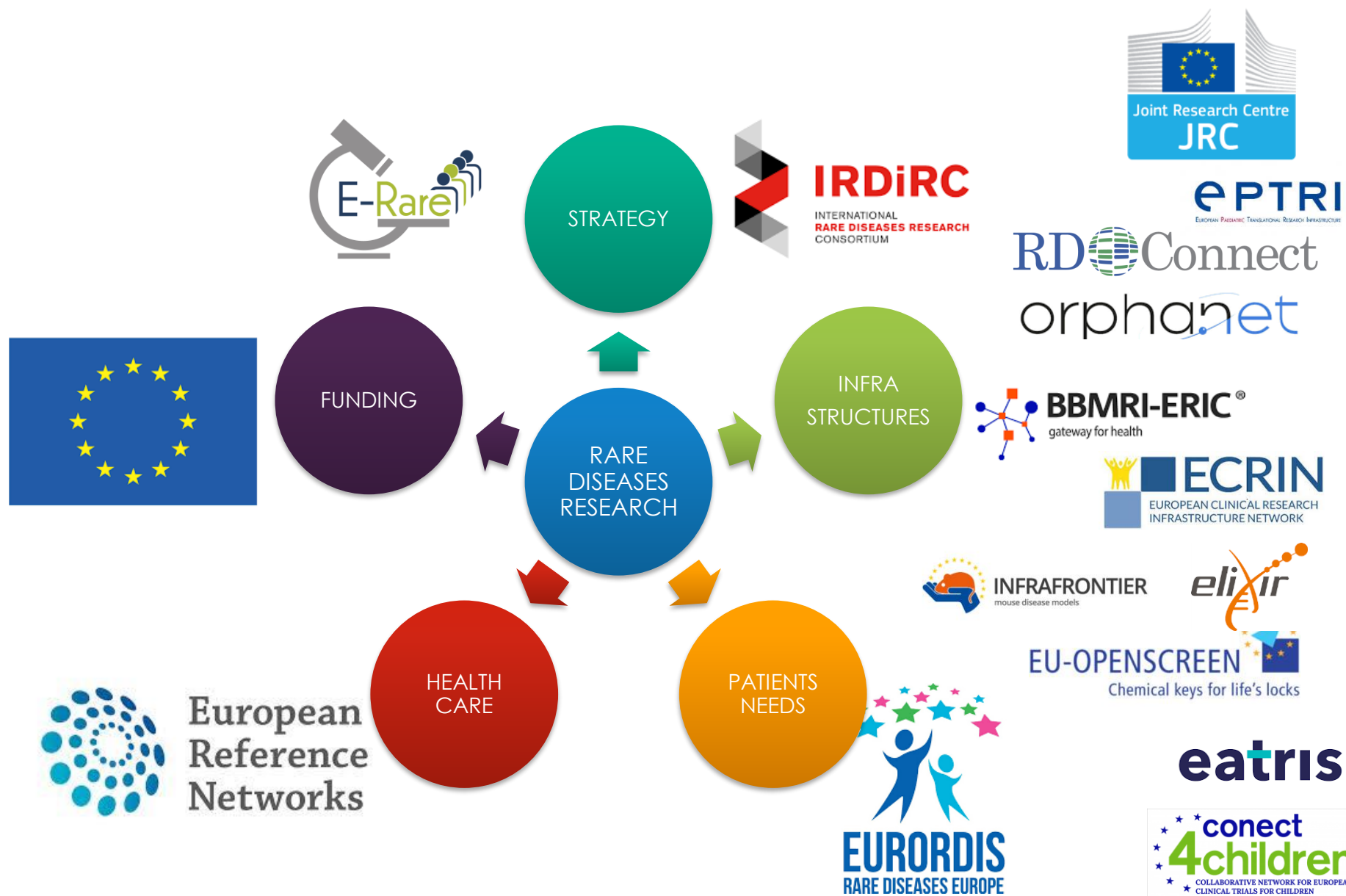



RESEARCHERS, NETWORKS, NEW FUNDS FOR RARE DISEASES

DARIA JULKOWSKA
ANR & INSERM, FRANCE

XI FORESIGHT TRAINING COURSE
Changes in Regulatory Sciences in the EU
25 – 27 of October 2018
Pavia, ITALY

