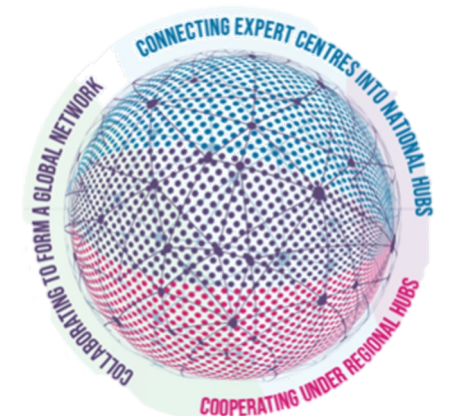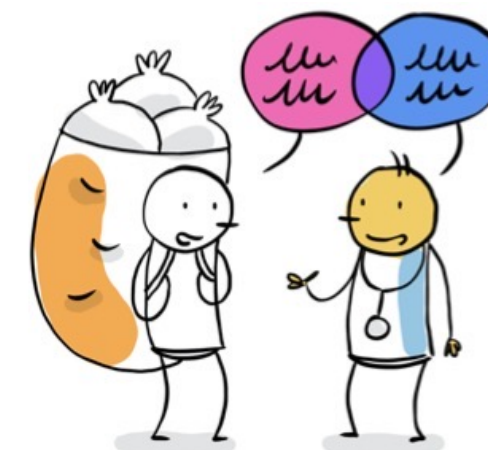**Informal Side-Event**
75th World Health Assembly
25 May 2022 | 9:30 to 12:30 (CET)

WORKSHOP ON
THE GLOBAL RARE
DISEASE NETWORK

Chair: Prof. Carmencita Padilla
University of the Phillipines, Manila

RARE
DISEASES
INTERNATIONAL

Strategic Planning & Mobilisation Session

Reflections on the Stepwise Approach for the Pilot

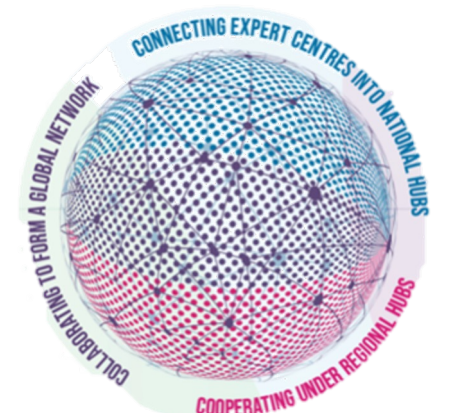|		
09:40 – 09:50	Opening Remarks	• Prof. Magdy El-Ekiby, Shabrawishi Hospital, Egypt (Session Chair)
09:50 – 10:00	• Rare Diseases Hub & National Networks: Brazil Case Study	• Prof. Robtero Giugliani, Universidade Federal do Rio Grande do Sul
10:00 – 10:10	• Cross Regional Collaboration: EURO & AFRO Regional Case Study	• Michelle Battye, ERN eUROGEN
10:10 – 10:20	• Mobilisation of the rare disease community: LATAM Case Study	• Alba Ancochea, ALIBER
10:20 – 10:40	**Panel Discussion & Q&A:** • Idenitfy key strategic actions needed to support the successful launch of the pilot • Mapping early adopters and future partners	• Victoria Hedley, University of Newcastle, UK

