

European Reference Networks Research Opportunities

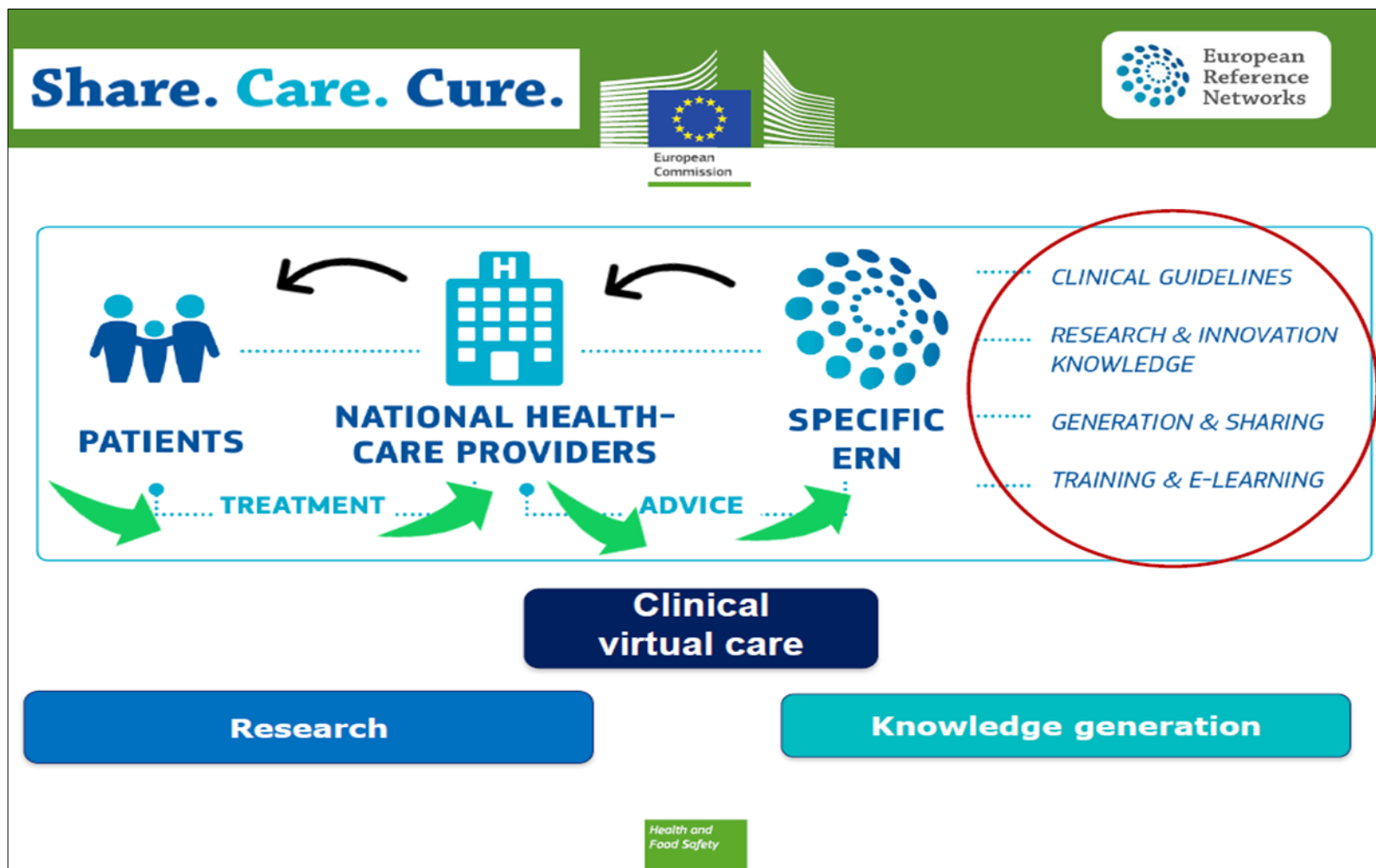
Prof Eileen Treacy

Clinical Lead, National Rare Diseases Office

February 23, 2023



Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive



WHICH DISEASES ARE CURRENTLY COVERED BY THE ERNS?