RARE DISEASES LANDSCAPE IN EUROPE





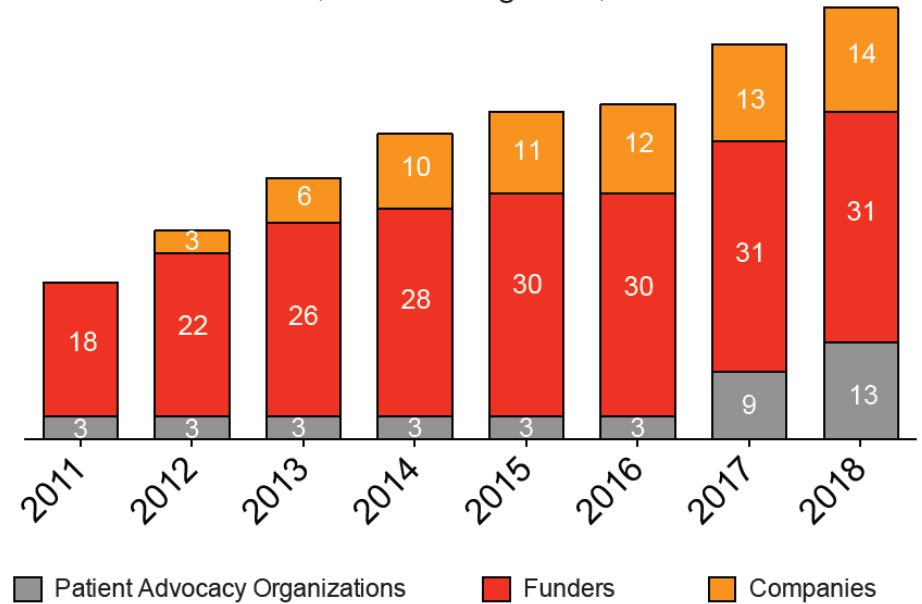
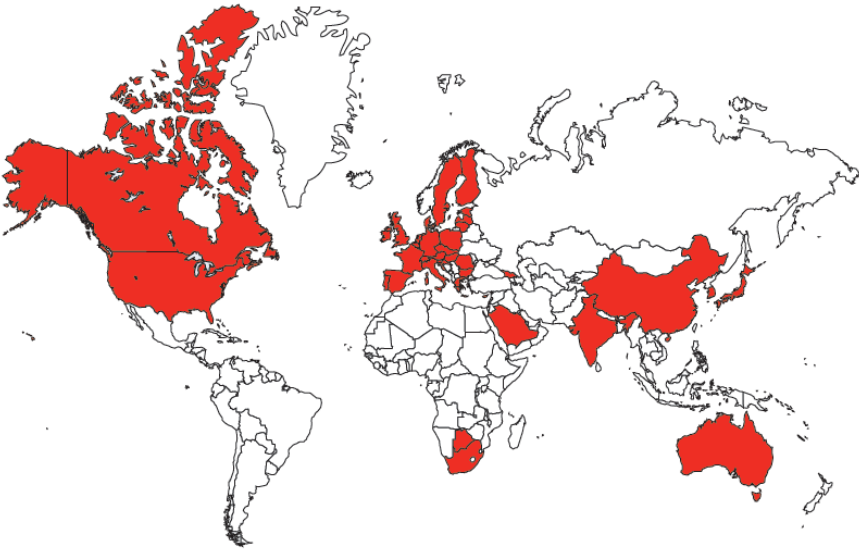
International Rare Diseases Research Consortium (IRDiRC)

**Co-operation at international level
to stimulate, better coordinate &
maximize output of rare disease
research efforts around the world**

IRDiRC Consortium Assembly

Representation

IRDiRC membership evolution from May 2011 to March 2018
(Cumulative growth)



► 58 IRDiRC members

- 31 funders
- 14 companies
- 13 patient advocates organizations

IRDiRC Goals 2017–2027

Released 9 August 2017

VISION: Enable all people living with a rare disease to receive an accurate diagnosis, care, and available therapy within one year of coming to medical attention

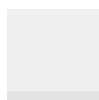
GOAL 1: All patients coming to medical attention with a suspected rare disease will be diagnosed within one year if their disorder is known in the medical literature; all currently undiagnosable individuals will enter a globally coordinated diagnostic and research pipeline.

GOAL 2: 1000 new therapies for rare diseases will be approved, the majority of which will focus on diseases without approved options.