RARE DISEASES INTERNATIONAL

Conceptual Implementation of Regional Hub: EURO & AFRO Region Case Study

Michelle Battye ERN eUROGEN Programme Manager

RARE DISEASES INTERNATIONAL
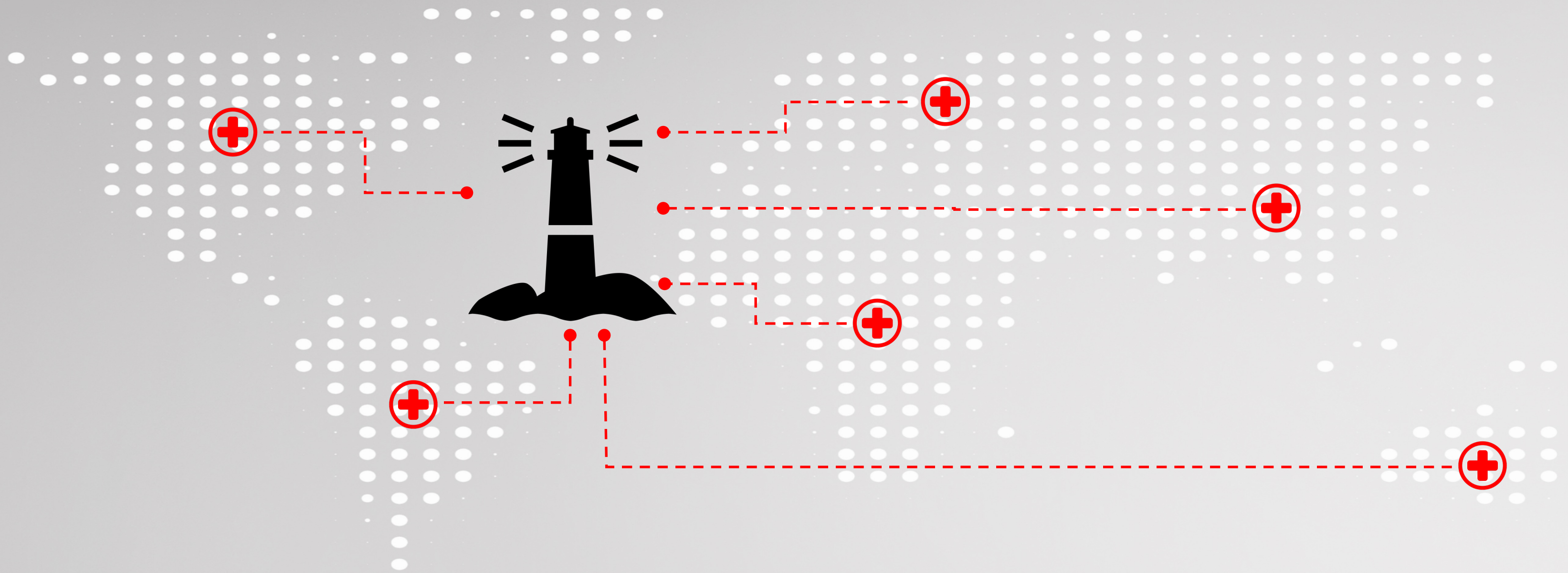
CONNECTING EXPERT CENTRES INTO NATIONAL HUBS

COLLABORATING TO FORM A GLOBAL NETWORK
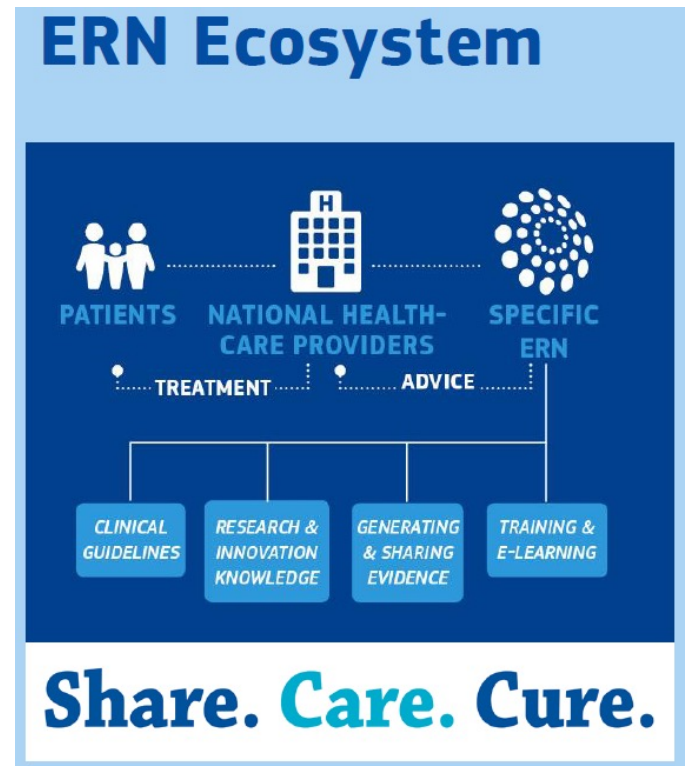
COOPERATING UNDER REGIONAL HUBS

# Connect ERN system to Global RD network pilot

The evolution of the ERNs

- A long process since 2004 with the first Rare Disease Task Force
- Council Recommendation in 2009
- Cross-Border Health Directive 2011/24/EU
- 2015: Implementation by the Commission
- 2017: Launch of 24 ERNs for five years (end March 23)
- 2019: ERNs enlarge to approximately 1600 specialised units
- 24 ERNs – wide fields – differing "natural histories" – heterogeneous entities

