



ERICA

European Rare Disease Research
Coordination and Support Action



Amsterdam UMC
Universitair Medische Centra



Endo-ERN

European Reference Network
on Rare Endocrine Conditions



European
Reference
Networks

#ERNcare4Ua

Rare Diseases Doctors

Session 3: The Way Ahead. *ERDERA: Research funding – opportunities for ERNs*

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The Netherlands



Rare Diseases in the EU: Joint Action shaping the future of ERNs

JARDIN kick-off meeting

8 March 2024, Brussels



ERICA has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 964908.



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ERICA (in which all 24 ERN's take part) has been established to strengthening ERN's research and innovation capacity, through:

- facilitating collaboration between ERNs
- increase the visibility and impact of ERNs
- integration of the results into the new European Rare Disease Alliance (ERDERA) 2024-2034





EUROPEAN RARE DISEASES RESEARCH ALLIANCE

2024 – 2034



**Co-funded by
the European Union**

ERDERA proposal **was approved under Horizon Europe** on 25/01/2024 for funding under the EU Research & Innovation funding programme [Horizon Europe](#) as a co-funded partnership between the European Commission, European Member States, and beyond.

ERDERA

SUPPORT

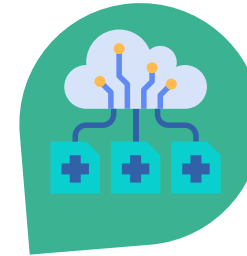
robust patient need-led research



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UTILIZE

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



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ERDERA

Rare Diseases
Partnership
Vision

DEVELOP

new treatments and diagnostic pathways



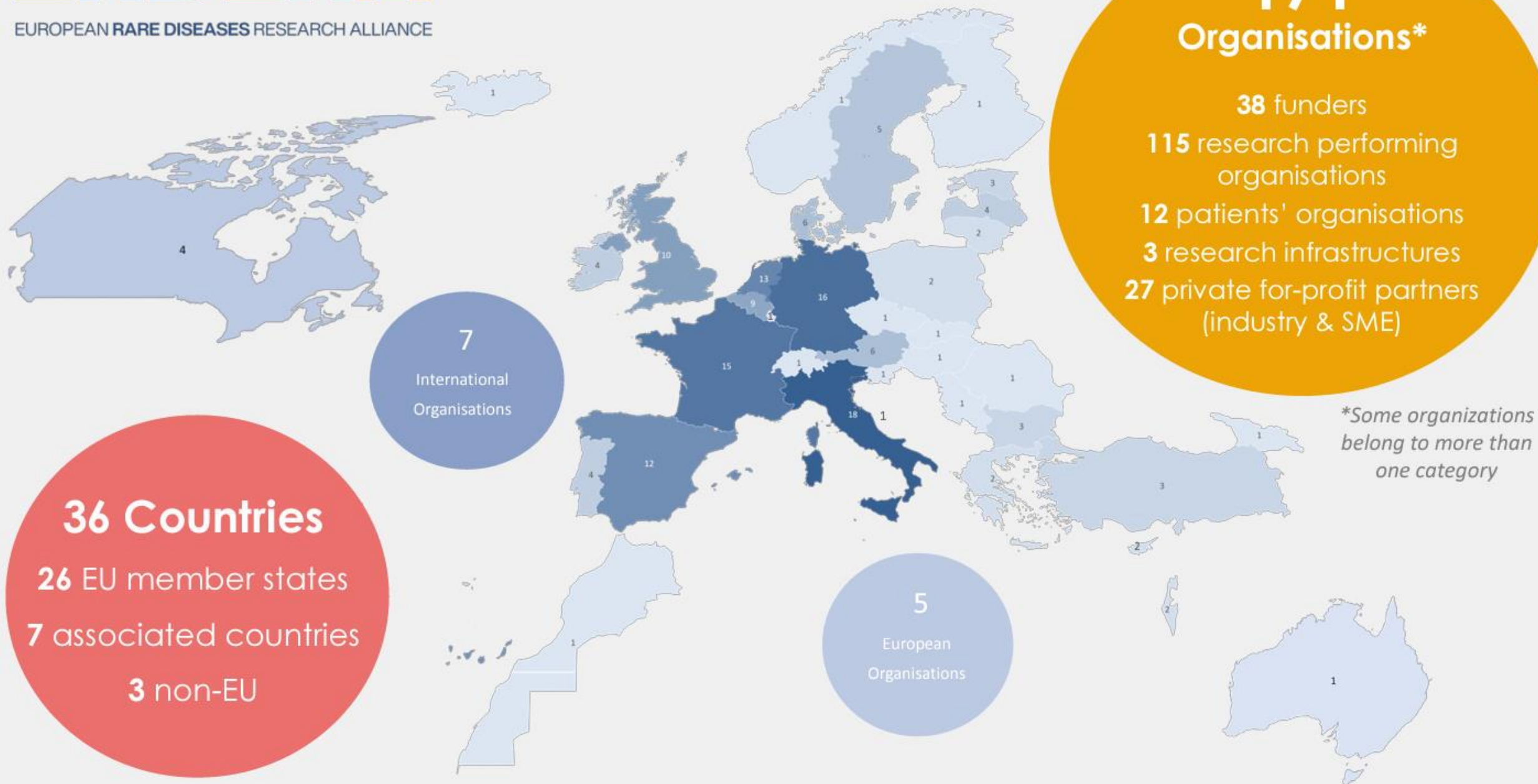
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SUPPORT

