PATIENT ENGAGEMENT IN RD RESEARCH

CNA/CEF MEETING, 11th December 2018, Paris

Virginie Bros-Facer

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OVERVIEW

TODAY:

1. RD-Connect 6 years 2012-2018 (FP7)
2. Solve-RD 5 years 2018-2022 (H2020)
3. European Joint Programme on Rare Diseases for 5 years 2019-2023 (H2020)

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RD-CONNECT

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RD-CONNECT IN BRIEF (2012-2018 UNDER FP7)

Integrated platform that connects data from Next Generation Sequencing, biobanks, registries and –omics technologies to facilitate diagnosis and improve treatments for rare diseases

- **30 partner organisations**
- **8 Work Packages (Management, Biobanks, Registries, Genome-Phenome analysis platform, Bioinformatic tools, ELSI, Patient engagement & Communication) – 12M euros**

Within the European rare disease community, RD-Connect has been highlighted as a successful infrastructure that is facilitating and accelerating research and diagnosis.

PATIENT ADVISORY COUNCIL(PAC)

Chris Sotirelis, UK Thalassaemia Society

Lydie Lemmonier, Vaincre la mucoviscidose (Cystic Fibrosis)

Joseph Irwin, Spinal Muscular Atrophy Support UK

Veronica Popa, Allan-Herndon-Dudley syndrome

Ciaran Scott, The Alkaptonuria Society

Alexandre Méjat, French Muscular Dystrophy Association - Téléthon

Virginie Bros-Facer, EURORDIS

Rainald von Gizycki, PRO RETINA Deutschland e.V.

Daniel Renault, Federation of European Associations of Patients affected by Renal Genetic Diseases

Dorthe Lykke, European Federation of Hereditary Spastic Paraplegia

Elizabeth Vroom Duchenne Parent Project/United Parent Projects MD

Muriel Arcaute-Gevrey, CMT-France association (Charcot-Marie-Tooth disease)

Julian Isla, Dravet Syndrome Foundation

Kay Parkinson
Director, Alstrom Syndrome Europe
CEO, Cambridge Rare Disease Network

Marieke van Meel, NephcEurope

Sigurður Jóhannesson, Alternating Hemiplegia association of Iceland (AHCAI)/AHCFE Europe

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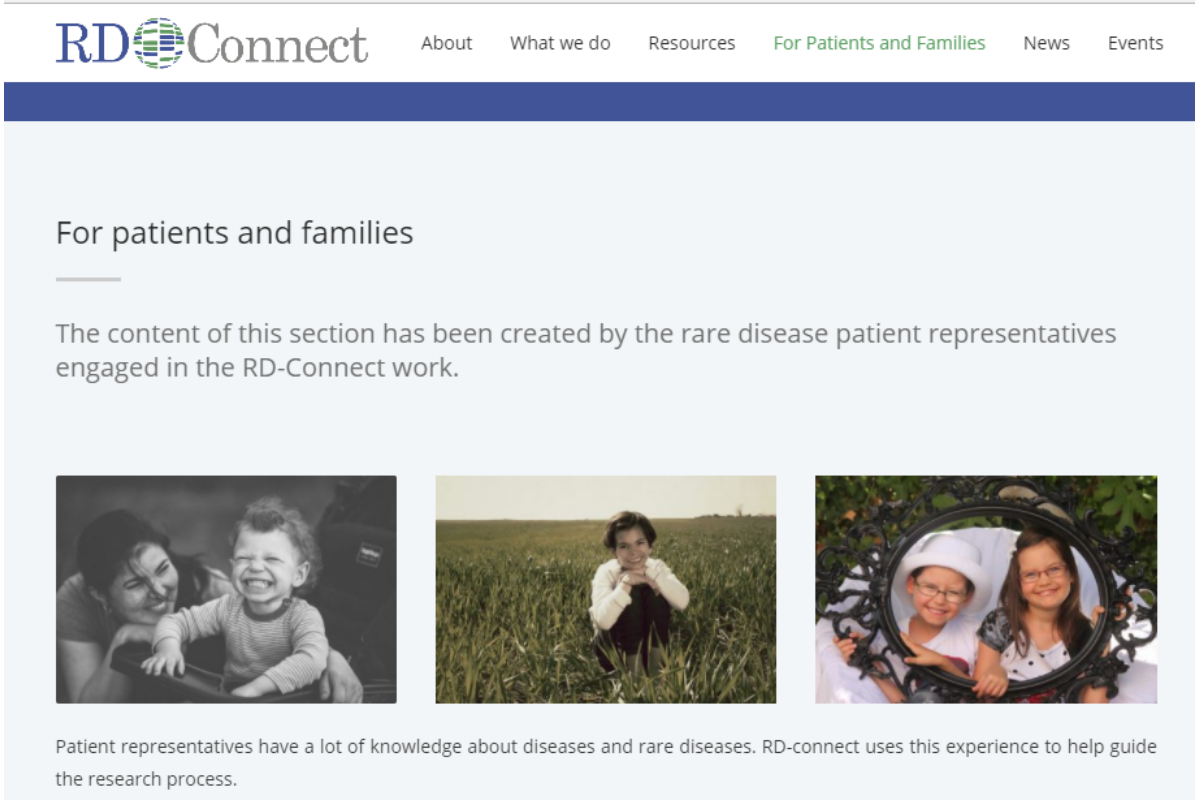
PAC MAIN ACHIEVEMENTS – COMMUNICATION (WP7)