Each of the **24 ERNs** address a specific area of intervention, though they also often work together.

- **ERN BOND**: bone disorders
- **ERN CRANIO**: craniofacial anomalies and ear, nose and throat (ENT) disorders
- **Endo-ERN**: endocrine conditions
- **ERN EpiCARE**: epilepsies
- **ERKNet**: kidney diseases
- **ERN-RND**: neurological diseases
- **ERNICA**: inherited and congenital anomalies
- **ERN LUNG**: respiratory diseases
- **ERN Skin**: skin disorders
- **ERN EURACAN**: adult cancers (solid tumours)
- **ERN EuroBloodNet**: oncological and non-oncological hematological diseases
- **ERN eUROGEN**: urogenital diseases
- **ERN EURO-NMD**: neuromuscular diseases
- **ERN EYE**: eye diseases
- **ERN GENTURIS**: genetic tumour risk syndromes
- **ERN GUARD-HEART**: diseases of the heart
- **ERN ITHACA**: congenital malformations and rare intellectual disability
- **MetabERN**: hereditary metabolic disorders
- **ERN PaedCan**: paediatric cancer
- **ERN RARE-LIVER**: hepatological diseases
- **ERN ReCONNET**: connective tissue and musculoskeletal diseases
- **ERN RITA**: immunodeficiency, autoinflammatory and autoimmune diseases
- **ERN TRANSPLANT-CHILD**: conditions and complications linked to the transplantation in children
- **VASCERN**: rare multisystemic vascular diseases

