EUROPEAN JOINT PROGRAMME ON RARE DISEASES

DARIA JULKOWSKA
INSERM, FRANCE

RARE DISEASES LANDSCAPE IN EUROPE

RARE DISEASES RESEARCH

STRATEGY

INFRA STRUCTURES

PATIENTS NEEDS

HEALTH CARE +

FUNDING

E-Rare

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

INFRAFRONTIER
mouse disease models

ePTRI
European Paediatric Translational Research Infrastructure

RD Connect

orphanet

BBMRI-ERIC®
gateway for health

ECRIN
EUROPEAN CLINICAL RESEARCH
INFRASTRUCTURE NETWORK

Joint Research Centre
JRC

elixir

EU-OPENSCREEN
Chemical keys for life's locks

eatris

conect4children
COLLABORATIVE NETWORK FOR EUROPEAN
CLINICAL TRIALS FOR CHILDREN

European
Reference
Networks

EURORDIS
RARE DISEASES EUROPE

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.

IRDiRC
INTERNATIONAL
RARE DISEASES RESEARCH
CONSORTIUM

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

EUROPEAN JOINT PROGRAMME ON RARE DISEASES

EUROPEAN JOINT PROGRAMME ON RARE DISEASES

- **Union contribution:** 55 M€ (70% reimbursement rate)
- **Total budget (min. submitted):** 101 M€ (→ expected > 110 M€)
- **Number of partners:** 88
- **Number of participating countries(beneficiaries and LTPs):** 35 including 27 EU MS (AT, BE, BG, CZ, DE, ES, EE, FI, FR, GR, HU, HR, IE, IT, NL, LT, LV, LU, MT, PL, PT, RO, SE, SK, SL, SV, UK), 7 associated (AM, CH, GE, 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

24 ERNs

  - 5 EU infrastructures (BBMRI, EATRIS, ECRIN, ELIXIR, INFRAFRONTIER) + EORTC
  - EURORDIS & ePAGs
  - 5 charities/foundations (FTELE, AFM, FFRD, FGB, BSF)

Coordinated by
Inserm
La science pour la santé
From science to health

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

P1

WP6
Joint Transnational Calls

WP7
Networking scheme

WP8
RDR Challenges

WP9
Monitoring of funded projects

P2

WP 10
User-driven strategic planning for P2

WP 11
Virtual Platfform for data & resources

WP 12
Enabling sustainable FAIRness

WP 13
Holistic approaches for rare disease diagnostics and therapeutics

P3

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

P4

WP 19
Facilitating partnerships and accelerating translation

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

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PILLAR 1: COLLABORATIVE RESEARCH FUNDING

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

PILLAR 1

WP6: Joint Transnational Calls for collaborative research projects

2 cofunded calls + 2 non-cofunded – 27 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

PILLAR1

- Joint Transnational Calls for collaborative research projects
- Networking to share knowledge on rare diseases
- Rare disease research challenges
- Monitoring of funded projects

- Achievement of critical mass of knowledge & resources
- Accelerated diagnosis and treatment development
- New and expanded networks – inclusion of stakeholders, share of knowledge
- New diseases targeted
- Public-private partnerships
- PoC and optimisation studies

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

Pillar leaders: Ana RATH (INSERM-Orphanet, FR) & Franz Schaeffer (Univ Haidelberg, DE)

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

Alignement 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 2

- User-driven strategic planning and transversal activities for Pillar 2 data ecosystem
- Common virtual platform for discoverable data and resources for RD research
- Enabling sustainable FAIRness and Federation at the record for RD data, patients and samples
- Enabling multidisciplinary, holistic approaches for rare diseases diagnostics and therapeutics

- Building the next generation data strategy
- Removing obstacles to finding and sharing of data & resources
- FAIR data for the RD community
- Data driven RD innovation

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mouse disease models

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elixir

European
Reference
Networks

PILLAR 3: CAPACITY BUILDING AND EMPOWERMENT

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

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 – eduocation 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 3

Training on data management & quality

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

Online academic education course

ERN RD training & support programmes

Development and adaptation of training activities

- Contributing to Responsible Research & Innovation goals
- Increasing the capacity of next generation of RD stakeholders
- Open access RD education
- Sustainability and scalability of competence transmission
- Outreach to less developed communities