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



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Coordination & Strategy

WP1

Coordination and management

WP2

Communication & dissemination

RD Funding

WP3

Joint Transnational Calls for collaborative research projects

WP4

Clinical trial call management

WP5

Networking to share knowledge on research

Clinical Research Network

WP6

Diagnostic data availability

WP7

Genome re-analysis research pipeline

WP8

Innovation to shorten time to RD diagnosis

WP9

Real world data

WP10

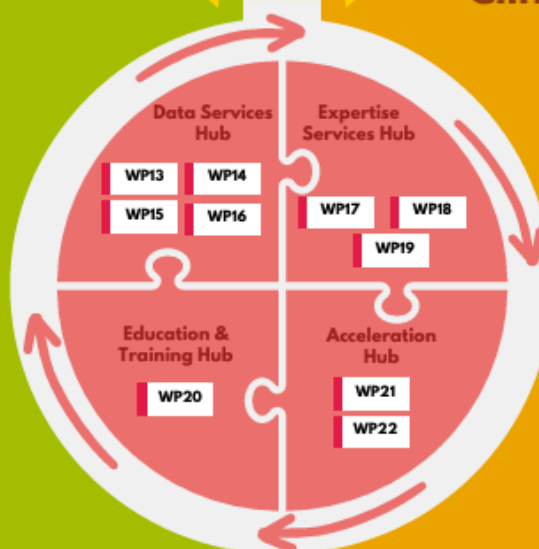
Clinical Outcome Assessment

WP11

Advanced Therapeutic Medicinal Products

WP12

N-of-few approach



Inter(national) Capacity Alignment

WP23

NMGs promotion and national alignment

WP24

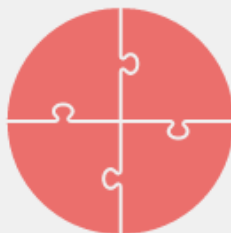
Fostering engagement of underrepresented countries in ERDERA

