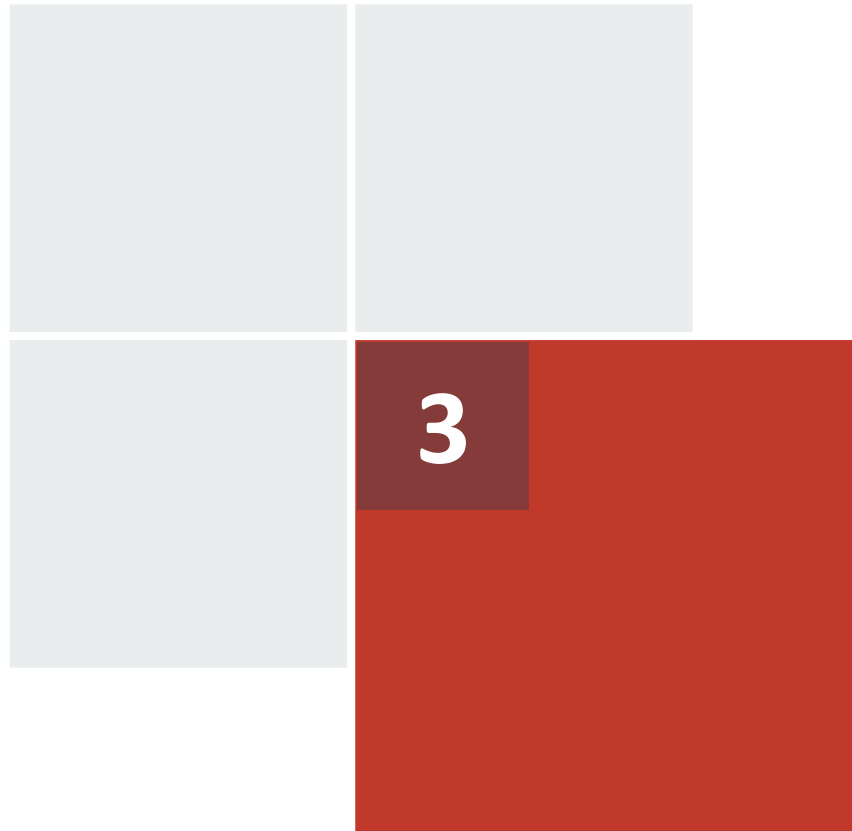
**WP2: Development and unification of infrastructures, technologies, data and standards to create a FAIR and federated next-generation information infrastructure for rare disease**

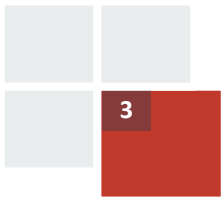
**WP3: Developing innovative approaches of the transcribed and translated genome**

**WP4: Innovative strategies for development of therapies for RD**

**WP5: Innovative small funding service interoperable with an international brokerage system**

# PILLAR 3: CAPACITY BUILDING AND EMPOWERMENT





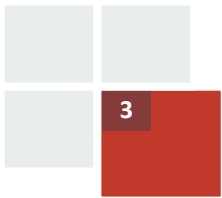
# BACKGROUND AND JUSTIFICATION

- **Patient-centered approach**
- **Key success factor for patient research partnerships is building the capability of researchers and patients to engage meaningfully with each other, maintain transparency and build trust**
- **Empowering stakeholders to contribute proactively and help inform research objectives**
- **Raising the level of knowledge and build capacity within the RD research and care community is critical to ensure equity and optimising outcomes for people living with rare diseases**