Video interviews of PAC members on their involvement in RD-Connect, full articles for the RD-Connect Newsletter, glossary on terms used in the project and beyond, EURORDIS webinar RD-Connect for Summer School Alumni, infographics on registries plus a lot of dissemination

PAC MAIN ACHIEVEMENTS – ELSI (WP6)

EJHG Open

European Journal of Human Genetics (2016), 1–7

www.nature.com/ejhg

ARTICLE

Improving the informed consent process in international collaborative rare disease research: effective consent for effective research

Sabina Gainotti*[,1,9], Cathy Turner[2], Simon Woods[3,9], Anna Kole[4,9], Pauline McCormack[3,9], Hanns Lochmüller[2,9], Olaf Riess[5], Volker Straub[2], Manuel Posada[6,9], Domenica Taruscio[1,9] and Deborah Mascalzoni[7,8,9]

European Journal of Human Genetics (2016), 1–6

www.nature.com/ejhg

ARTICLE

'You should at least ask'. The expectations, hopes and fears of rare disease patients on large-scale data and biomaterial sharing for genomics research

Pauline McCormack*[,1], Anna Kole[2], Sabina Gainotti[3], Deborah Mascalzoni[4], Caron Molster[5], Hanns Lochmüller[6] and Simon Woods[1]

PAC MAIN ACHIEVEMENTS – REGISTRIES (WP2) AND BIOBANKS (WP3)

Integration in registry activities with WP2:

- Review and active input in recommendation to improve quality for RD registries (Poster and Paper with PAC members as co-authors Kodra Y. et al (2018)
- Presentation and dissemination at national and European workshops (registry workshop for patient groups, findacure UK; CHAFEA/Ciberer registry workshop; ISS registry Summer School)

Integration in Biobank activities with WP3:

- PAC involved in Biobank Assessment Panel
- EURORDIS is a member of BBMRI Stakeholders forum and liaise and feedback activities and discussions between the different projects and infrastructures

RD-CONNECT ASSETS AND OUTPUTS

A number of RD-Connect tools and resources have been given IRDiRC Recognized Resources label:

- The RD-Connect Genome Phenome Analysis Platform (4,160 data sets)
  - *Sample Catalogue 24,857 biosamples covering 112 RDs*
  - *Registry & biobank finder 382 patient registries and biobanks covering 1500 RDs*
- The FAIR Guiding Principles document for scientific data management and stewardship
- International Charter of Principles for sharing Bio-Specimens and Data
- Guidelines for the informed consent process in international Rare Disease Research

The Data Access Committee reviews requests to grant autorisation to the GPAP and includes EURORDIS as a member

The RD-Connect consortium made the decision in May 2017 to continue the RD-Connect brand and community (thank you Alexandre!) as well as to seek funding to support the individual assets

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SOLVE-RD

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SOLVE-RD IN BRIEF (2018-2022)

Solve-RD aims to find a diagnosis for rare disease patients who did not get a molecular diagnosis so far (beyond the exome).