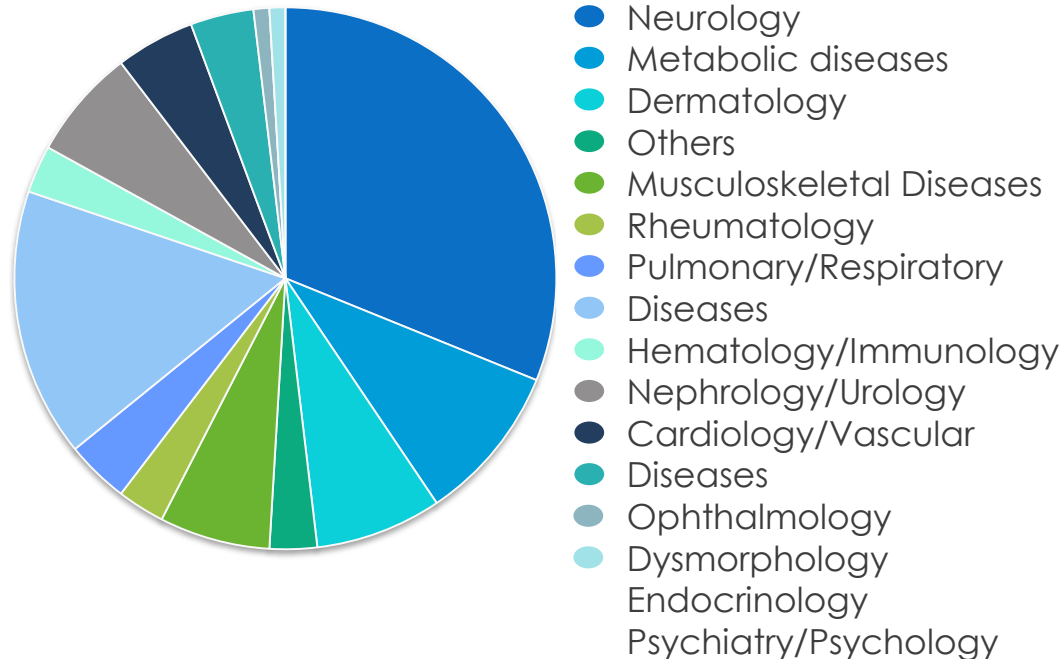
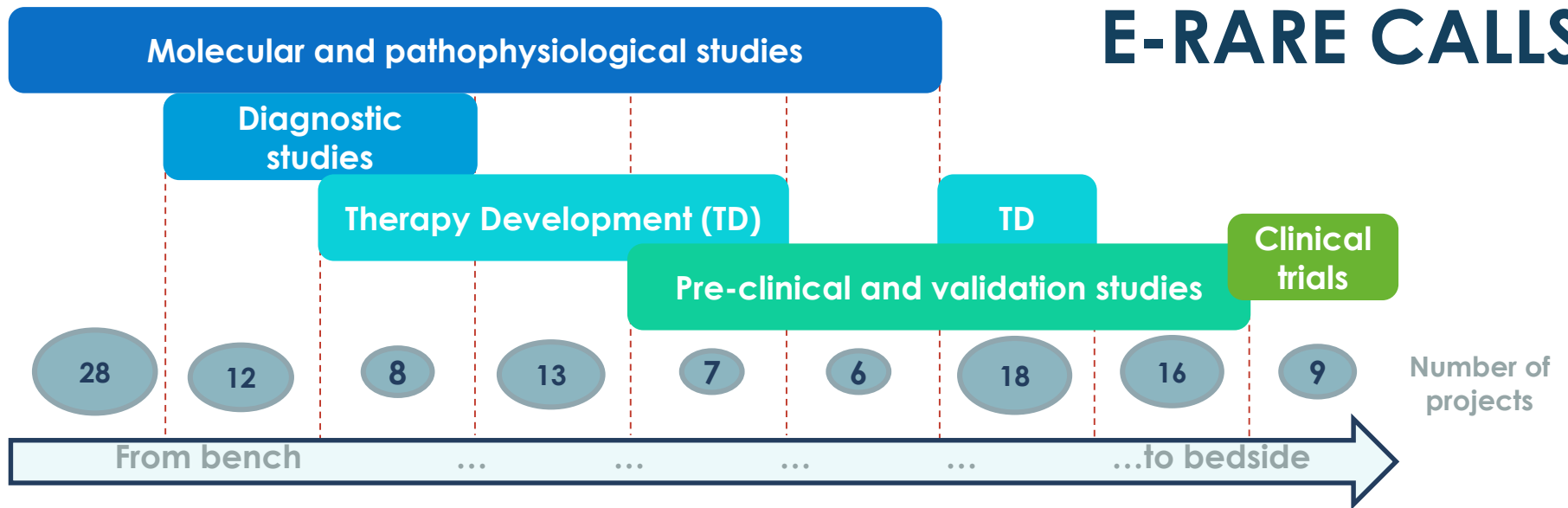
GOAL 3: Methodologies will be developed to assess the impact of diagnoses and therapies on rare disease patients.