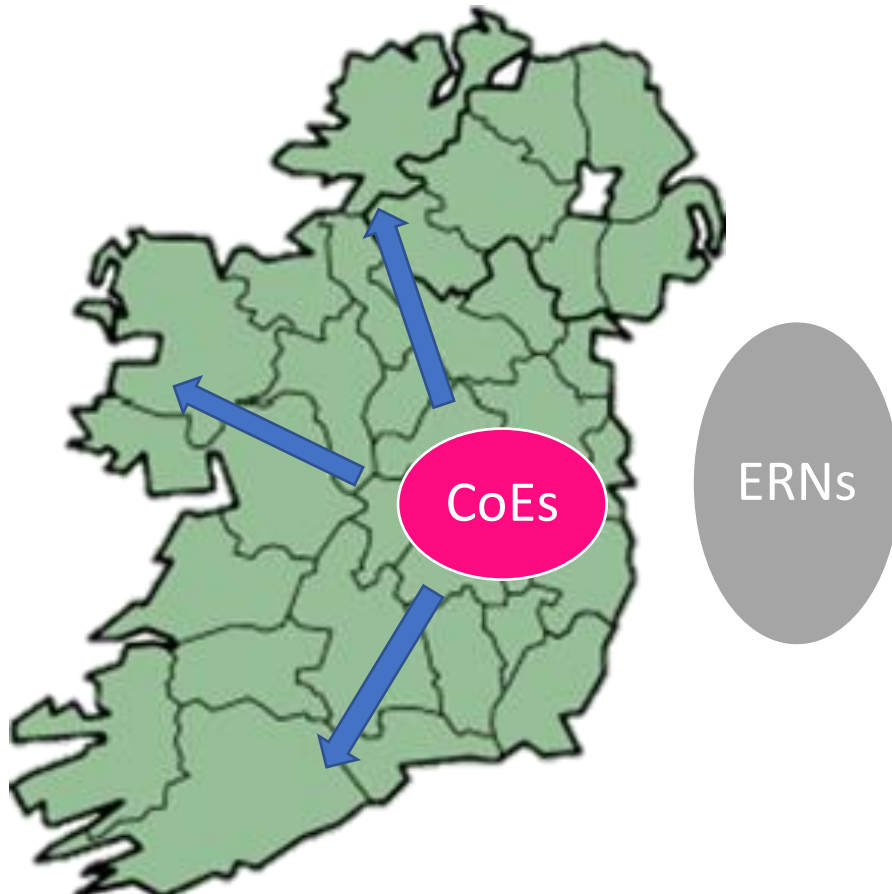


Ireland- Integration to ERNs



European
Reference
Networks

Hub and Spoke

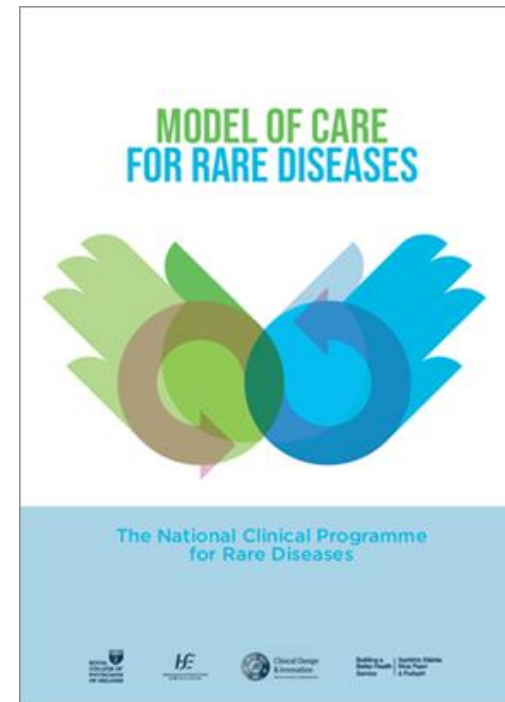
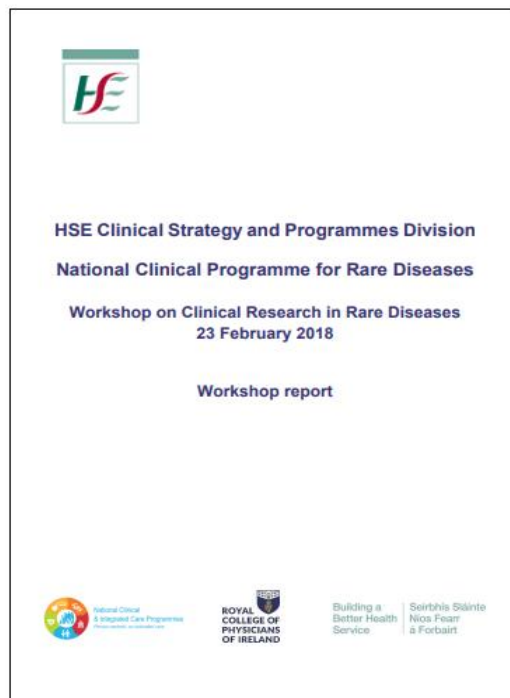


Irish Networks
NRDO-Coordination 'Hub'
(www.rarediseases.ie)