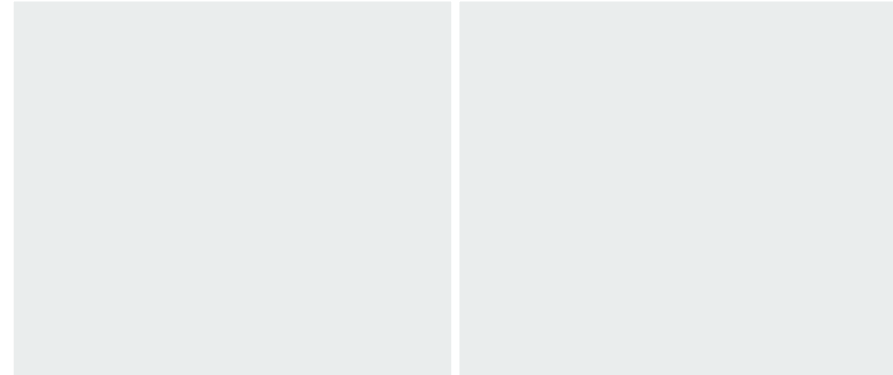
**Pillar 3 Chairs:**

**Virginie Bros-Facer (EURORDIS)**

**Birute Tumiene (Univ Vilnius, LT)**



# PROPOSED STRUCTURE



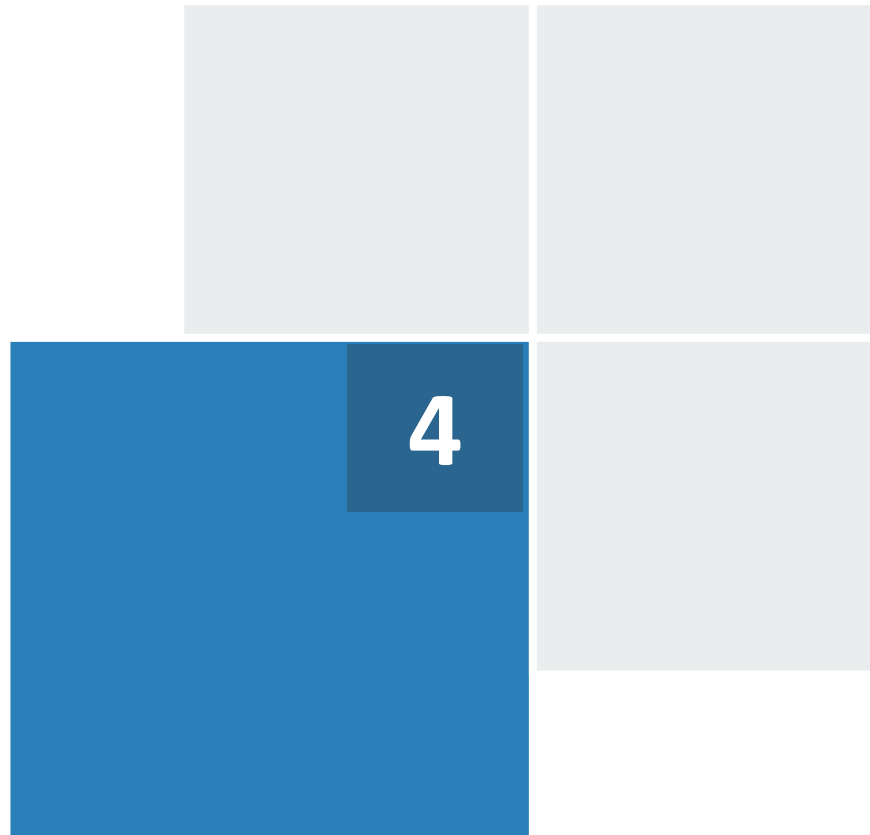
**WP1: Training on data management & quality**

**WP2: Capacity building & training of patients and researchers in RD research and processes**

**WP3: Online academic education courses**

**WP4: ERN RD training & support programmes**

# PILLAR 4: INNOVATION





# BACKGROUND AND JUSTIFICATION

- **The resources and expertise exists but is hard to reach, underused and still scattered**
- **Patient-driven research is often undertaken by small academic groups with limited possibilities of translation and funding resources**
- **Lack of structured and continuous support leading to the uptake of final results**
- **Fragmenting of cohorts between national/regional existing registries, biobanks and data resources with limited bridging possibilities**
- **Newly created ERNs are major resources for a proper and accelerated translation from bench to bedside of clinical programs**

**Pillar 4 Chairs:**

**Rima Nabbout (Imagine, FR)**

**Maurizio Scarpa (HSK, DE)**

# PROPOSED STRUCTURE

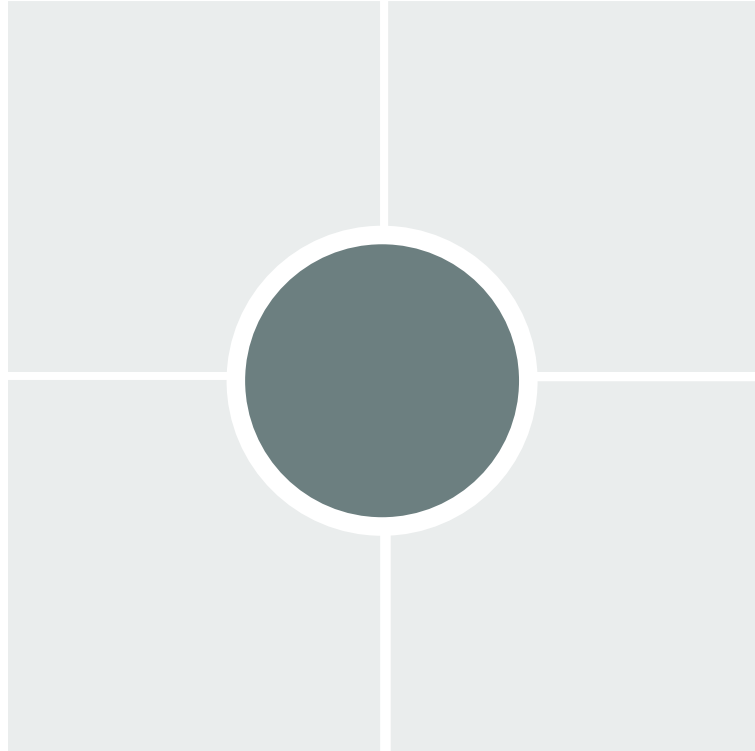
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**WP1: Support to accelerate translational research**

