E-RARE & THE EUROPEAN JOINT PROGRAMME ON RARE DISEASES

RE(ACT) 2018 Congress
7-10 of March 2018
Bologna, ITALY
E-RARE –
THE ERA-NET FOR RESEARCH PROGRAMMES ON RARE DISEASES
MAJOR FACTS ABOUT E-RARE

• **ERA-NET** for research programs on rare diseases
• Created in 2006 (E-Rare-1) renewed in 2010 (E-Rare-2) at present funded under Horizon 2020 ERA-Net Co-fund
• **E-Rare-3 (2014 - 2019)**

• 28 partners from 20 European, Associated and non-European countries (AT, BE, CA, CZ, CH, ES, FR, GR, HU, IT, IL, JP, LT, LV, NL, PO, PL, RO, TR)

• **Objective**: coordinate and promote European and international efforts for funding research on rare diseases
• E-Rare is mentioned in more than half of National Plans for Rare Diseases
Medical domains represented in the funded projects:

- Neurology
- Metabolic diseases
- Dermatology
- Others
- Musculoskeletal Diseases
- Rheumatology
- Pulmonary/Respiratory Diseases
- Hematology/Immunology
- Nephrology/Urology
- Cardiology/Vascular Diseases
- Ophthalmology
- Dysmorphology
- Endocrinology
- Psychiatry/Psychology

9 Joint Transnational Calls:

117 funded projects

104 M€ invested

Including 556 research partners funded
E-Rare facilitates & connects

**RESEARCH COMMUNITY**

**PATIENT ORGANIZATIONS**
- Collaboration with small & big POs/Co-funding/Research projects

**EU RESEARCH INFRASTRUCTURES**
- Direct collaboration/Calls/Info platform

**OTHER STAKEHOLDERS**
- Training & workshops (EMA, ESHG)

**NETWORKING**
- Partnership with RE(Act) Congress
631 publications

More than 251* students trained

+ 1 M€ investment by POs

100% achieve critical mass

54% successful application to other calls

100* new genes discovered

77% new sustainable collaborations

104 M€

556 teams

117 research projects

77% new sustainable collaborations

* In 48 finalized projects
EJP RD –
THE EUROPEAN JOINT PROGRAMME
ON RARE DISEASES
RARE DISEASES LANDSCAPE IN EUROPE

- STRATEGY
- INFRA STRUCTURES
- RARE DISEASES RESEARCH
- FUNDING
- HEALTH CARE
- PATIENTS NEEDS

- E-Rare
- IRDiRC
- Orphanet
- BBMRI-ERIC
- ECRIN
- Eatris
- Elixir
- European Reference Networks
- EURORDIS
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 1st Concept Draft of the EJP RD
- April 2017 - 2nd 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 2nd Concept Draft of the EJP RD → more than 70 experts involved + 24 ERN coordinators representing EU, associated and non-EU countries
- 12 Oct 2017 – 2nd EJP RD Concept Draft sent to the EC and WGs + ERN Coo
- 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
**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 third countries (AU, CA, JP, USA)
- **Timeline:** Jan 2019 – Dec 2024
EJP RD STRUCTURE

COORDINATION & TRANSVERSALE ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

SUSTAINABILITY

ETHICAL & REGULATORY

COMMUNICATION

1. FUNDING
2. COORDINATED ACCESS TO DATA & SERVICES
3. CAPACITY BUILDING & EMPOWERMENT
4. ACCELERATING TRANSLATION OF RESEARCH & THERAPY DEVELOPMENT

COORDINATED by
PILLAR 1: COLLABORATIVE RESEARCH FUNDING

Ralph SCHUSTER (DLR, DE) & Sonja van WEELY (ZonMw, NL)
# PILLAR 1 - PROPOSED STRUCTURE

<table>
<thead>
<tr>
<th>WP1: Joint Transnational Calls for collaborative research projects</th>
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<tr>
<td>WP2: Networking to share knowledge on rare diseases</td>
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<tr>
<td>WP3: Rare disease research challenges</td>
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<td>WP4: Monitoring of granted projects</td>
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PILLAR 2: COORDINATED ACCESS TO DATA AND SERVICES

Ana RATH (INSERM-Orphanet, FR) & Olaf Riess (Univ Tübingen, DE)
**PILLAR 2 - PROPOSED STRUCTURE**

<table>
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<tr>
<th>WP1: Management of the next-generation data ecosystem strategic plan</th>
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<tr>
<td>WP2: Common platform for discoverable data and resources for RD research</td>
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<td>WP3: Enabling sustainable FAIRness and Federation at the data record, patient and sample level for RD</td>
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<td>WP4: Enabling multidisciplinary, holistic approaches for rare disease diagnostics and therapeutics</td>
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<tr>
<td>WP5: Accelerating research capacity of the ERNs to increase diagnostic capacity and to hustle compound validation for therapy development</td>
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PILLAR 3: CAPACITY BUILDING AND EMPOWERMENT

Virginie BROS-FACER (EURORDIS), Birutė TUMIENE (Univ Vilnus, LT)
PILLAR 3 - PROPOSED STRUCTURE

WP1: Training on data management & quality

WP2: Capacity building & training of patients and researchers

WP3: Online academic education course

WP4: ERN RD training & support programmes

WP5: Development and adaptation of training activities
PILLAR 4: ACCELERATING TRANSLATION & THERAPY DEVELOPMENT

Rima NABBOUT (Imagine, FR), Anton USSI (EATRIS)
Maurizio SCARPA (ERN chair)
WP1: Accelerating Translation for Higher Patient Impact

WP2: Support to RD investigators in the preparation of Clinical Studies

WP3: Innovation in the methodologies of clinical studies in RD

WP4: Fostering private-public and public-public partnership
GOVERNANCE STRUCTURE

**GOVERNING BOARD**
Ultimate decision-making body

**COORDINATION LEVEL**

**MANAGEMENT OFFICE**
Day-to-day management, coordination & transversal activities

**EXECUTIVE COMMITTEE**
Implementation & progress monitoring

Coordinator, Pillar leaders, WP leaders

**STRATEGY LEVEL**

**OPERATIONAL LEVEL**

**European Commission**

National Mirror Groups

Policy Board (MS & EC)

Ensures alignment of strategies

Board of Funders

Ad hoc Advisory Board(s)

Independent advice

P1 call topics & management

P1
P2
P3
P4
WP1 WP2 WP3 WP1 WP2 WP3 WP1 WP2 WP3 WP1 WP2 WP3 WP1 WP2 WP3
THANK YOU

for any questions contact:

Daria julkowska
daria.julkowska@inserm.fr