113 Expert Clinics
5 Academic Teaching Hospitals
Universities:
UCD, Trinity College, RCSI



ERNs, EJPRD, RD Partnership and Clinical Research



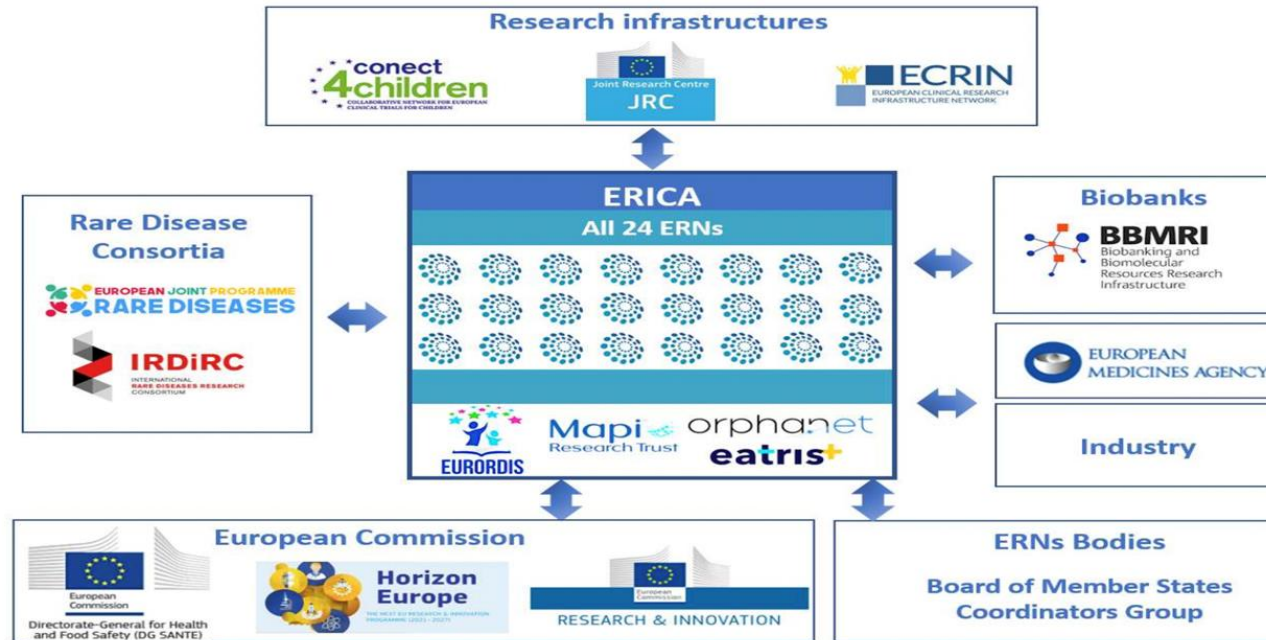
HRB: *“Ireland’s weaknesses include lack of patient registries and the lack of protected time for RD research for clinicians”*

“I am a clinician by choice and a researcher by necessity”- Aaron Ciechanover- Noble Institute



European Rare Disease Research Coordination and Support Action

Positioning of ERICA within the RD research ecosystem



<https://erica-rd.eu/>

Objectives

To record and disseminate ERN research activities

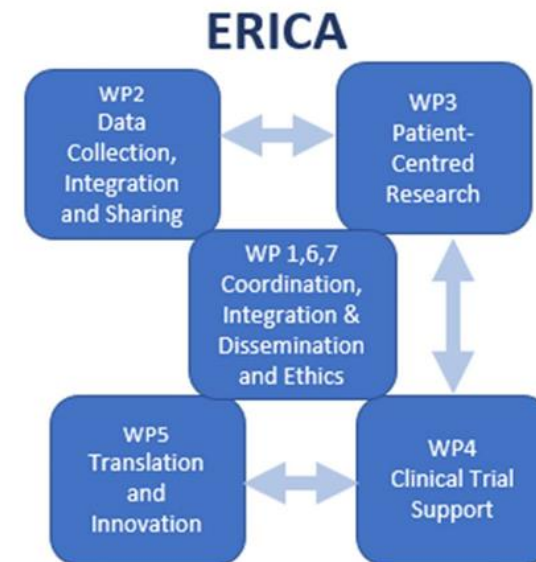
To facilitate collection sharing/analysis of research data between ERNs

To advance patient-centered research and outcomes monitoring

To map, integrate and link ongoing activities to facilitate international clinical trials

To encourage methodology sharing and innovation of ERNs

Work Packages



ERICA Repository in a nutshell:

D3.1 Central Repository of (validated) PCOMs for RDs August 2022

- Free of access (<https://erica-rd.eu/work-packages/patient-centred-research/proms-repository/>)
 - 811 unique PROMs/PCOMs:
 - ERN related to the PROMs/PCOM's population of development and validation
 - PROMs/PCOMs PROQOLID™ database page for additional information

PROMs Repository

The ERICA Patient Reported Outcome Measures (PROMs) Repository is the first attempt to identify and centralize Clinical Assessment Outcomes questionnaires of relevance for rare diseases and constitutes a milestone in the Europe-wide standardization of Patient-Centered Outcome Measures (PCOMs) and PROMs for rare diseases. It has been made possible through the joint collaboration between Orphanet, Mapi Research Trust/ICON and ERN EuroBloodNet (VHIR, APHP), and the active contribution of ERNs and ePAGs. The methodology for the constitution and future evolution of the repository can be found in [deliverable 3.1](#) (849 KB) and [deliverable 3.2](#) (661 KB).