ERN Ecosystem
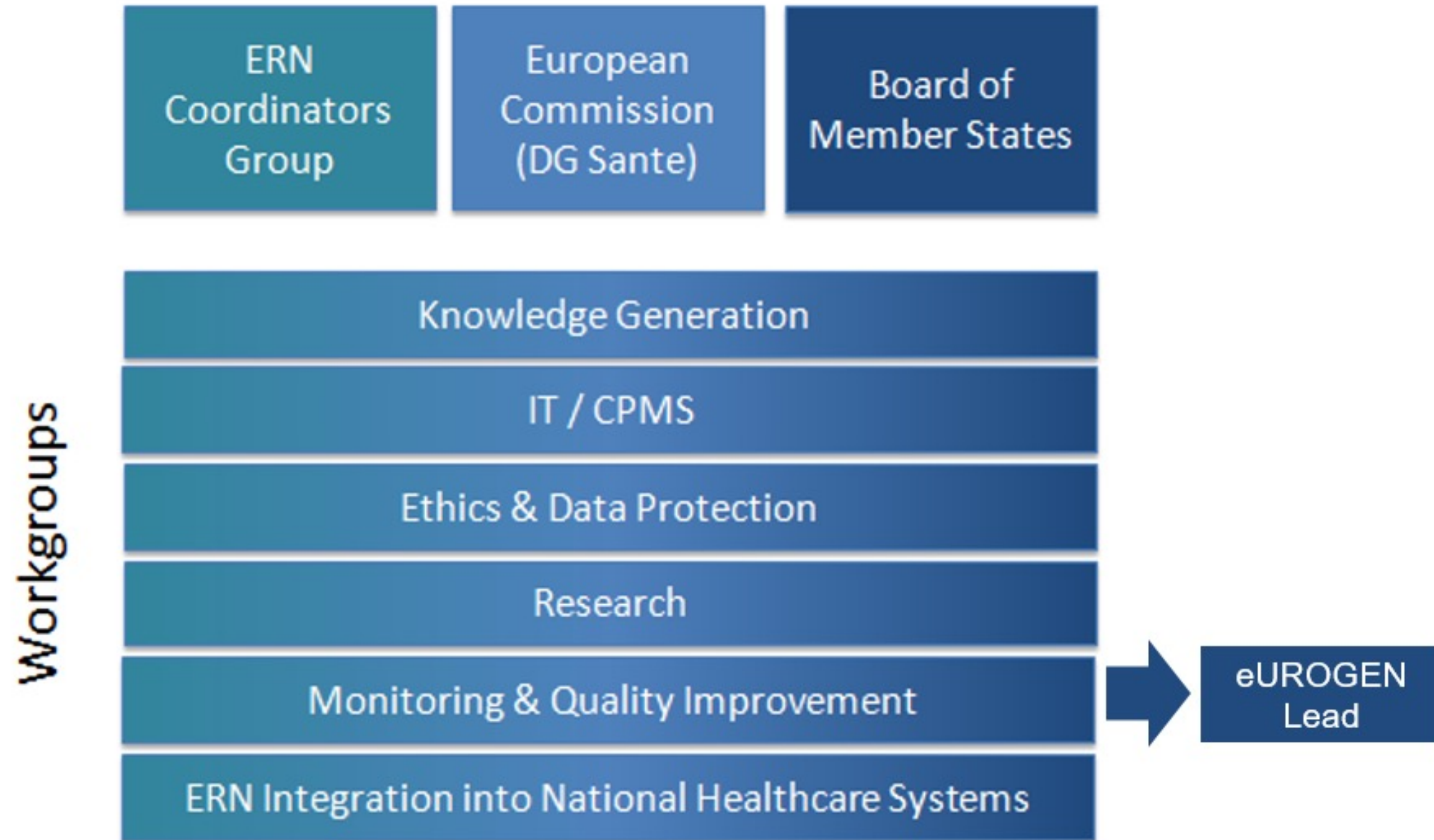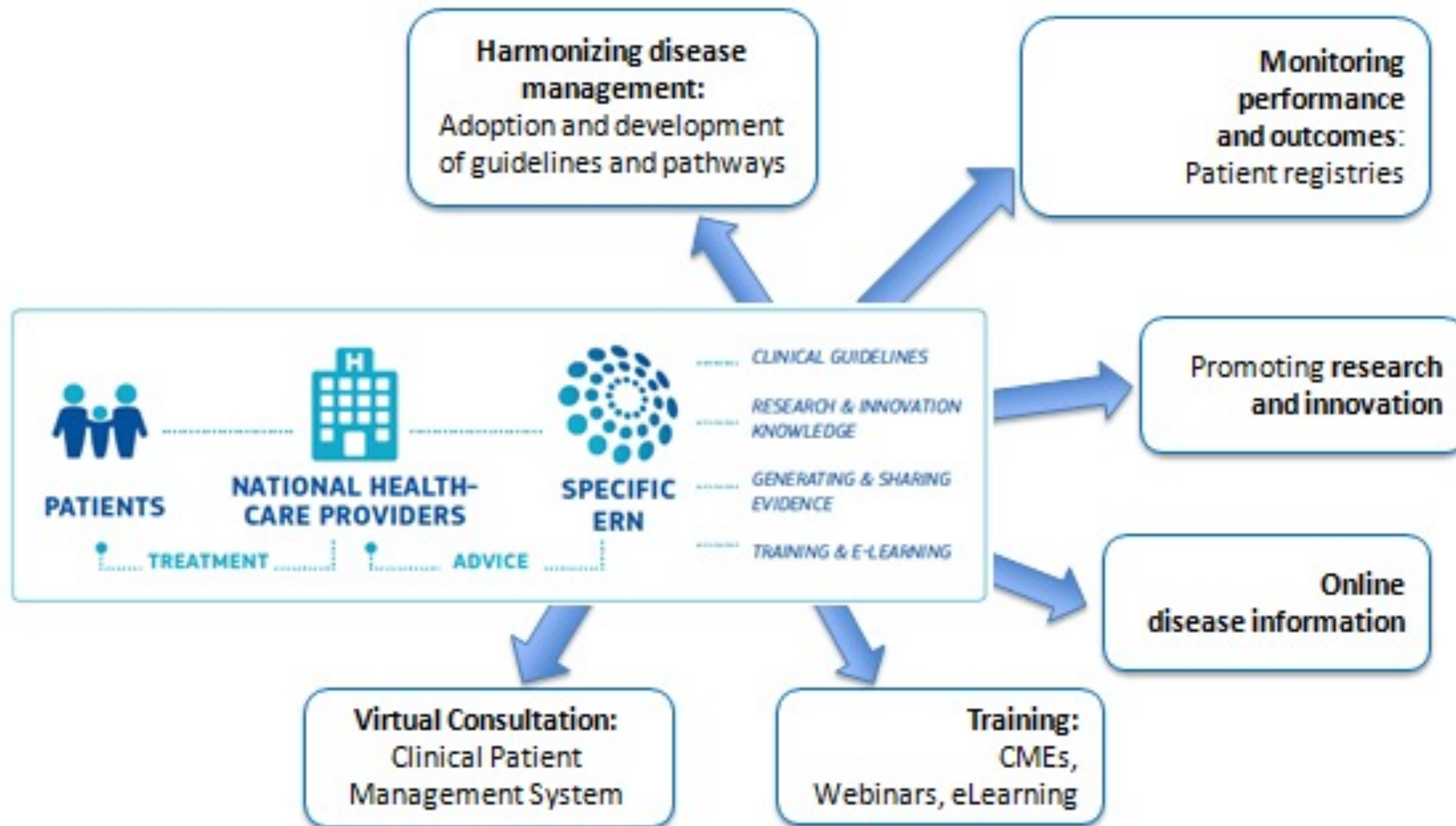
Share. Care. Cure.

ERN Governance

Pilot readiness?