- **Solve large numbers of rare disease, for which a molecular cause is not known yet by sophisticated combined omics approaches**
- **4 core ERNs (NMD, RND, ITHACA and GENTURIS)**
- **Use of the RD-Connect GPAP**
- **27 partners and 7 Work Packages** (phenotypes, molecular causes, **translation**, bioinformatics, **dissemination & impact**, coordination, **ethics**) **– 15M euros**

THE COMMUNITY ENGAGEMENT TASK FORCE PATIENT ENGAGEMENT IN SOLVE-RD

The CETF aims to embed the patient voice in all stages of the project, supporting appropriate patient involvement in the project and for any diseases still unsolved at the end of the project, leave a legacy of a strengthened support community for the undiagnosed community.

- Create a **united and engaged multi-stakeholder community** of patients, scientists and clinicians **committed** to improving diagnosis and care of **ultra-rare diseases** and **supporting** the needs of the **undiagnosed community**.
- Ensure that the **patient voice is heard and represented** in all stages of the project, by i) acting as a point of reference for patient voice across the Solve-RD project and ii) providing a 'critical friend' function to those engaged in delivering the project;

THE COMMUNITY ENGAGEMENT TASK FORCE PATIENT ENGAGEMENT IN SOLVE-RD

- **Demonstrate the added value of patient involvement** by bringing useful and impactful input in specific areas of Solve-RD to be identified by the CETF
- **Support and facilitate engagement of stakeholders within, and across**, initiatives and networks in the field of diagnosis at European and international levels (SWAN Europe, UDNI, ERNs)
- **16 confirmed members (project partners, UDNI, SWAN Europe, ePAG rep)**
- **Teleconference every 3 months (first one next week) and 3 F2F meetings (first one scheduled back to back with annual meeting in February 2019)**

✓ Terms of Reference - finalised

✓ Action Plan – in progress

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EURORDIS WINTER SCHOOL: PATIENT EMPOWEREMENT IN SOLVE-RD AND BEYOND

- Updated programme according to feedback of first edition
- 135 applications – 30 selected and invited to attend
- Pre-training initiated and evaluation quizzes developed and tested

Next edition: 11-15 March 2019 at Imagine Institute (Solve-RD)

Budget secured through EJP for 4 years (2020-2023)

EURORDIS WINTER SCHOOL

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EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD)

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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.

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

24 ERNs

EJP RD STRUCTURE

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

COORDINATION & TRANSVERSAL ACTIVITIES

INTEGRATIVE RESEARCH STRATEGY

SUSTAINABILITY

ETHICAL & REGULATORY

COMMUNICATION

1 FUNDING

2 COORDINATED ACCESS TO DATA & SERVICES

3 CAPACITY BUIDLING & EMPOWERMENT

4 ACCELERATING TRANSLATION OF RESEARCH & THERAPY DEVELOPMENT

PILLAR 1: COLLABORATIVE RESEARCH FUNDING

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

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

- 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

BBMRI-ERIC gateway for health

orphanet

INFRAFRONTIER mouse disease models

Joint Research Centre JRC

RD Connect

elixir

European Reference Networks

PILLAR 3: CAPACITY BUILDING AND EMPOWERMENT

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

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**

European Reference Networks

TEDDY NETWORK
European Network of Excellence for Paediatric Clinical Research

FONDATION maladies rares

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UNIVERSITY

Telethon

orphanet

Joint Research Centre JRC

RD Connect

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)