WP25

ERDERA Global Collaboration



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WP13

Rare Diseases-Virtual Platform (RD-VP): Finding and accessing the data ecosystem

WP14

Data readiness services

WP15

Data sharing and analysis services

WP16

Knowledge bases and ontologies for RD research

WP17

Mentoring and consultancy

WP18

Regulatory support service

WP19

Methodological Support

WP20

Education and training in rare diseases research

WP21

Technology accelerator

WP22

Public-Private Collaboration Accelerator

RD funding

Joint transnational calls for collaborative research projects

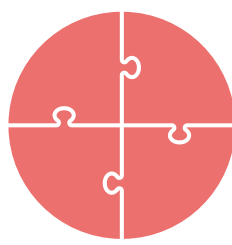
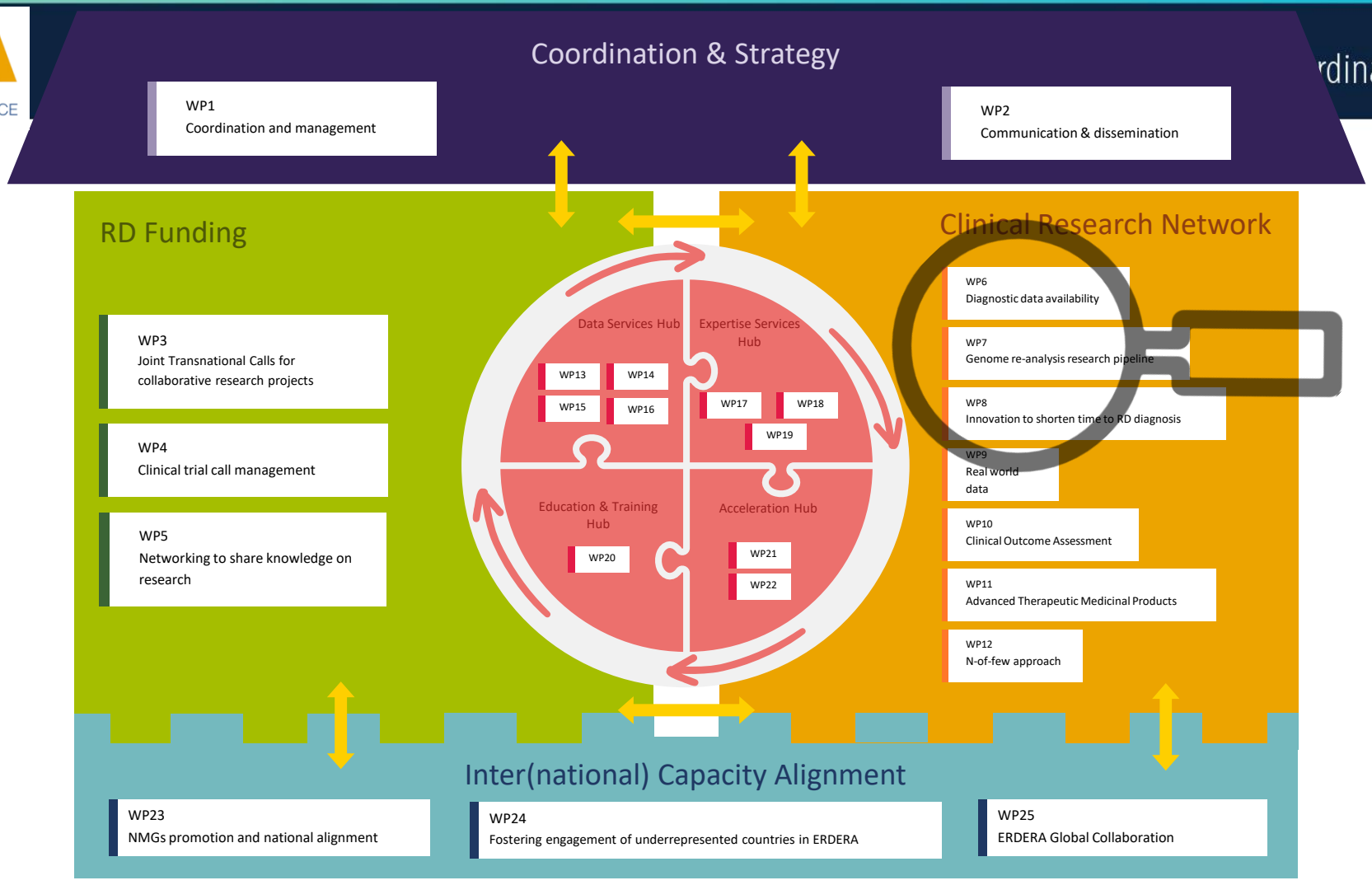
- **Tasks 3.1** – Topics selection for the JTCs and Clinical Trial Call
- **Task 3.2** – Joint Transnational Call implementation
- **Task 3.3** – Working group on patient engagement in research project funding