The central repository is a centralized and standardized access gate to more in depth information contained in PROQOLID™.

Full-access requests for ERICA partners: info@erica-rd.eu

Filters list of PCOMs/PROMs

PCOM/PROM Name ¹⁾	<input type="text"/>	PCOM/PROM Type ²⁾	<input type="text"/>
Target Age ⁴⁾	<input type="text"/>	Domains ⁵⁾	<input type="text"/>
Disease (OrphaName) ⁶⁾	<input type="text"/>	OrphaCode ⁷⁾	<input type="text"/>
Group of Diseases ⁸⁾	<input type="text"/>	ERNs ⁹⁾	<input type="text"/>

[Legend](#)

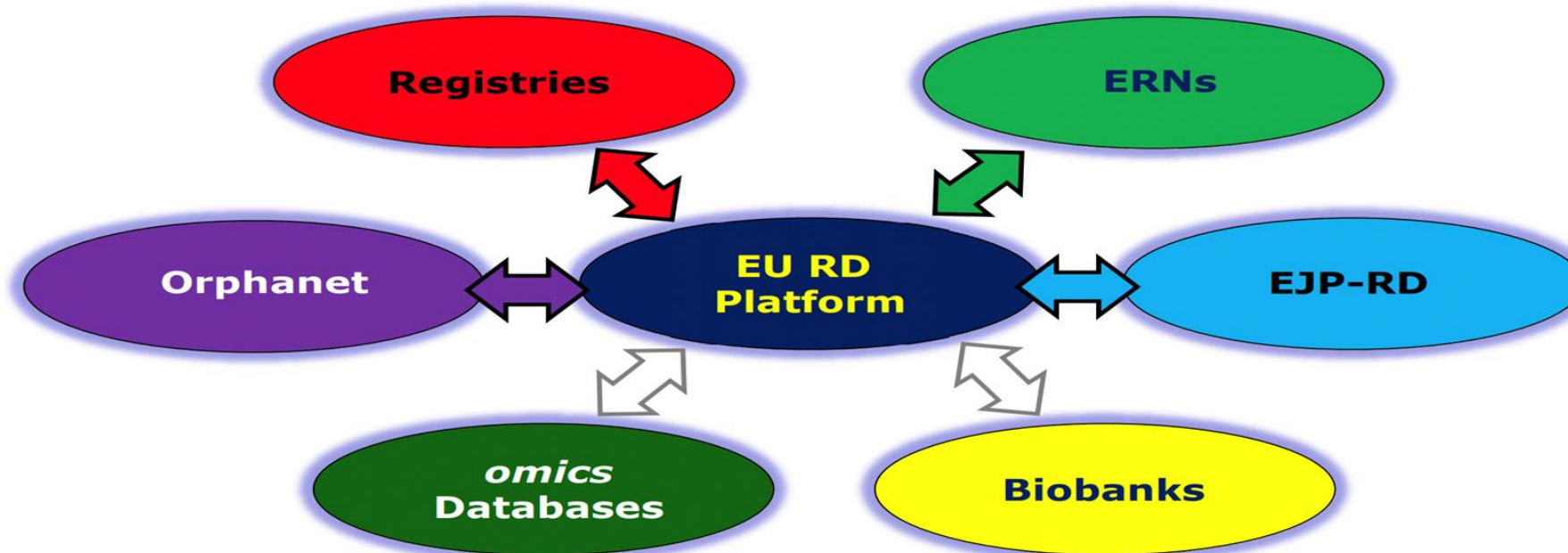
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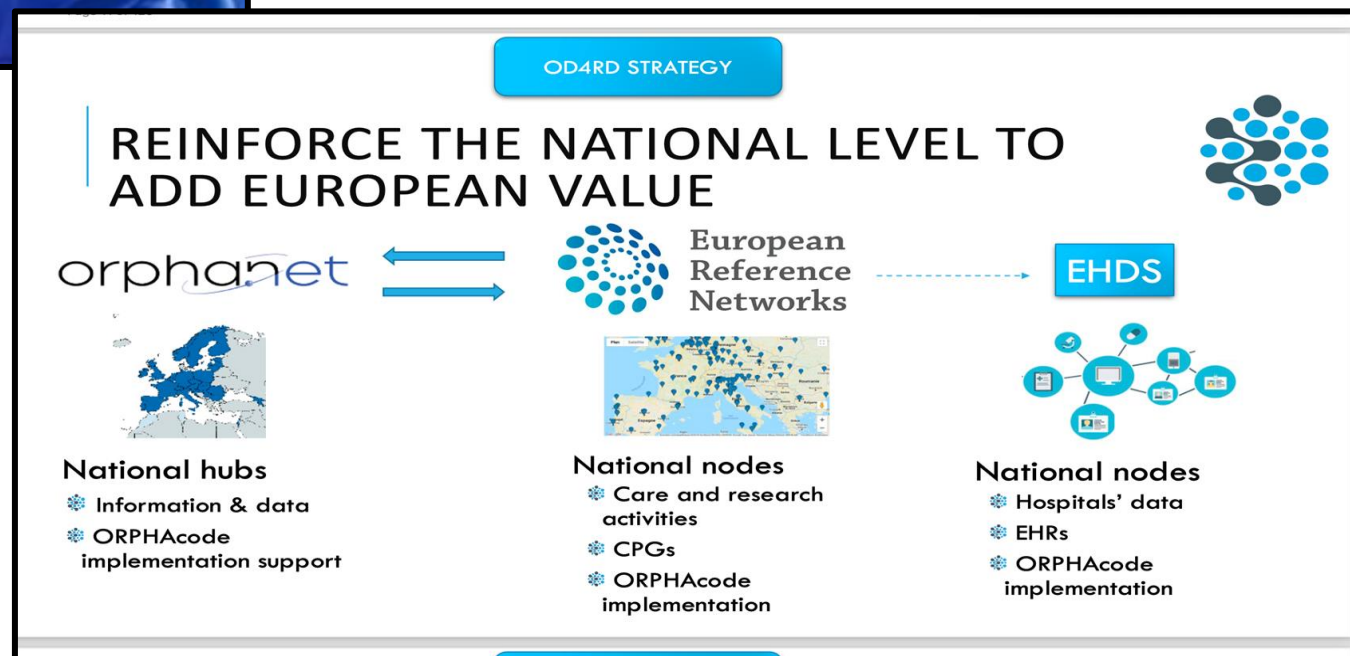
Showing 811 PCOMs/PROMs

