

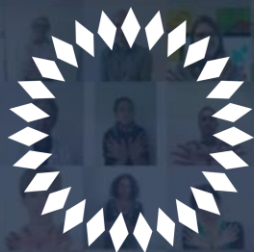
Rare Diseases Partnership

—
improving R&I potential and accelerating clinical trial
readiness of the rare diseases

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INSERM, France



EU2022.CZ

Czech Presidency of the Council
of the European Union

Expert Conference on Rare Diseases
Towards a New European Policy Framework:
Building the future together for rare diseases
25-26 of October 2022, Prague, Czech Republic



STRATEGY

International Rare Diseases Research Consortium (IRDiRC), EC, Member States

INFRASTRUCTURES

Orphanet, RD Connect, ERDRI, Solve-RD, EATRIS, ECRIN, BBMRI, etc.

FUNDING

ERA-Net Rare (2006-2018) + EC + industry

PATIENTS NEEDS

EURORDIS

HEALTHCARE +

European Reference Networks (ERN)

RESEARCH ECOSYSTEM

EJP RD



WHAT ARE THE REMAINING NEEDS?



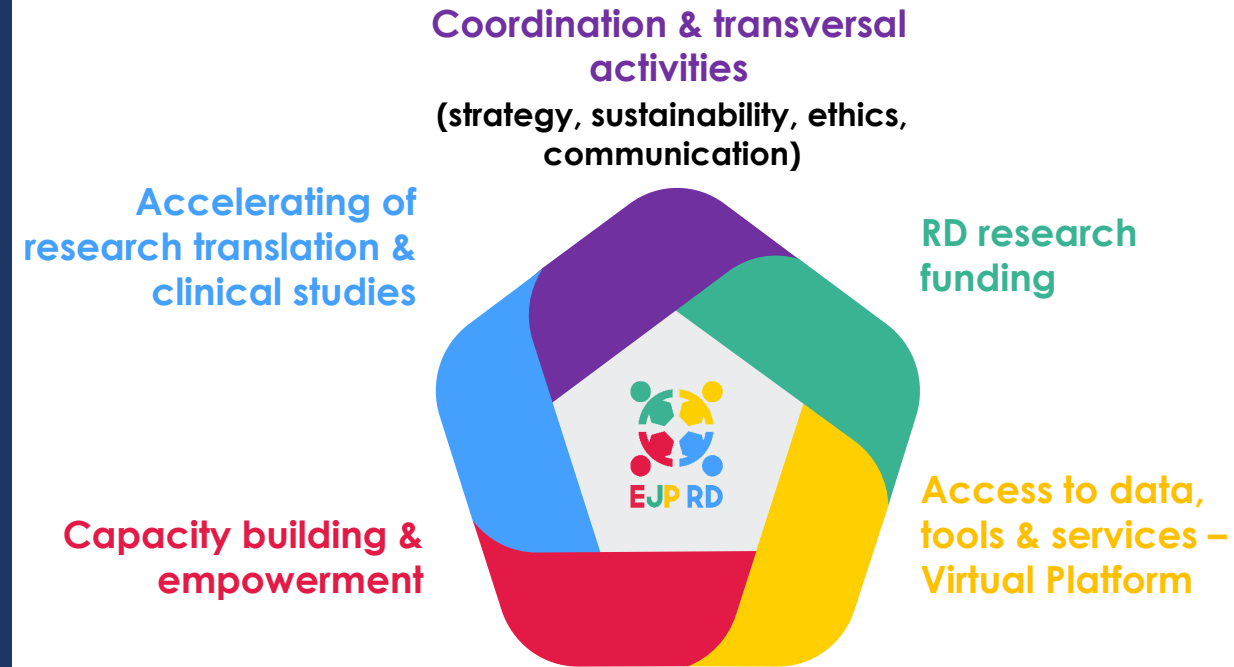
European Joint Programme on Rare Diseases – objectives & structure

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

Mode of action:

Large programme that integrates existing infrastructures, trainings, funding programmes and tools, expands them and develops new essential ones to offer harmonized (and centralized) RD research ecosystem that is easy to use for scientists and produces benefits for patients in the most efficient way





HOW EJP RD EMPOWERS STAKEHOLDERS & PERFORMANCE OF RESEARCH ACTIVITIES?