Clinical trial call management

- **Tasks 4.1** – Develop the call framework
- **Task 4.2** – Open the call and select trials for funding
- **Task 4.3** – Project implementation, project monitoring and financial management

Networking to share knowledge

- **Tasks 5.1** – Preparation and launching of the funding scheme
- **Task 5.2** – Evaluation of the selected proposals after each collection date
- **Task 5.3** – Quality management



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|---|---|---|
| <p>WP13
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Technology accelerator</p> |
| | | <p>WP22
Public-Private Collaboration Accelerator</p> |

CLINICAL RESEARCH NETWORK _ Diagnostic research

Data Readiness

- **Task 6.1** – Coordinate Pan-European diagnostic research data readiness and collation effort
- **Task 6.2** – Data standardisation, submission and harmonisation
- **Task 6.3** – Data archival & data access

Genome re-analysis research pipeline

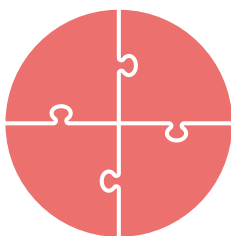
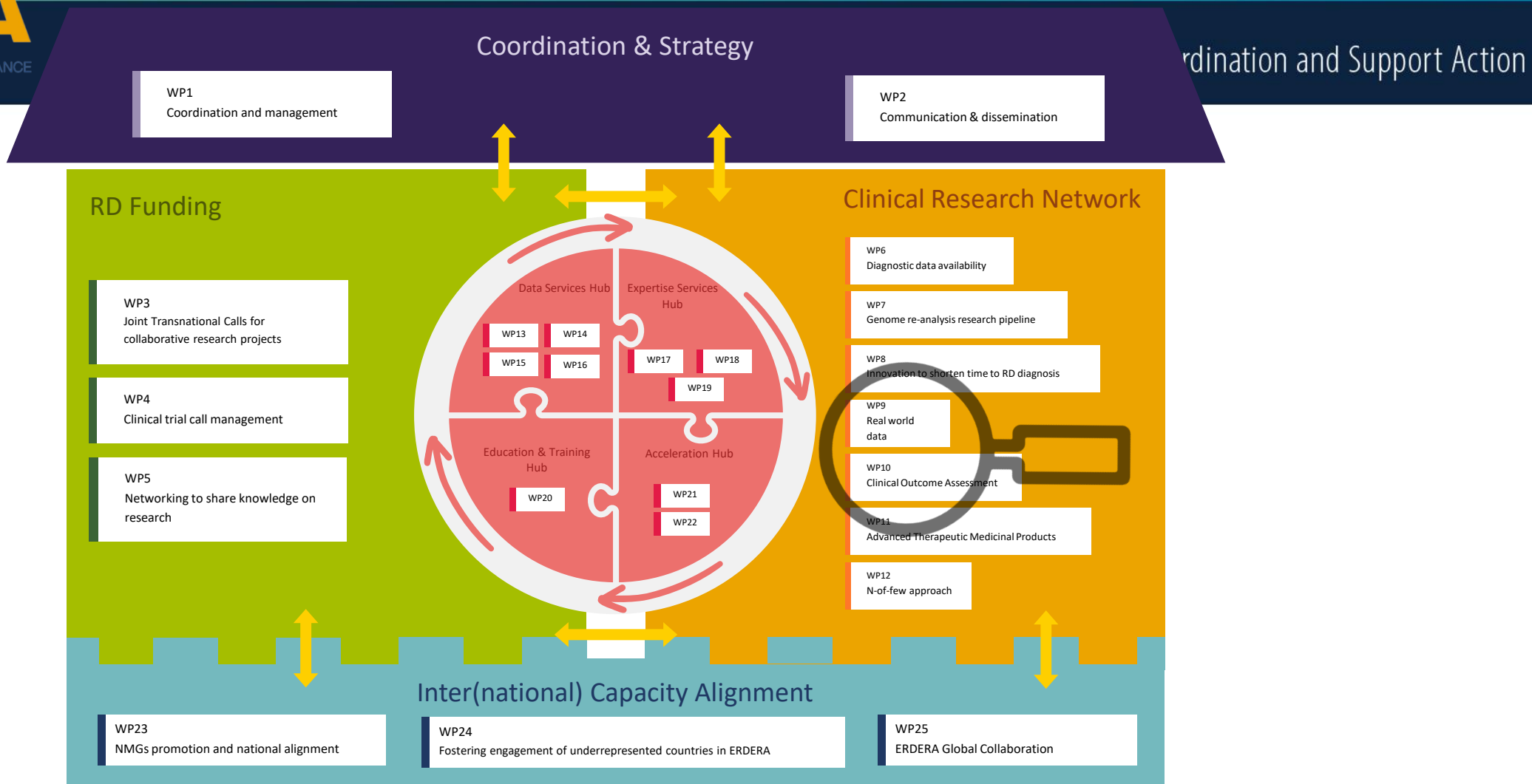
- **Task 7.1** – Exome and genome re-analysis pipeline coordination and monitoring
- **Task 7.2** – Standardised exome and genome re-analysis beyond state-of-the-art diagnostics
- **Task 7.3** – Develop and leverage knowledge for variant interpretation
- **Task 7.4** – Translation to clinic