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

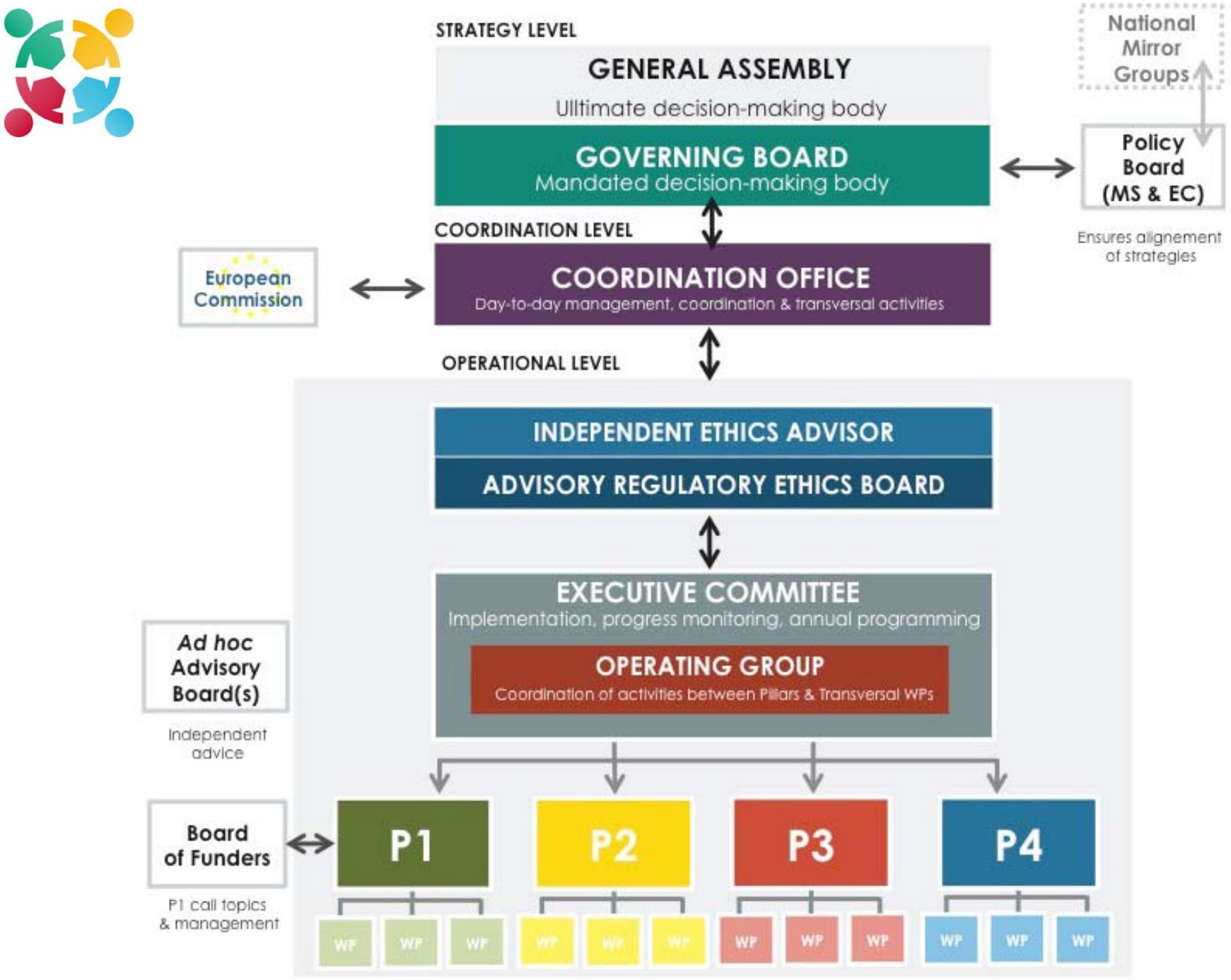
eatris

COORDINATION & TRANSVERSAL ACTIVITIES

COORDINATION & TRANSVERSAL ACTIVITIES

- PROGRAMME MANAGEMENT & COORDINATION
- INTEGRATIVE RESEARCH & INNOVATION STRATEGY
- SUSTAINABILITY
- ETHICS, LEGAL, REGULATORY & IPR
- COMMUNICATION & DISSEMINATION

GOVERNANCE


STRATEGY LEVEL
GENERAL ASSEMBLY
Ulltimate decision-making body
National Mirror Groups
GOVERNING BOARD
Mandated decision-making body
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
P1
P2
P3
P4
WP

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.