E-RARE: ERA-NET ON RESEARCH PROGRAMMES FOR RARE DISEASES



E-RARE CALLS



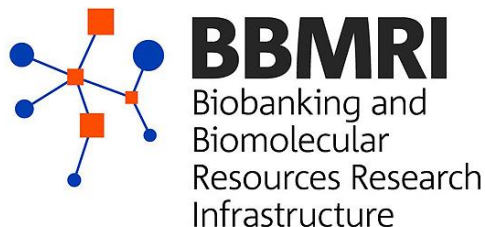
9 Joint Transnational Calls:

117 funded projects

104 M€ invested

Including **556** research partners funded

Medical domains represented in the funded projects



EUROPEAN RESEARCH INFRASTRUCTURES:

BIOBANKS, CLINICAL TRIALS, TRANSLATIONAL MEDICINE,
PAEDIATRIC RESEARCH, MOLECULE SCREENING, MOUSE
MODELS, DATA, RD ONTOLOGY, REGISTRIES





EURORDIS-Rare Diseases Europe is a unique, non-profit alliance **of 812 rare disease patient organisations from 70 countries** that work together to improve the lives of the 30 million people living with a rare disease in Europe.



European Reference Networks

European Reference Networks (ERNs) are virtual networks involving healthcare providers across Europe. They aim to facilitate discussion on complex or rare diseases and conditions that require highly specialised treatment, and concentrated knowledge and resources.

There are **24 ERNs, representing together 300 hospitals (900 care units)**

RARE DISEASES LANDSCAPE IN EUROPE

TOPIC : Rare Disease European Joint Programme Cofund

Topic identifier: SC1-BHC-04-2018

Publication date: 27 October 2017

Types of action: COFUND-EJP COFUND (European Joint Programme)

DeadlineModel: single-stage

Opening date: 07 November 2017

Deadline: 18 April 2018 17:00:00

Time Zone : (Brussels time)



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

EUROPEAN JOINT PROGRAMME ON RARE DISEASES





EUROPEAN JOINT PROGRAMME ON RARE DISEASES

- **Union contribution:** 55 M€ (70% reimbursement rate)
 - **Total budget (min. submitted):** 93,53 M€ (→ expected > 110 M€)
 - **Number of partners:** 88
 - **Number of participating countries(beneficiaries and LTPs):** 33
including 25 EU MS (AT, BE, BG, CZ, DE, ES, EE, FI, FR, HU, IE, IT, NL, LT, LV, LU, MT, PL, PT, RO, SE, SK, SL, SV, UK), 8 associated (AM, CH, GE, HR, IL, NO, RS TK) and third countries (CA)
 - **Timeline:** Jan 2019 – Dec 2023
-
- **Types of partners:**
 - 31 research funding bodies/ministries
 - 12 research institutes
 - 22 universities/hospital universities
 - 11 hospitals
 - 5 EU infrastructures (BBMRI, EATRIS, ECRIN, ELIXIR, INFRAFRONTIER) + EORTC
 - EURORDIS & ePAGs
 - 5 charities/foundations (FTELE, AFM, FFRD, FGB, BSF)