Genomic innovation to shorten time to diagnosis

- **Task 8.1** – Enable access of complete genome sequencing for RD in underrepresented countries
- **Task 8.2** – Enable complete genome sequencing and analysis for RD to shorten time to diagnosis
- **Task 8.3** – Complete genome mapping for RD to shorten time to diagnosis
- **Task 8.4** – New genomics/transcriptomics analysis capabilities to understand genetic variation in RD
- **Task 8.5** – Multi-omics data integration to shorten time to diagnosis in RD



ALL Other ERNs
(in addition to Euro-NMD, GENTURIS, ITHACA, RND + EpiCare and RITA)



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CLINICAL RESEARCH NETWORK _ Outcome research

Real World Data

- **Task 9.1** – Use of primary healthcare data (EHRs) for RD outcome research
- **Task 9.2** – Use of population-based data for RD outcome research
- **Task 9.3** – Integrating patient cohorts for natural history / standard-of-care reference studies
- **Task 9.4** – Development of a blueprint and inventory of regulatory-grade natural history cohort data
- **Task 9.5** – Disease progression modelling and prognostic biomarker research
- **Task 9.6** – Development of a regulatory grade clinical trial simulation platform for rare diseases



T9.1: eUROGEN; EURO-NMD; CRANIO, EpiCare, EuroBloodNet;
T9.2: EpiCare; ERNICA; MetabERN; T9.3: ITHACA, ERKNet, ERNs ENDO-BOND;
T9.4: RND, EuroBloodNet, ERKNet, EYE; T9.5: EURO-NMD; T9.6: ERKnet; DDF