Networking support to build ideas, engage patients, align needs

Profit from multinational funding opportunities



Follow on funding support

Clinical studies support office

Knowledge sharing



Use VP to find suitable tools (models, screening, biosamples, pathways for drug targets)

Benefit from expert mentoring support at application stage



Mentoring follow up

Training for PIs & students

Data FAIRification

Tools for phen-gen analysis





HOW EJP RD EMPOWERS STAKEHOLDERS & PERFORMANCE OF RESEARCH ACTIVITIES **FOR EARLY DIAGNOSIS?**



Joint transnational calls covering diagnosis related topics



Mentoring follow up
Follow on funding support for diagnostics tools, biomarkers, etc.




VP resources: biobanks, animal models, registries, RD-Connect
Tools for phen-gen analysis




Training for PIs & students
Data FAIRification
Biobanking
Quality assurance, variant interpretation in NGS
Undiagnosed diseases
Orphanet nomenclature




Remaining needs



95% of RDs are disregarded in terms of research and lack effective treatment options



50% of RD patients still do not have confirmed molecular diagnosis



4 years - is an average time to be diagnosed when RD is known

52% of RD patients and carers, RD translates into severe impact on their daily life



SUPPORT

robust patient need-led research



UTILIZE

the power of health and research data and spearhead the digital transformational change in RD research and innovation



Rare Diseases Partnership Vision



DEVELOP

new treatments and diagnostic pathways




SUPPORT

the coordination and alignment of national and regional research strategies, including the establishment of strong public-private collaborations



Rare Diseases Partnership Mission



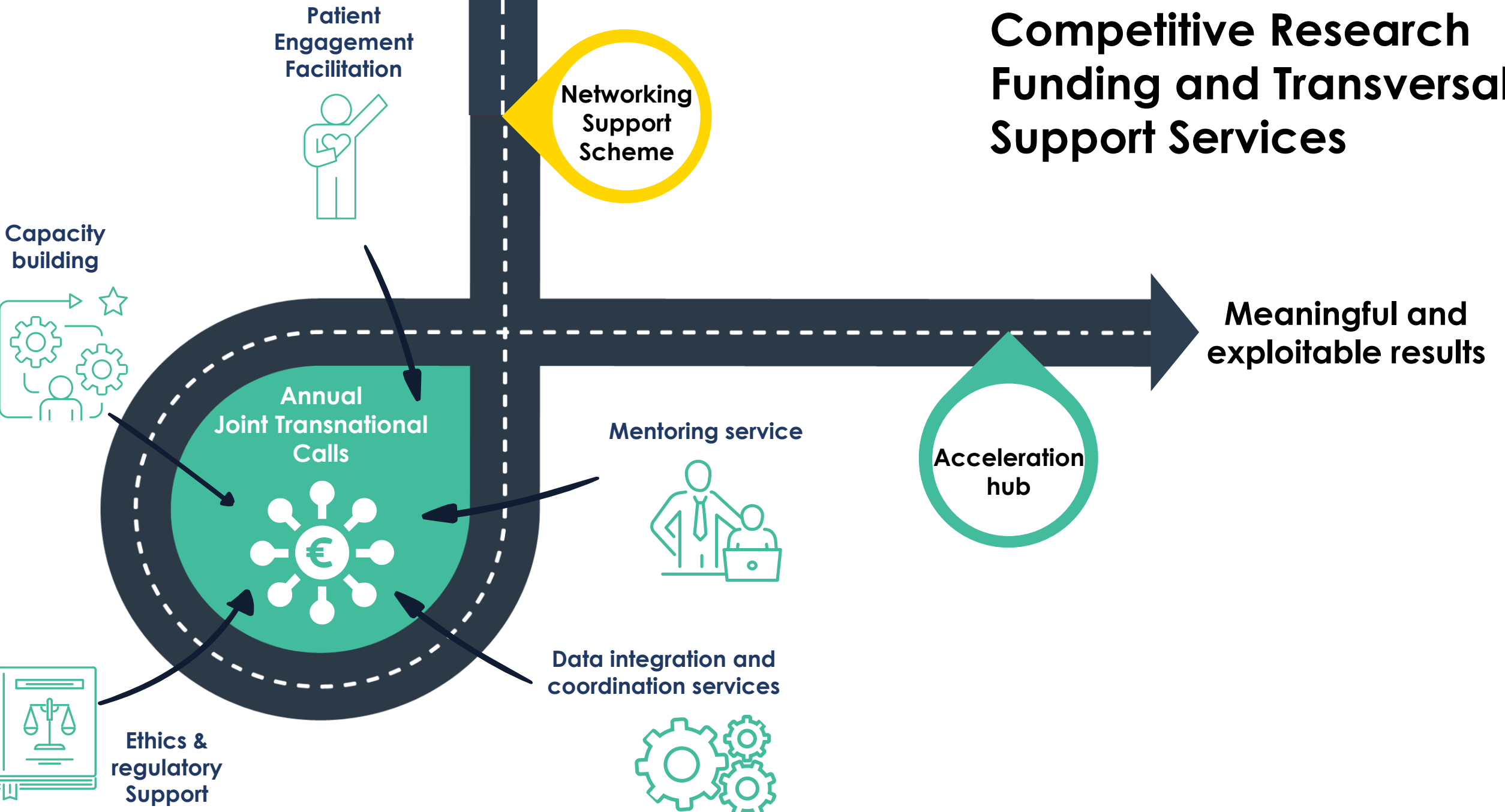
Bring supporting R&I services from across Europe under one roof so that every high-quality RD research project will benefit from cross-disciplinary expertise, goal-oriented study planning and efficient execution