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

Pillar leaders: Rima NABBOUT (Imagine, FR), Anton USSI (EATRIS)

PILLAR4

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

PILLAR 4

- Facilitating partnerships and accelerating translation for higher patient impact
- Accelerating the validation, use and development of innovative methodologies tailored for clinical trials in RDs

- Improved patient impact potential
- More sustainable and exploitable academic research
- Improved clinical trial methodologies for small populations
- Roadmap for RD innovation funding

imagine
INSTITUT DES MALADIES GÉNÉTIQUES

Telethon

ECRIN
EUROPEAN CLINICAL RESEARCH INFRASTRUCTURE NETWORK

European Reference Networks

eatris

COORDINATION & TRANSVERSAL ACTIVITIES

COORDINATION & TRANSVERSAL ACTIVITIES

PROGRAMME MANAGEMENT & COORDINATION

INTEGRATIVE RESEARCH & INNOVATION STRATEGY

SUSTAINABILITY

ETHICS, LEGAL, REGULATORY & IPR

COMMUNICATION & DISSEMINATION

European Reference Networks

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ISTITVTO SVPERIORE DI SANITA

CVBF
Consorzio per Valutazioni Biologiche e Farmacologiche
Branch of Albania
TIRANA

CORBEL

FONDAZIONE PER LA RICERCA FARMACOLOGICA GIANNI BENZI ONLUS

EURORDIS
RARE DISEASES EUROPE

isc
Instituto de Salud Carlos III

conect4children
COLLABORATIVE NETWORK FOR EUROPEAN CLINICAL TRIALS FOR CHILDREN

TEDDY NETWORK
European Network of Excellence fo Paediatric Clinical Research

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 – alignement if national and EU strategies