Clinical Outcome Assessment

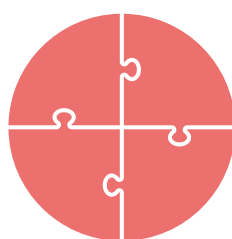
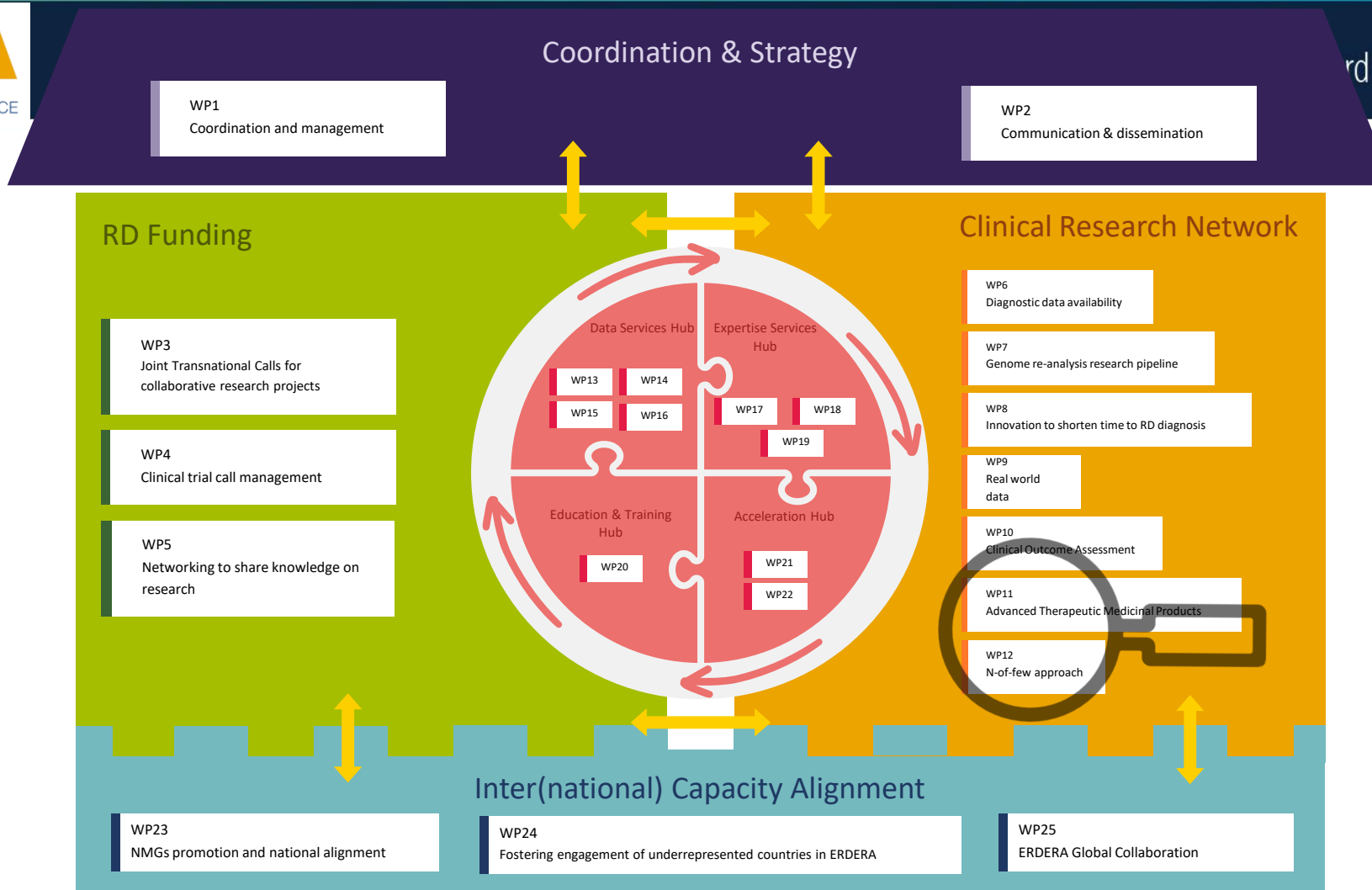
- **Task 10.1** – Platform for regulatory-grade patient-centred COA development and validation
- **Task 10.2** – Development and Implementation of Clinical Outcome Assessment Tools
- **Task 10.3** – Unveiling the Hidden Burden: Estimating the Socioeconomic Impact of Rare Diseases for Informed Decision Making and Resource Allocation