Enable every consenting patient living with a rare disease to be findable and enrolled in a suitable clinical study, by boosting generation of regulatory-level and FAIR-compliant data from diversity of sources, with the ultimate goal to fasten advances in prevention, diagnosis, disease knowledge and treatment

Make Europe a global leader on rare disease research through a significant increase in investment to spur innovation, leading to job creation and improving EU competitiveness in R&I

Competitive Research Funding and Transversal Support Services

Meaningful and exploitable results



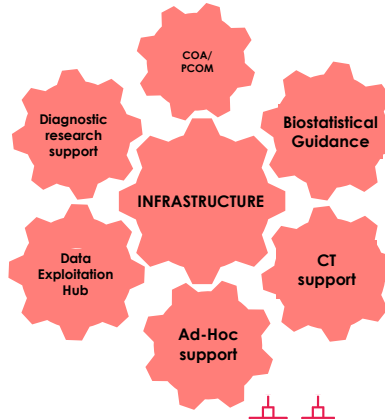
Clinical Research Network

National alignment & capacities



Innovative diagnostics
Natural History Studies
RWE generation
Identification & validation of PCOMs
Biomarkers & surrogate endpoints

Technological expertise



Infrastructure

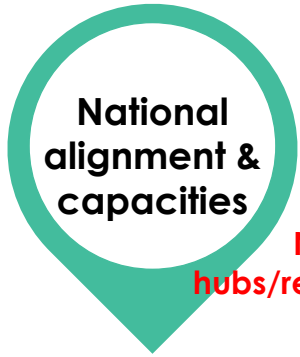
Public-private collaboration

Accelerated diagnosis
Clinical trial readiness
New therapies

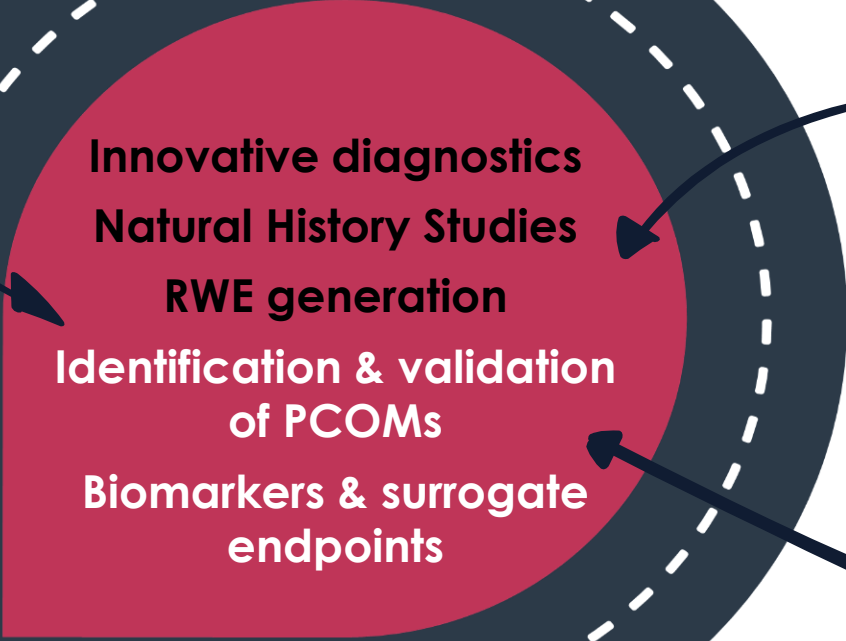
Clinical Research Network – contribution to early diagnosis