1. Governance – ERN Coordinators Group

2. Identification of lighthouse healthcare provider – connection point

3. Identification of priorities for Global RD Network
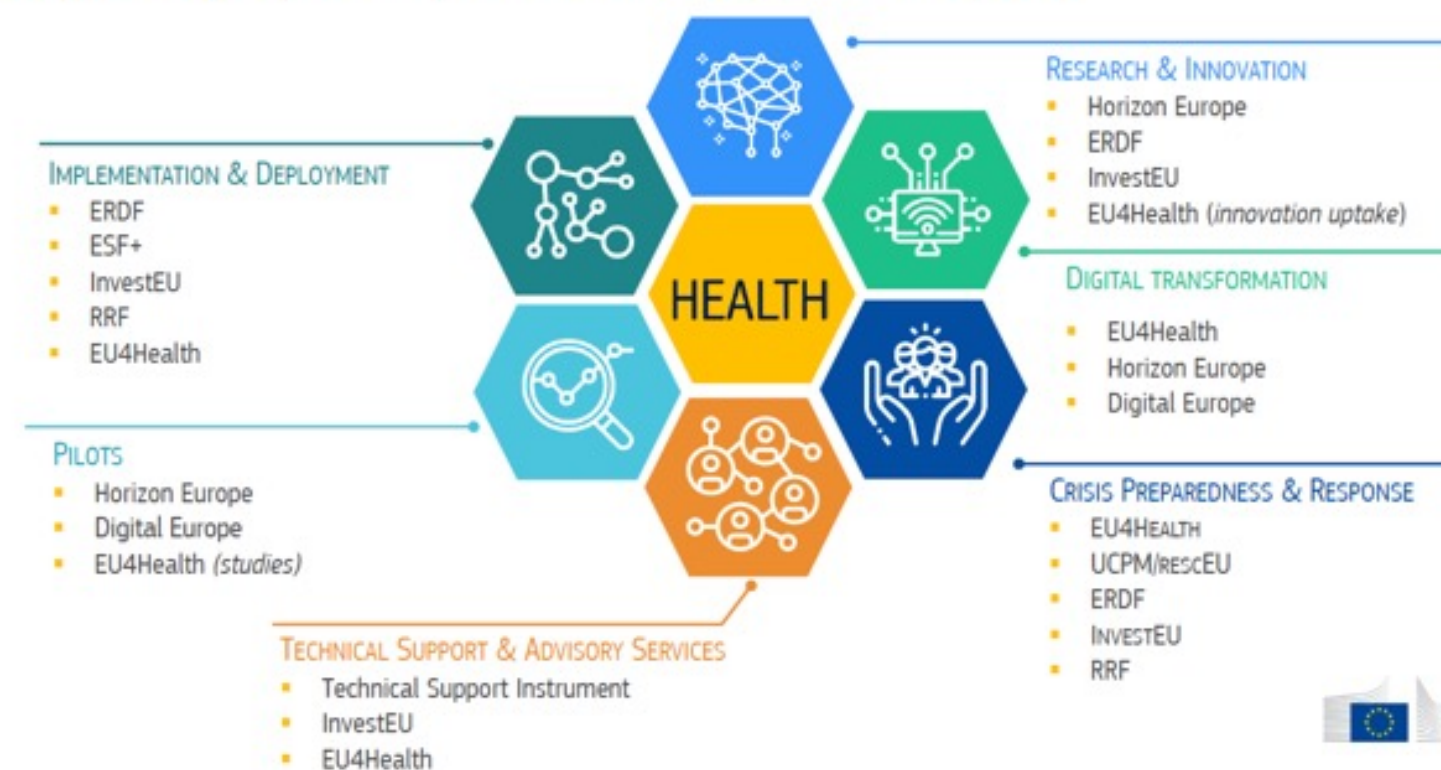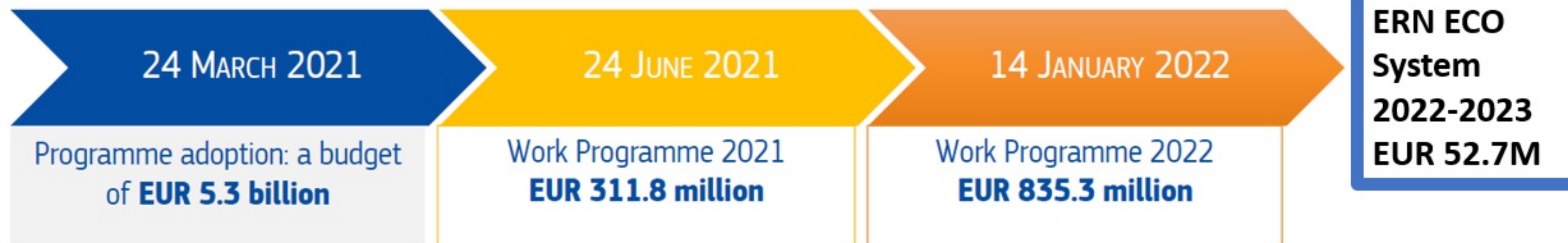
What ERNs can deliver

Harmonizing disease management: Adoption and development of guidelines and pathways

Monitoring performance and outcomes: Patient registries

PATIENTS — NATIONAL HEALTH-CARE PROVIDERS — SPECIFIC ERN

TREATMENT — ADVICE

CLINICAL GUIDELINES

RESEARCH & INNOVATION KNOWLEDGE

GENERATING & SHARING EVIDENCE

TRAINING & E-LEARNING

Promoting research and innovation

Online disease information

Virtual Consultation: Clinical Patient Management System

Training: CMEs, Webinars, eLearning

Next steps

STAKEHOLDERS
(INPUT)

EU4HEALTH STEERING GROUP – MS
(CONSULTATION)

EUROPEAN PARLIAMENT – ENVI COMM.
(INFORMATION)

EUROPEAN COMMISSION
DG SANTE

EU4HEALTH PROGRAMME COMMITTEE –
MS (OPINION)

EUROPEAN COMMISSION
(DECISION)

EU4Health – synergies & complementarities

An approach by policy area: many instruments contributing to the same objective

IMPLEMENTATION & DEPLOYMENT
- ERDF
- ESF+
- InvestEU
- RRF
- EU4Health

RESEARCH & INNOVATION
- Horizon Europe
- ERDF
- InvestEU
- EU4Health (*innovation uptake*)