} **24 ERNs**



EJP RD STRUCTURE

COORDINATION
& TRANSVERSAL 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



WP1 COORDINATION & MANAGEMENT

WP2
STRATEGY

WP3
SUSTAINABILITY

WP4
ETHICS, LEGAL, REGULATORY & IPR

WP5
COMMUNICATION & DISSEMINATION



WP6
Joint Transnational Calls

WP7
Networking scheme

WP8
RDR Challenges

WP9
Monitoring of funded projects



WP 10
User-driven strategic planning for P2

WP 11
Virtual Platform for data & resources

WP 12
Enabling sustainable FAIRness

WP 13
Holistic approaches for rare disease diagnostics and therapeutics



WP 14
Training on data management & quality

WP 15
Capacity building and training of patients and researchers

WP 16
Online Academic education course

WP 17
ERN RD training and support programme

WP 18
Development and adaptation of training activities

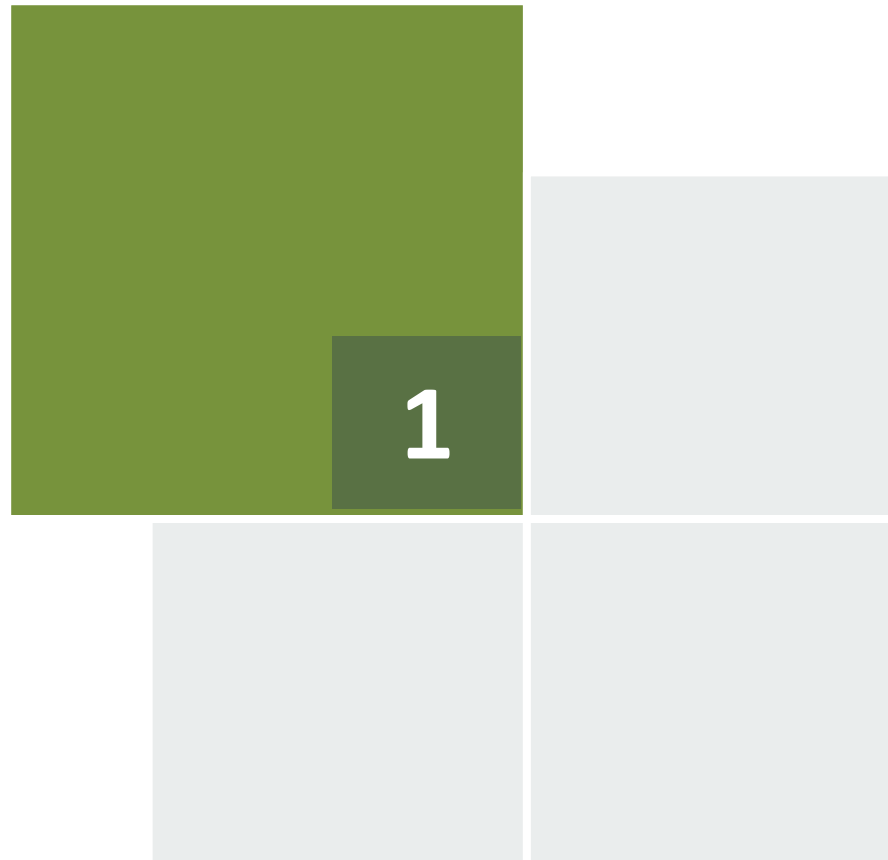


WP 19
Facilitating partnerships and accelerating translation

WP 20
Validation, use and development of innovative methodologies for clinical studies

Coordinated by

PILLAR 1: COLLABORATIVE RESEARCH FUNDING



Ralph SCHUSTER (DLR, DE) & Sonja van WEELY (ZonMw, NL)



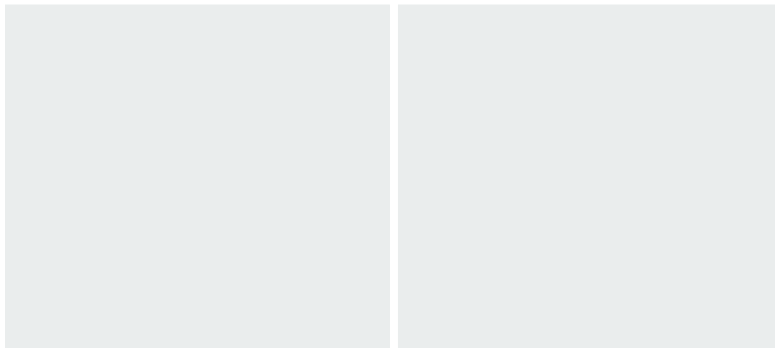
PILLAR1 STRUCTURE

WP6: Joint Transnational Calls for collaborative research projects

WP7: Networking to share knowledge on rare diseases

WP8: Rare disease research challenges

WP9: Monitoring of funded projects





PILLAR 1

WP6: Joint Transnational Calls for collaborative research projects

2 cofunded calls + 2 non-cofunded – 23 M€ budget for call 1 – open to research teams from countries with funders involved – min of 3 teams from 3 countries

WP7: Networking to share knowledge on rare diseases

Small support schemes for networking (workshops/events/share of knowledge) – 30 K€ max – open all year long – open to all countries involved in EJP RD