T10.1: RND; ITHACA; mito-InterERN workgroup (EURO-NMD, RND, MetabERN, Eye, EpiCare); EuroBloodNet; CRANIO;
T10.2: EpiCare; EURO-NMD; ERKNet; EuroBloodNet; ERN-RND

CRN: Outcome Research workstream _ Involvement of ERNs

	9.1	9.2	9.3	9.4	9.5	9.6	10.1	10.2
BOND			X					
CRANIO	X						X	
ENDO			X					
EpiCare	X	X					X [*]	X
ERKNet			X	X		X		X
ERNICA		X						
EURO-NMD / DDF	X				X	X	X [*]	X
RND				X	X		X [*]	X
EuroBloodNet	X			X	X		X	X
eUROGEN	X							
EYE				X			X [*]	
ITHACA			X				X	
MetabERN		X					X [*]	

* Mito-InterERN workgroup



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CLINICAL RESEARCH NETWORK _ Innovative therapies

Advanced Therapeutic Medicinal Products

- **Task 11.1** – Identify and rank disease indications requiring ATMPs
- **Task 11.2** – Select and adapt the technical platforms with prioritised need
- **Task 11.3** – Design the proof of concept studies to evaluate the selected pipelines
- **Task 11.4** – Evaluate the selected platforms for clinical trials requirement and joint transnational call

N-of-few approach

- **12.1** – Academic Platform for Tailored Antisense Oligonucleotide Therapies
- **12.2** – Identification of patient relevant-outcomes (n-of-1/few) and run in natural history study
- **12.1** – Treatment/study design and analysis
- **12.1** – Implementation of first in human treatment infrastructure
- **12.1** – Case studies

(Inter)national capacity alignment

National Mirror Groups promotion & national alignment

- **Task 23.1** – Fostering creation of National Mirror Groups
- **Task 23.2** – Deployment and operations of National Mirror Groups
- **Task 23.3** – Animation of National Mirror Groups synergies


Fostering engagement of underrepresented countries

- **Task 24.1** – Promoting capacity development actions
- **Task 24.2** – Undertaking advocacy and awareness efforts to UCs added value
- **Task 24.3** – Support actions to improve UC participation in all RDP activities

RDP Global collaboration

- **Task 25.1** – Strategic Alliances
- **Task 25.2** – Support to IRDiRC Scientific Secretariat
- **Task 25.3** – Promote the International Dimension of the Clinical Research Network of RDP by building global networks among clinical research networks, universities, and patient organizations.
- **Task 25.4 – Alignment with the ERNs Research strategy** 

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Goals: global collaboration

- To ensure that the **ERN-CG represent a dissemination point for the benefit of ERN HCPs members about the activities performed inside ERDERA**, and, vice versa, it will provide the RDP with advices on the research priorities distilled from thematic workshops aimed at the identification of potential solutions to patients' unmet needs.
- To facilitate the **Collaboration with the Joint Action** for the Integration of ERNs into National Health Systems (NHS) to enforce and highlight research activities as a driving force to improve patient care and outcomes through innovation.
- This Task aims at ensuring the **alignment of the JARDIN** program with the ERNs activities and strategies.
- To this aim, this task will contribute by proposing the ERN-CG as advisory body in order to facilitate that ERNs have a common voice in the JARDIN implementing actions. The JARDIN subtask will be lead by Dr. Luca Sangiorgi IOR-ERN BOND

Thank you very much for your attention!