DIGITAL TRANSFORMATION
- EU4Health
- Horizon Europe
- Digital Europe

HEALTH

PILOTS
- Horizon Europe
- Digital Europe
- EU4Health (*studies*)

CRISIS PREPAREDNESS & RESPONSE
- EU4HEALTH
- UCPM/rescEU
- ERDF
- InvestEU
- RRF

TECHNICAL SUPPORT & ADVISORY SERVICES
- Technical Support Instrument
- InvestEU
- EU4Health

European Commission

	EUR Million
Health systems & healthcare workforce	**126.5**
REFORMING AND STRENGHTENING HEALTH SYSTEMS	0.7
TRAINING FOR HEALTH WORKFORCE, INCLUDING DIGITAL SKILLS	29
ENHANCED EUROPEAN REFERENCE NETWORKS	52.7
IMPLEMENTATION OF THE PHARMACEUTICAL LEGISLATION AND STRATEGY	10.2
STRENGTHENING THE IMPLEMENTATION OF THE LEGISLATION ON BLOOD, TISSUES AND CELLS AND ORGANS	6.9
IMPLEMENTATION OF REGULATIONS ON MEDICAL DEVICES AND IN VITRO DIAGNOSTIC MEDICAL DEVICES	19.8
PREPARATION AND IMPLEMENTATION OF THE HEALTH TECHNOLOGY ASSESSMENT (HTA) REGULATION	7.2

24 MARCH 2021
Programme adoption: a budget of **EUR 5.3 billion**

24 JUNE 2021
Work Programme 2021
EUR 311.8 million

14 JANUARY 2022
Work Programme 2022
EUR 835.3 million

ERN ECO System 2022-2023 EUR 52.7M

THANK
YOU

RARE
DISEASES
INTERNATIONAL

Regional Preparedness & Planning Session:
Reflection of Learning from Regional Workshops
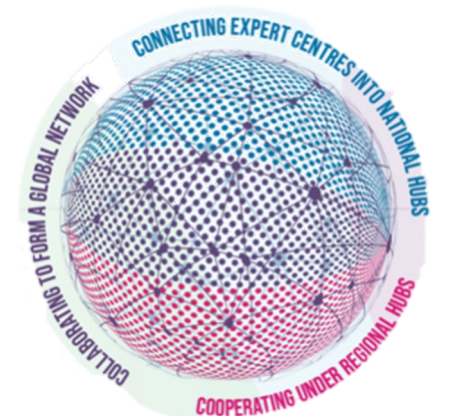
10:55 – 11:00	Opening Remarks	• Matt Bolz-Johnson, RDI (Chair)
11:00 – 11:10	• AFRO Action Plan	• Prof. Johnny Mahlangu & Kelly du Plessis, RD South Africa
11:10 – 11:20	• AMRO Action Plan	• Lisa Sarfaty, NORD & Durhane Wong-Rieger, CORD
11:20 – 11:30	• LATAM Action Plan	• Prof. Roberto Giugliani & Alba Ancochea, ALIBER
11:30 – 11:40	• EMRO Action Plan	• Prof. Magdy El-Ekiby & Salman Saif, CareforU
11:40 – 11:50	• EURO Action Plan	• Dr. Loredana D'Amato, Orphanet & Michelle Battye, ERN eUROGEN
11:50 – 12:00	• SEARO & WPRO Action Plan	• Prof. Carmencita Padilla, Rachel Yang, CHARD & Ritu Jain, APARDO
12:00 – 12:20	**Panel Discussion & Q&A:** • Identify the operational activities	• Louise Clément, Health Standards Orgnisation
12:20 – 12:30	**Closing & Take-Home Messages**	• Prof Gareth Baynam, UDP-WA & Yann La Cam, EURORDIS

RARE DISEASES INTERNATIONAL

Overview of the European Region

The western parts of the European region, **27 countries are members of the European Union (EU) & more countries will join**

Population: 0,93 Billion (12,13%)
52 Countries (26.67%)
10,1 million square km (6%)

Overall mature HCS, with inequalities in patients' rights & access to diagnosis, care and treatment.