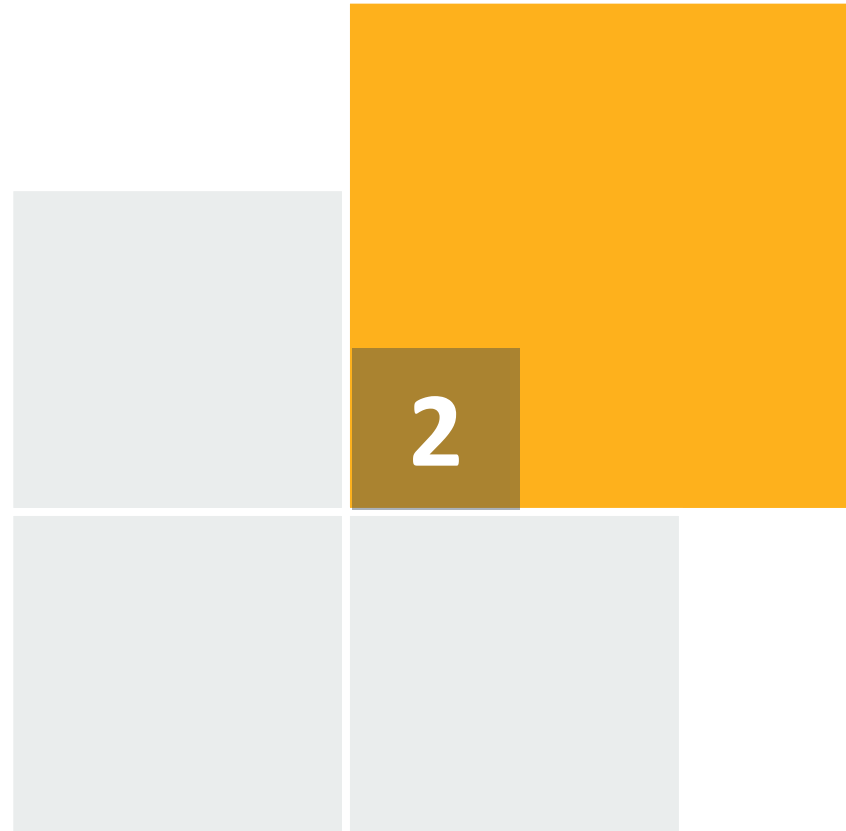
WP8: Rare disease research challenges

Public-private (small scale) partnerships – challenges set by industry and validated by EJP RD – short term (max. 18 months) projects

WP9: Monitoring of funded projects

Monitoring of all projects funded through EJP RD and previous E-Rare projects

PILLAR 2: INNOVATIVE COORDINATED ACCESS TO DATA AND SERVICES FOR TRANSFORMATIVE RARE DISEASES RESEARCH



Ana RATH (INSERM-Orphanet, FR) & Franz Schaeffer (Univ Heidelberg, DE)



PILLAR 2 - PROPOSED STRUCTURE

WP10: User-driven strategic planning and transversal activities for Pillar 2 data ecosystem

WP11: Common virtual platform for discoverable data and resources for RD research

WP12: Enabling sustainable FAIRness and Federation at the record for RD data, patients and samples

WP13: Enabling multidisciplinary, holistic approaches for rare diseases diagnostics and therapeutics





PILLAR 2

WP10: User-driven strategic planning and transversal activities for Pillar 2 data ecosystem

Annual strategic meetings with users (ERNs) & developers to define the priorities – coordination of outputs & needs – technical GDPR implementation – quality, sustainability and scaling up

WP11: Common virtual platform for discoverable data and resources for RD research

Metadata & ontological models – FAIR compliance – data deposition & access to data infras – online tools

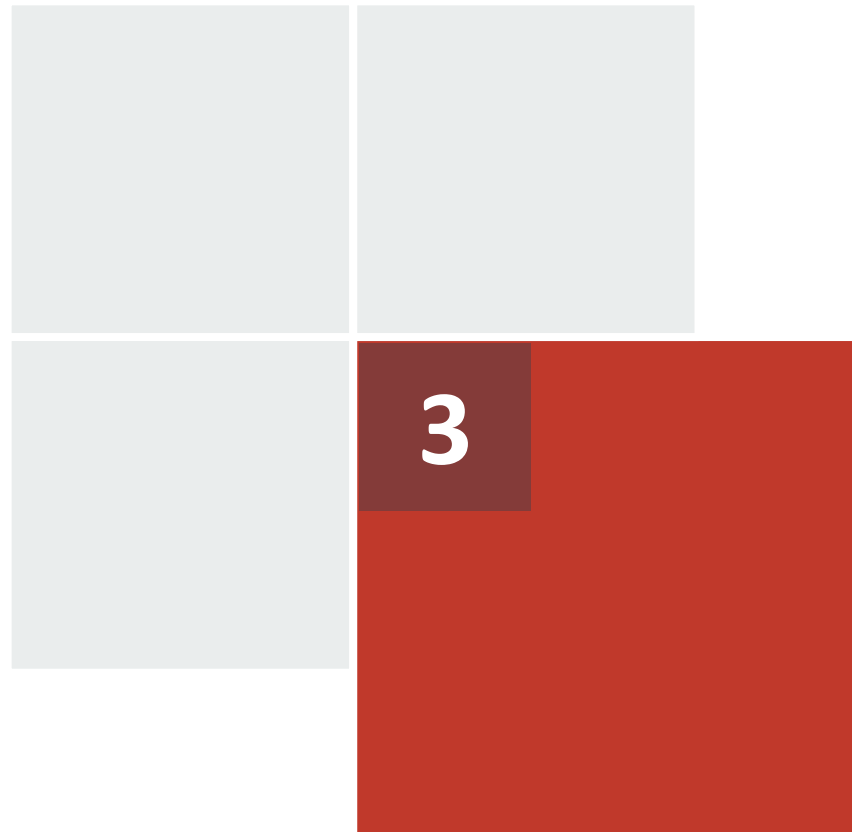
WP12: Enabling sustainable FAIRness and Federation at the record for RD data, patients and samples

Alignment of core interoperability standards – software for FAIR ecosystem – FAIRification support