EU RD Platform - interactions



EU4Health programme
for a healthier and safer Union

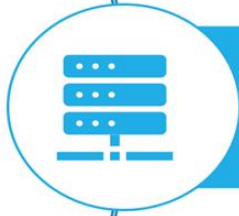
#EUBudget #EU4Health



OD4RD GENERAL OBJECTIVES



To contribute to the generation of standardised, interoperable data on RD diagnosis for primary and secondary use, by the maintenance and the support to the implementation of the Orphanet nomenclature of RD



To contribute to the harmonization of data collection amongst settings (health records, registries) and amongst countries, by the dissemination of coding good practices at the data source level



To contribute at supporting evidence-based decision-making in the frame of the European strategy around ERNs, by supporting the exploitation of reference corpus of data and information on RD



2022 EU4Health Work Programme



- **HS-g-16.02-22. Direct grant to Member States to support ERNs integration to the national healthcare systems of Member States**

This action supports the integration of the ERNs into the national healthcare systems that will ensure long term sustainability of the ERN system, enable the Member States to strengthen the resilience of their national health system and improve accessibility of the ERN system to rare disease patients and their health professionals at national, regional, and local level with the aim to enable access to timely diagnosis and appropriate treatment



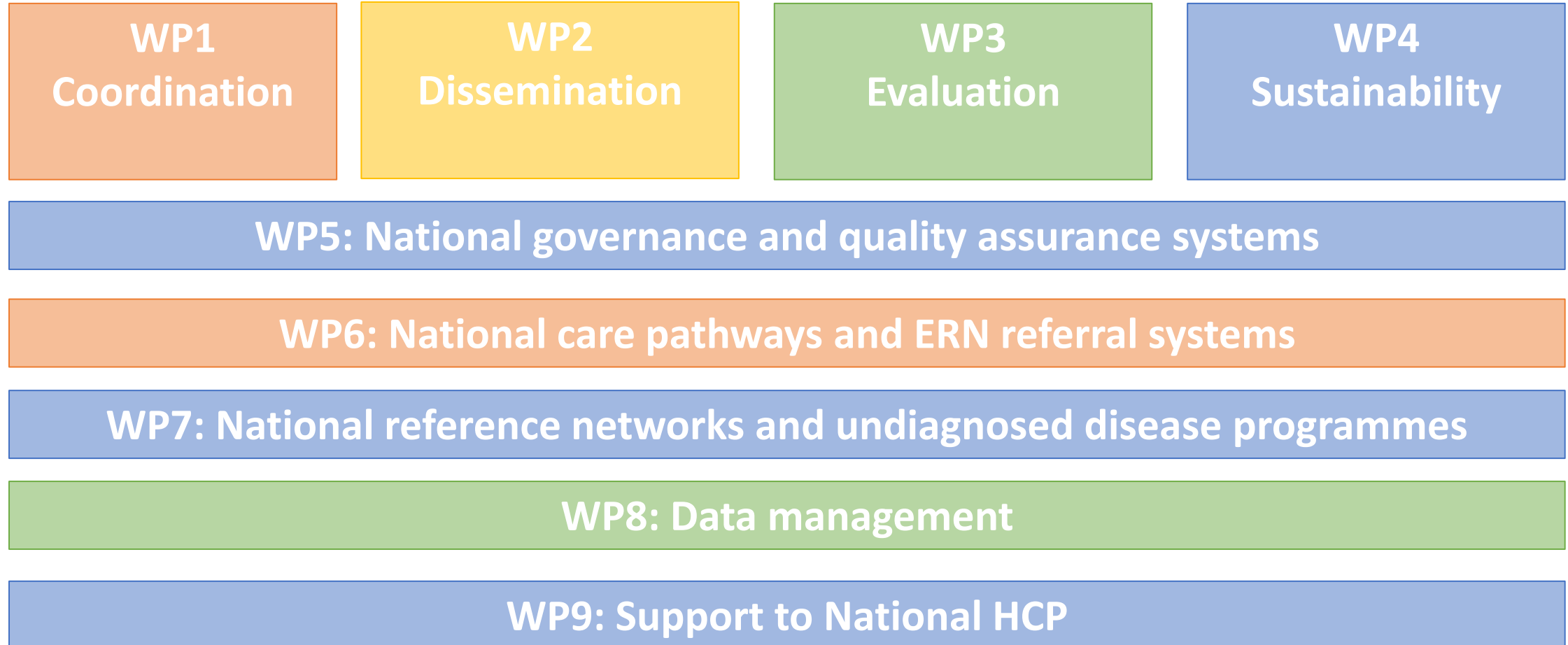
Expected Results and Impact



- Exchange of best practices
- Create proposals, guidelines, models and recommendations for better integration of ERNs in national healthcare systems, *including well defined patient pathways and referral procedures*
- Development of national networks for rare diseases
- Guidelines for development of national teleconsultation tools interoperable with the ERN CPMs



JA-05: Support ERNs integration to the national healthcare systems of MS





HSE – National Competent Authority

Key Work Packages



WP6: National care pathways and ERN referral systems

WP7: National reference networks and undiagnosed disease programmes

In collaboration with Eurordis





Thank you!