WP3: SUSTAINABILITY

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

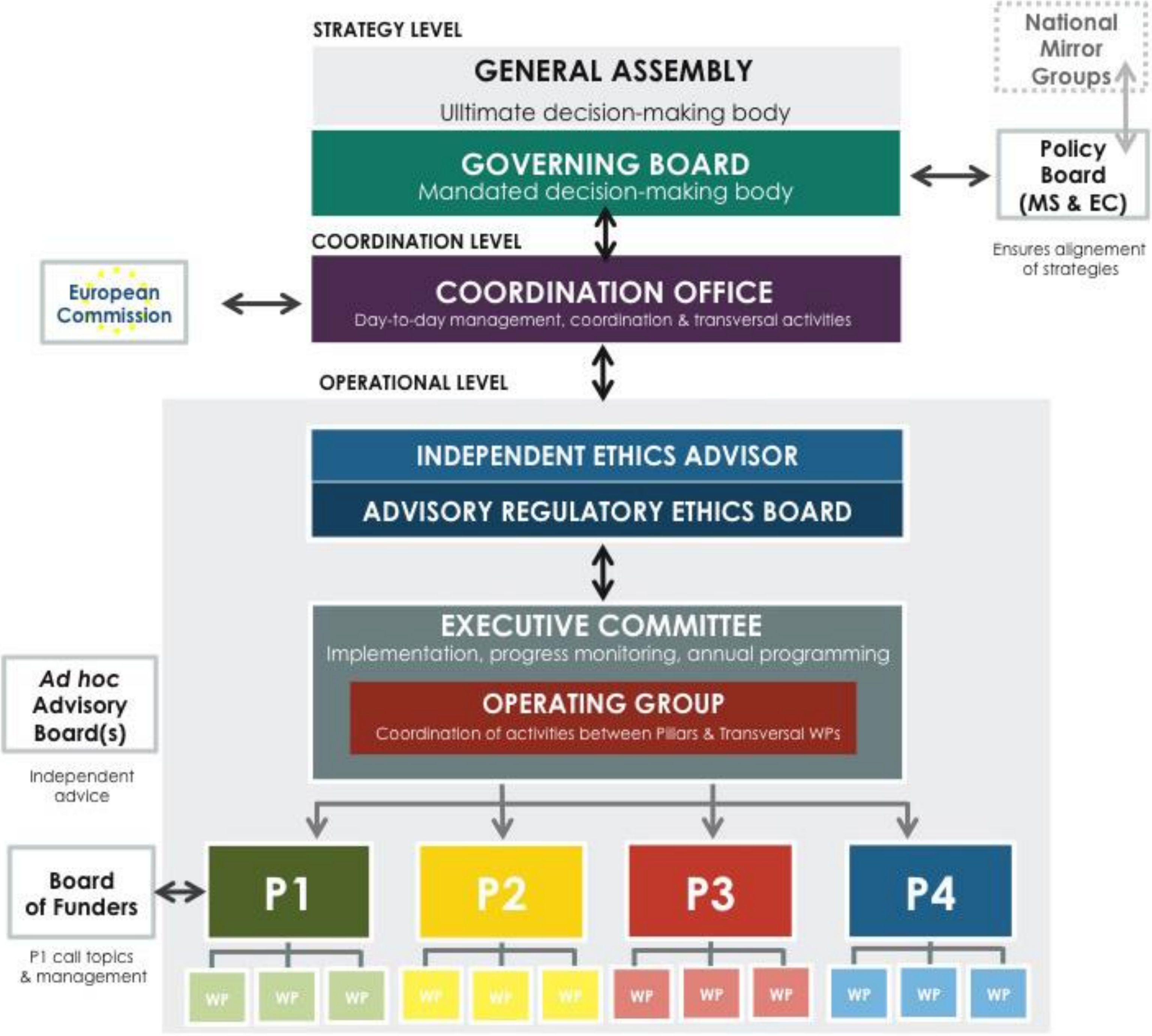
WP4: ETHICS, LEGAL, REGULATORY & IPR

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

Ulltimate decision-making body

GOVERNING BOARD

Mandated decision-making body

National Mirror Groups

Policy Board (MS & EC)

Ensures alignement of strategies

COORDINATION LEVEL

European Commission

COORDINATION OFFICE

Day-to-day management, coordination & transversal activities

OPERATIONAL LEVEL

INDEPENDENT ETHICS ADVISOR

ADVISORY REGULATORY ETHICS BOARD

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

POLICY BOARD & NATIONAL MIRROR GROUPS

The **POLICY BOARD** will have a major role in ensuring this dialogue and translation through its participation is EJP RD strategy and sustainability development. It will meet once a year.

The Policy Board will be constituted from:

- Representatives of national ministries of research and health;
- Representatives of European Commission Directorates: DG RTD, DG Santé, DG Connect;
- Representative of the pharmaceutical industry and public-private initiatives (e.g. European Federation of Pharmaceutical Industries and Associations, EFPIA; Innovative Medicines Initiative, IMI);
- Representative of EuropaBio;
- Representative of regulatory authorities (e.g. European Medicines Agency, EMA, esp. Committee for Orphan Medicinal Products, COMP, EuNetHTA);
- Chair of the European Strategy Forum on Research Infrastructures (ESFRI);
- Chair and vice-chair of the International Rare Diseases Research Consortium (IRDiRC).

NATIONAL MIRROR GROUPS:

- NMG ensures national coordination, contribute to the objectives of the EJP RD and benefit from it
- Is expected to include representatives of the National plan for RD, national nodes of the European Reference Networks, relevant national authorities and research institutions (whether participating to the EJP RD or not), as well as the relevant national partners of the EJP RD and **GB member** that will report NMG views and positions during GB meetings.

JOINT TRANSNATIONAL CALL

2019

FLORENCE GUILLOT

ANR, FRANCE

PILLAR 1

WP6: Joint Transnational Calls for collaborative research projects

1st Joint Transnational Call (2019)

- Total budget 27 M€ from participating Member States + min 5 M€ of EC contribution
- 31 funders from 23 countries (AT, BE, CA, CZ, EE, FI, FR, DE, GR, HU, IE, IL, IT, LT, LU, PL, PO, SK, ES, SE, CH, NL, TK)
- Topic: Research projects to accelerate diagnosis and/or explore disease progression and mechanisms of rare diseases
  - <u>Research to accelerate diagnosis, e.g:</u>

o New schemes for finding diagnosis for undiagnosed patients;

o Improved annotation and interpretation of variants and development of diagnostic tests for the more prevalent variants;

o Novel modalities of functional analysis of candidate variants through in vitro, cell, tissue or animal studies.