WP13: Enabling multidisciplinary, holistic approaches for rare diseases diagnostics and therapeutics

System biology approaches for RD – biological pathways – variants to function – environmental toxicology – treatment drugs - proof of principle studies

PILLAR 3: CAPACITY BUILDING AND EMPOWERMENT



Virginie BROS-FACER (EURORDIS), Biruté TUMIENE (Univ Vilnus, LT)



European
Reference
Networks



EURORDIS
RARE DISEASES EUROPE



PILLAR3 STRUCTURE

orphanet



RD  **Connect**

WP14: Training on data management & quality

WP15: Capacity building & training of patients and researchers in rare diseases research and processes

WP16: Online academic education course

WP17: ERN RD training & support programmes

WP18: Development and adaptation of training activities



PILLAR 3

WP14: Training on data management & quality

Trainings on: orphanet nomenclature – standards & quality of genetics/genomics data in clinical practice – strategies to foster undiagnosed diseases – biobanks sample data management – rare diseases registries & FAIRification at source – European Rare Diseases Registry Infrastructure

WP15: Capacity building & training of patients and researchers in rare diseases research and processes

Expert Patients and Researchers EURORDIS Summer school – scientific innovation and translation research aspects in RDs for patient advocates – leadership & communication skills for patient advocates and representatives – education material and activities for paediatric patients

WP16: Online academic education course

Based on assessed needs of the RD community – in collaboration with universities – 10 to 12 modules with accreditation – e-learning format open to all – Future Learn platform

WP17: ERN RD training & support programmes



PILLAR 3

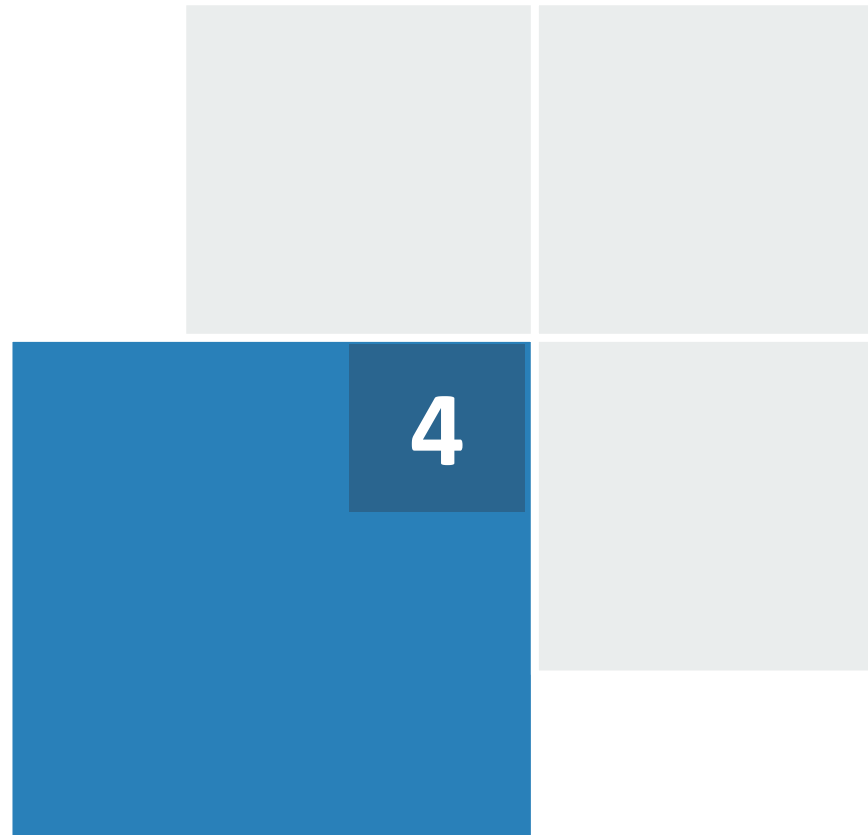
WP17: ERN RD training & support programmes

Based on four groups (Neuro, Neoplasm & malformation, Organs, Systemic) – preferences, needs and resources of ERNs – tailored for and performed by ERNs

WP18: Development and adaptation of training activities

Evaluation of developing needs according to progress of Pillars 2 & 4 – specific needs of EU 13 countries – emerging needs of ERNs

PILLAR 4: ACCELERATING THE TRANSLATION OF HIGH POTENTIAL PROJECTS & IMPROVING OUTCOMES OF CLINICAL STUDIES IN SMALL POPULATIONS



Rima NABBOUT (Imagine, FR), Anton USSI (EATRIS)



PILLAR4 STRUCTURE

imagine
INSTITUT DES MALADIES GÉNÉTIQUES



European
Reference
Networks



eatris



WP19: Facilitating partnerships and accelerating translation for higher patient impact