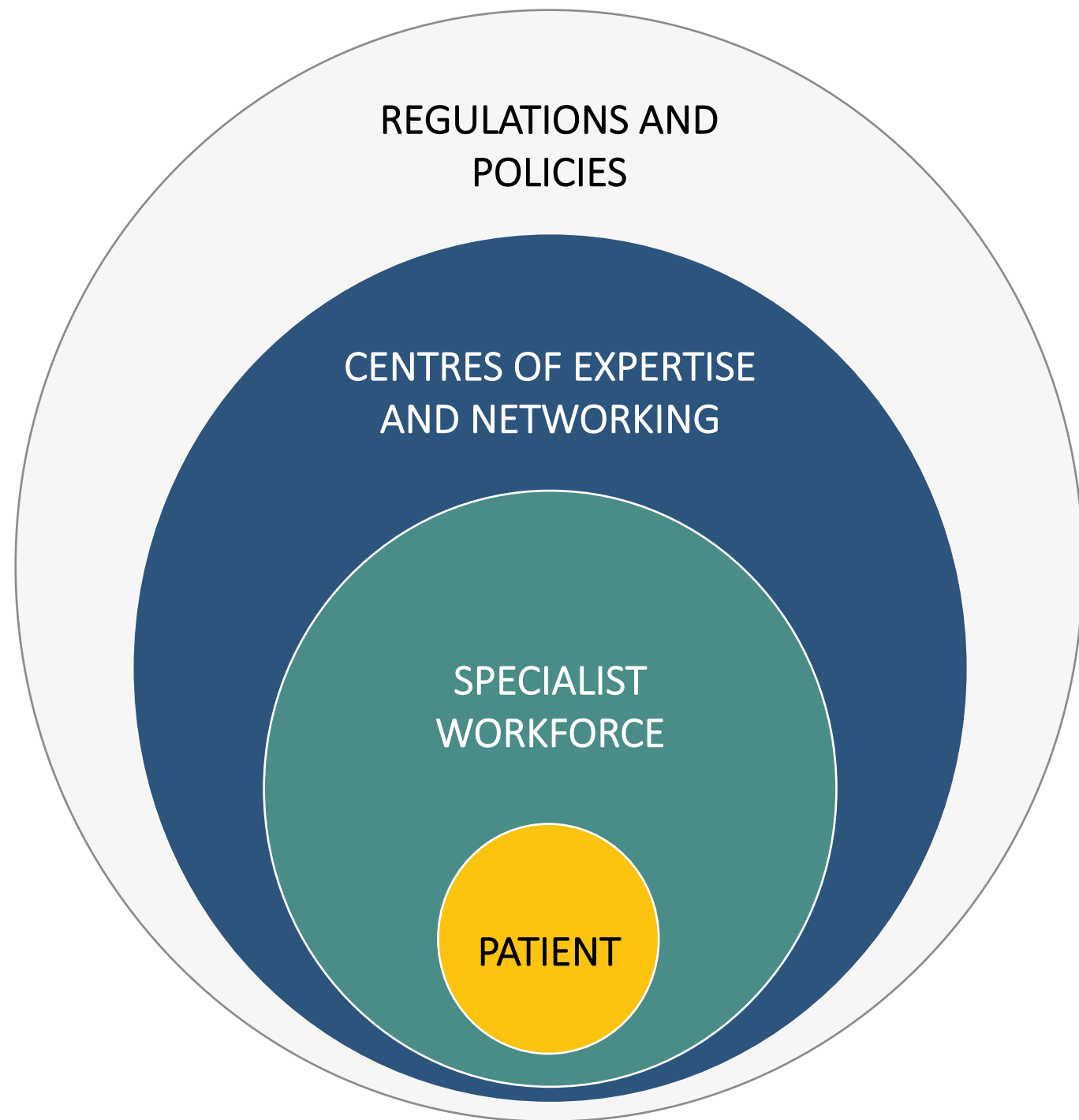**Orphan products approved by EMA**, but **each country controls pricing and reimbursement → variations in availability and access** of medicines
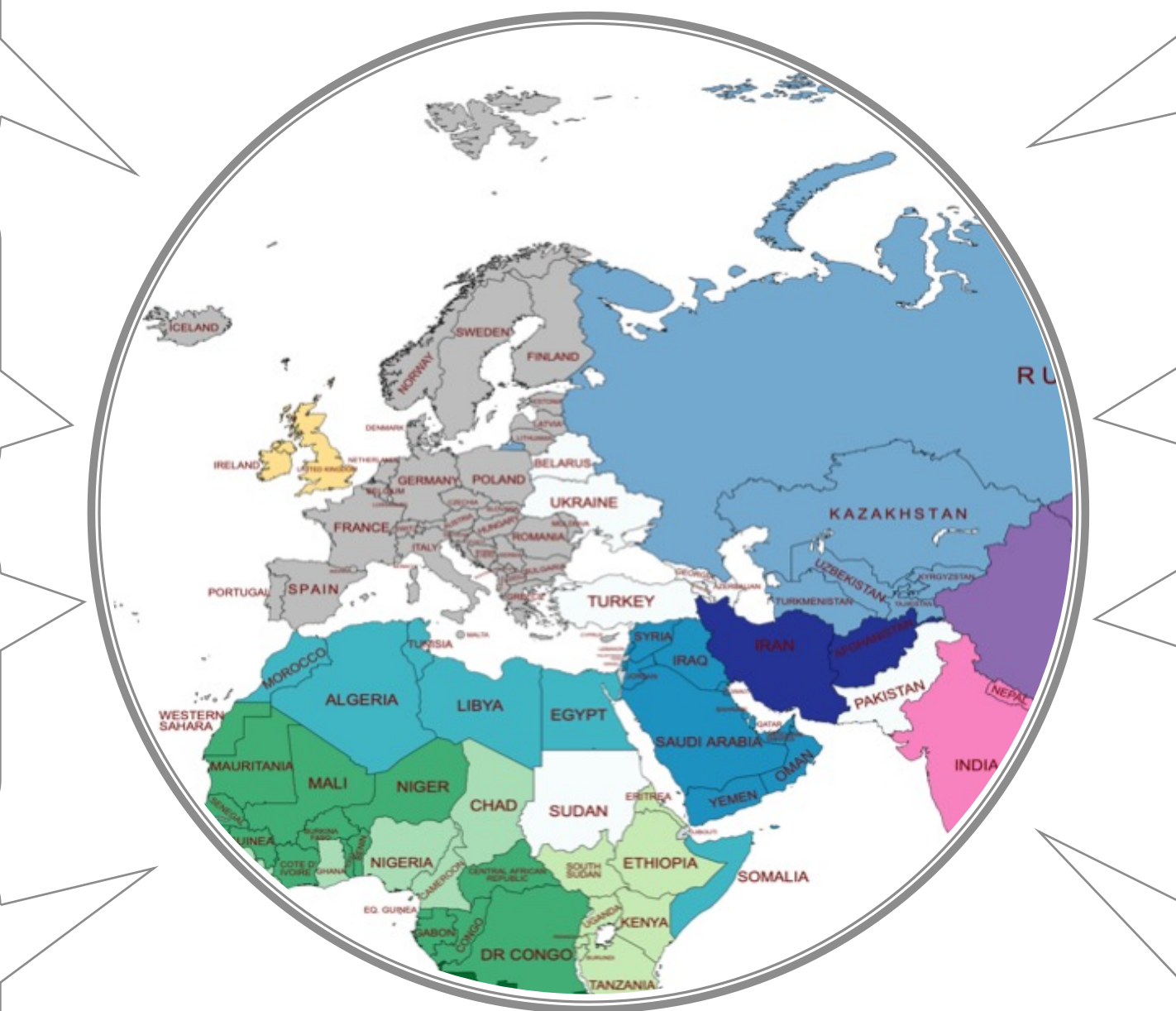
Russian Fed. has the **larger population,** 145.9M (15.6% of region) **& the biggest area,** (39% of the continent land)