o -omic or multi-omic integrated approaches for discovery of disease causes and mechanisms including development of relevant bioinformatic tools;

  - <u>Research to explore disease progression and mechanisms, e.g:</u>

o Natural history studies and patient registries (also for clinical trial readiness). Whenever possible these should include development and use of patient reported outcome measures. In addition, the exploration of the use of standardized M-Health-based surveillance instruments and of patient entered data to gather information for natural history studies is welcome;

o Identification of clinical biomarkers, clinical outcome measures and surrogate endpoints;

o Identification of novel pathophysiological pathways in appropriate disease models that effectively mimic the human condition

PILLAR 1

WP6: Joint Transnational Calls for collaborative research projects

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

- Approaches concerning rare infectious diseases or rare cancers;
- Approaches concerning rare adverse drug events/medical complications in treatments of common diseases;
- Studies that focus on pre-clinical therapy development and/or validation in cellular or animal models. These will be addressed in future calls;
- Interventional clinical trials;
- Rare neurodegenerative diseases which are within the main focus of the Joint Programming Initiative on Neurodegenerative Disease Research (JPND; http://www.neurodegenerationresearch.eu/). These concern: Alzheimer’s disease and other dementias; Parkinson’s disease (PD) and PD-related disorders; Prion disease; Motor Neuron Diseases; Huntington’s disease; Spinal Muscular Atrophy and dominant forms of Spinocerebellar Ataxia. Interested researchers should refer to the relevant JPND calls. Not excluded through this specification are childhood dementias/neurodegenerative diseases.

FOR OTHER DETAILS (ELIGIBILITY RULES PLEASE CHECK CAREFULLY THE CALL TEXT, GUIDELINES FOR APPLICANTS AND PROPOSAL FORMS)

PILLAR 1

WP6: Joint Transnational Calls for collaborative research projects

Eligible partners (check the national rules!):

- academia (research teams working in universities, other higher education institutions or research institutes)
- clinical/public health sector (research teams working in hospitals/public health and/or other health care settings and health organisations)
- enterprise (all sizes of private companies). Participation of small and medium-size enterprises (SMEs) is encouraged when allowed by national/regional regulations
- patient advocacy organisations (PAOs - see more information below and refer to the INSERM contact point) **– TBC**

Other eligibility rules:

- Min. of 4 eligible partners from 4 different countries
- Max. of 6 partners per project that can be extended to 8 if underrepresented countries (CZ,EE,HU, LT, PL, SK, TK) participate
- The coordinator must be always eligible
- Associated partners from countries NOT participating in the call may be part of the project if they participate with their own funding, their number is limited to 2 (check details in the call rules)
- Timetable:
  - Call opens on 14/12/2018
  - Pre-proposal submission closes on 14/02/2019
  - Invitation of selected project to submit full proposal – early May 2019
  - Submission of full proposals 11/06/2019
  - Final results – October 2019

PILLAR 1

WP6: Joint Transnational Calls for collaborative research projects

CONTACT ANR:
Florence Guillot
Florence.guillot@agencerechereche.fr
Tel. 01 78 09 80 01

We strongly advise you to contact Florence by mail (to facilitate the management and record of responses)

CONTACT Fondation Maladies Rares (FFRD):
Website: https://fondation-maladiesrares.org/eng/
Ingrid Zwaenepoel
Tel. 01 58 14 22 85 or

Diana Désir-Parseille
Tel : + 01 58 14 22 81
aap-bio@fondation-maladiesrares.com

CONTACT INSERM (only questions related to participation of Patient Advocacy Organisations)
Daria Julkowska
Daria.julkowska@inserm.fr

PILLAR 1

WP6: Joint Transnational Calls for collaborative research projects

EJP RD JTC 2019 SECRETARIAT (contact for any issues related to the call and not specific to FR participation):

Dr. Katarzyna Saedler
katarzyna.saedler@dlr.de
+49 228 3821 1947

Dr. Michaela Fersch
+ 49 228 3821 1268
michaela.fersch@dlr.de

Dr. Ralph Schuster
+ 49 228 3821 1233
ralph.schuster@dlr.de

INFO ON:

Currently: www.erare.eu

Starting from 14 December 2018: www.ejprarediseases.org

MERCI

DARIA.JULKOWSKA@INSERM.FR

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