**WP2: Support to design of clinical trials**

**WP3: Public-public & public-private partnerships**

# TRANSVERSAL ACTIVITIES





# TRANSVERSAL ACTIVITIES

**STRATEGY**

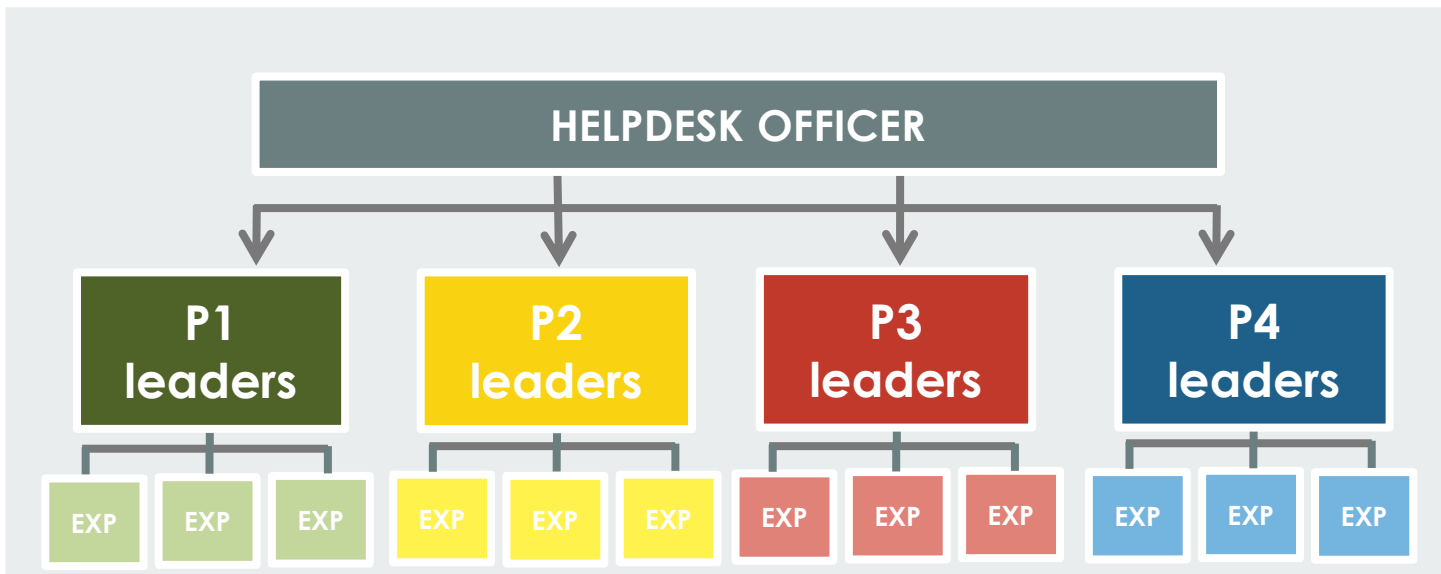
**SUSTAINABILITY**

**COMMUNICATION & DISSEMINATION**

**HELPDESK**

## HELPDESK IS A FAST TRACK TO INFORMATION & SUPPORT

- A single entry point for the whole EJP RD
- It's role is to respond to questions/requests from the RD community in relation to the activities of EJP RD
- **Based on the competences & experts of each of the pillars**
- It is NOT replacing already existing helpdesks or supports of other organizations BUT facilitates the flow of information and provides response in a most rapid, structured and complete manner



# E-RARE JOINT TRANSNATIONAL CALL 2018

**TOPIC: 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**

- With participation of 18 countries: AT, BE, CA, CZ, CH, DE, ES, FI, FR, GR, HU, IL, IT, LV, NL, PL, RO, TK
- Tentative budget above 18 M€
- Call opened on 7 December 2017 → pre-proposal submission deadline **6 of Feb 2018**
- Focus on collaborative projects on rare diseases (min. 3 teams from 3 different countries)
- Excluded: rare cancers, rare infectious diseases, rare adverse drug events/medical complications in treatments of common diseases & interventional clinical trials
- MORE INFO AT: **[www.erare.eu](http://www.erare.eu)**



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# THANK YOU

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