The **10 most populated countries** represent **the 71,8% of region** people

Estimated PLWARD in the Region is **between 32.7M and 55M** persons living with a rare disease

In **eastern Europe** 9 countries cooperate at **Commonwealth of Independent States (CIS)**; formed after the Soviet Union dissolution

How Healthcare is Organized for RD

REGULATIONS AND POLICIES

CENTRES OF EXPERTISE AND NETWORKING

SPECIALIST WORKFORCE

PATIENT

Regulations & Policies:

- Rare disease has been recognised as a public health priority in the EU and some Eastern European countries increasing the cross-border cooperation between States.

- Greater acknowledgement across the European region. Twenty-five EU MSs have a National RD Plan as well as outside the EU and under development (the former Yugoslav Republic of Macedonia, Norway). Some countries have none (Russian Federation, Island).

- Orphan medicine designation is coordinated under the European Medicine Agency (EMA) and regulated by the Orphan Medicine Regulation and Advance Therapies Regulation, encouraging investment in research and therapeutic development.

Centres of Expertise & Networking:

- Well-funded healthcare systems in some EU Member States, compared to other WHO Reg.

- An EU healthcare infrastructure is developing under the European Reference Networks, and there are a growing number of accredited national Centres of Excellence in the Member States. There is an opportunity to establish ERNs further and connect them throughout the WHO European Region. There is also a well-structured research infrastructure and collaboration under the European Joint Programme for RD, the ERNs research communities.

- HC 4RD is developing outside the EU, (Israel & Switzerland) with the recognition of Reference Centres, Centres of Expertise and Rare Disease Units.

Patient level:

- Patient community and representatives have been growing and is well organised and positioned to represent the community in developing strategy, policy, legislation, and operationalising front-line services. This facilitates the process of "incorporating" rare diseases into the national healthcare system.

Connecting with ERN networks
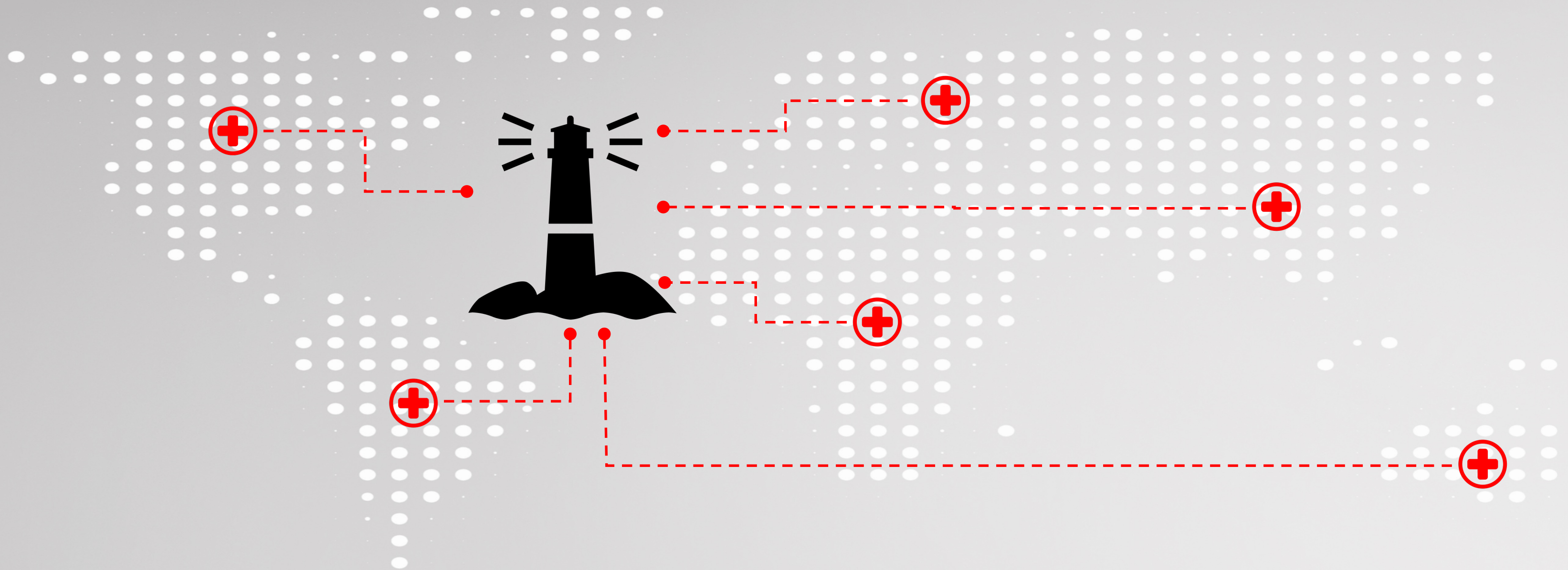
EURO Region

2 Regional Hubs:

- **Regional Hub 1:** Central and Eastern Europe: ERNs with Iceland, Israel, Switzerland and the UK

- **Regional Hub 3:** Commonwealth of Independent States (CIS)

However, more discussions are needed with leads from these countries to further explore the different possible configurations.