National experts centers

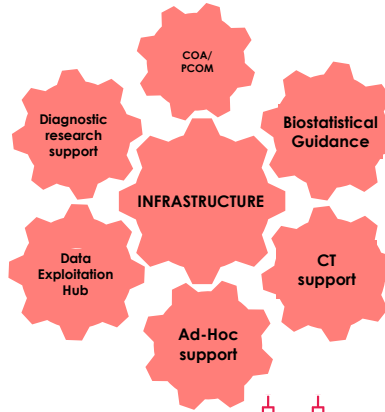
Clinical expertise &
capacities



National data
hubs/registries/omics data
sources



Technological
expertise



Infrastructure

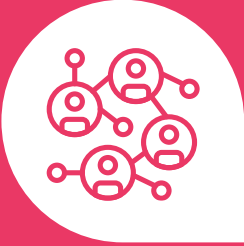
Collaboration with Solve-RD,
1+MG



Accelerated diagnosis
Clinical trial readiness
New therapies

EUROPEAN CLINICAL RESEARCH NETWORK FOR RARE DISEASES

*RD Clinical Research Infrastructure
Data exploitation hub
Diagnostic research support
COA/PCOMs support
Biostatistical guidance
Clinical trials support*



COMPETITIVE RESEARCH FUNDING AND SUPPORT

*Joint Transnational Calls,
Networking*

TRANSVERSAL SUPPORT SERVICE

*Mentoring service
Data integration
Capacity building
Ethics & regulatory support
Acceleration hub*



COORDINATION, STRATEGY, GOVERNANCE

*Public-private collaboration
Maximisation of national alignment & contributions
Joint multi-stakeholder strategy
Patients as drivers*



Rare Diseases Partnership

THANK YOU

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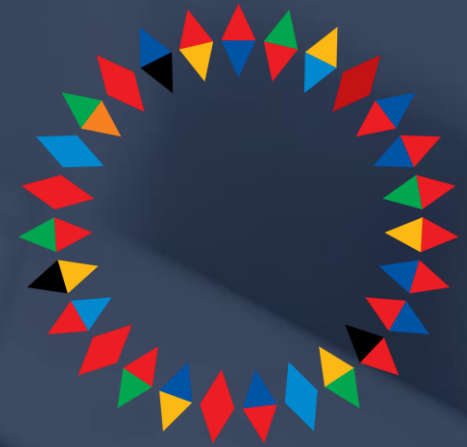
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