WP20: Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs



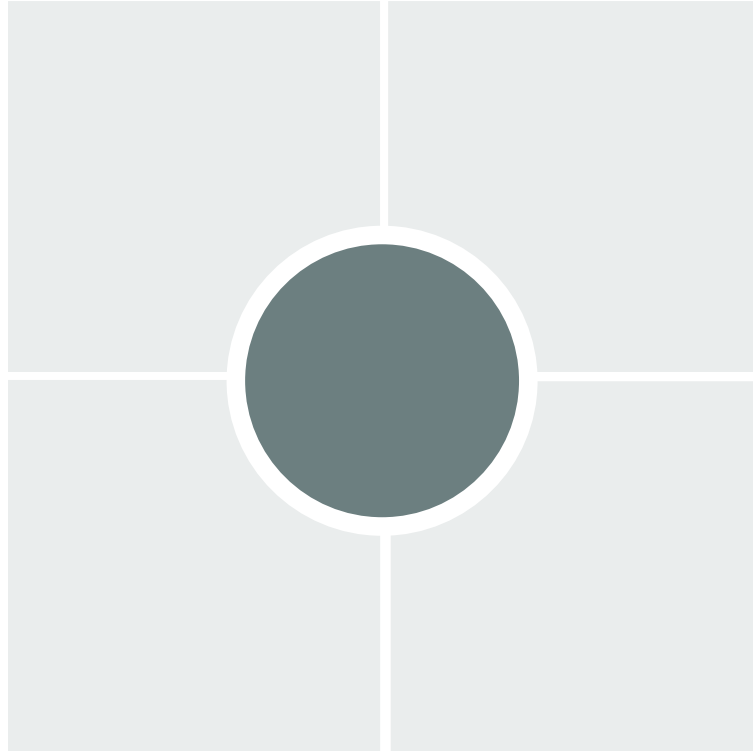
WP19: Facilitating partnerships and accelerating translation for higher patient impact

Innovation management toolbox – assessment and real time mentoring of translational projects – support in exploitation and follow-on funding – partnering support – roadmap for European investment platform for RD

WP20: Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs

Key Task Force group - Support in design and planning of RD clinical studies with ECRIN – demonstration projects on existing statistical methodologies to improve RD clinical trials – innovative methodologies to improve RD clinical trials in limited populations

TRANSVERSAL ACTIVITIES





TRANSVERSAL ACTIVITIES

WP1: PROGRAMME MANAGEMENT & COORDINATION

WP2: INTEGRATIVE RESEARCH & INNOVATION STRATEGY

WP3: SUSTAINABILITY

WP4: ETHICS, LEGAL, REGULATORY & IPR

WP5: COMMUNICATION & DISSEMINATION



European
Reference
Networks



Inserm

La science pour la santé
From science to health



FONDAZIONE
PER LA RICERCA FARMACOLOGICA
GIANNI BENZI
ONLUS





TRANSVERSAL ACTIVITIES

WP1: PROGRAMME MANAGEMENT & COORDINATION

Coordination office & governance – SciSec of IRDiRC – Central Helpdesk – Annual work plans – monitoring/KPIs – Data management plan

WP2: INTEGRATIVE RESEARCH & INNOVATION STRATEGY

Prioritization strategy – mapping of research needs – scientific programming of calls – medium & long term strategy – alignment of national and EU strategies

WP3: SUSTAINABILITY

Roadmap of needs & expectations – sustainable service catalogue – EJP RD sustainability business plan

WP4: ETHICS, LEGAL, REGULATORY & IPR

AREB – management of transversal legal, ethical & IPR issues

WP5: COMMUNICATION & DISSEMINATION

External communication & dissemination of EJP RD & IRDiRC results – expansion to & interactions with stakeholders

GOVERNANCE



STRATEGY LEVEL

GENERAL ASSEMBLY

Ultimate decision-making body

GOVERNING BOARD

Mandated decision-making body

COORDINATION LEVEL

COORDINATION OFFICE

Day-to-day management, coordination & transversal activities

European
Commission

National
Mirror
Groups

Policy
Board
(MS & EC)

Ensures alignment
of strategies

OPERATIONAL LEVEL

EXECUTIVE COMMITTEE

Implementation, progress monitoring, annual programming

OPERATING GROUP

Coordination of activities between Pillars & Transversal WPs

**Ad hoc
Advisory
Board(s)**

Independent
advice

**Board
of Funders**

P1 call topics
& management

P1

P2

P3

P4

WP

WP

WP

WP

WP

WP

WP

WP

WP

WP

WP

WP

ARE YOU EJP RD SUPERHERO?



THANK YOU

DARIA.JULKOWSKA@INSERM.FR