Additional considerations around hub distribution:

- EURO 1: Baltic countries do not naturally collaborate with each other – will remain in EURO1

- EURO 2: Outstanding actions - to confirm the grouping with representatives from CIS

- Turkey (EURO-EMRO) is a bridge country

RARE
DISEASES
INTERNATIONAL
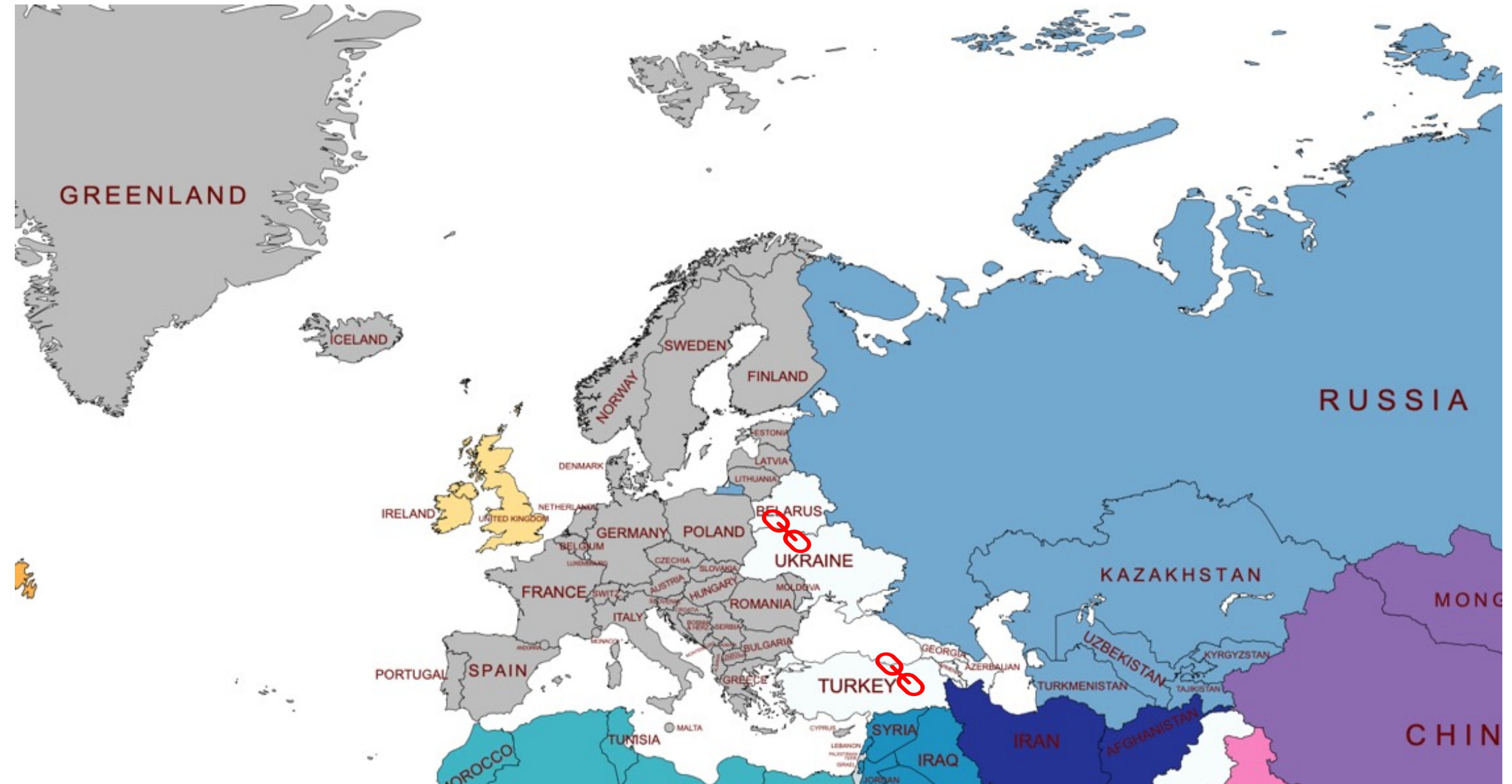
Action Plan

Strengths

- EC legislative framework - Comm in 2008, Council Recommendations 2009
- Maturity of networks (ERNs, Orphanet, Infrastructures)
- ERNs have legal base to collaborate with other international centres of expertise
- ERN registries and European Health Data Space
- EJP RD research ecosystem
- Data collection – interoperable global registries for RD based on ORPHAcodes
- EC funding
- ERNs have legal base to collaborate with other international centres of expertise
- Maturity of Patient Organisations

Actions to Be Ready

- Linking into existing networks – how?
- Identification of "lighthouse" healthcare providers, support
- Build on existing tools eg CPMS used in campaign to help rare disease doctors and patients in the Ukraine https://www.erncare4ua.com/
- GN4RD Regional Hubs define priority actions/disease areas – linked with existing networks eg knowledge sharing, clinical exchanges, virtual consultations
- No forum to bring national policy makers together to share best practice and peer review NAPs on RD
- Use Orphanet as a global network for information and orientation

Pilot Readiness?

1. Governance – ERN Coordinators Group

2. Identification of lighthouse healthcare provider – connection point

3. Identification of priorities for Global RD Network

4. Funding - WHO EC collaboration?
    EU4health and other programmes to stimulate ERN international collaboration?

orphanet

Free access reference resource on rare diseases since 1997

NIH National Center for Advancing Translational Sciences

GARD Genetic and Rare Diseases Information Center

A global network improving **visibility**, **awareness**, **information** and **knowledge** in the field of rare diseases

CREATION
1997

42 COUNTRIES

110 PEOPLE

RARE DISEASES INTERNATIONAL

Provide Information on each Rare disease

Orient patients and doctors through the rare disease pathway
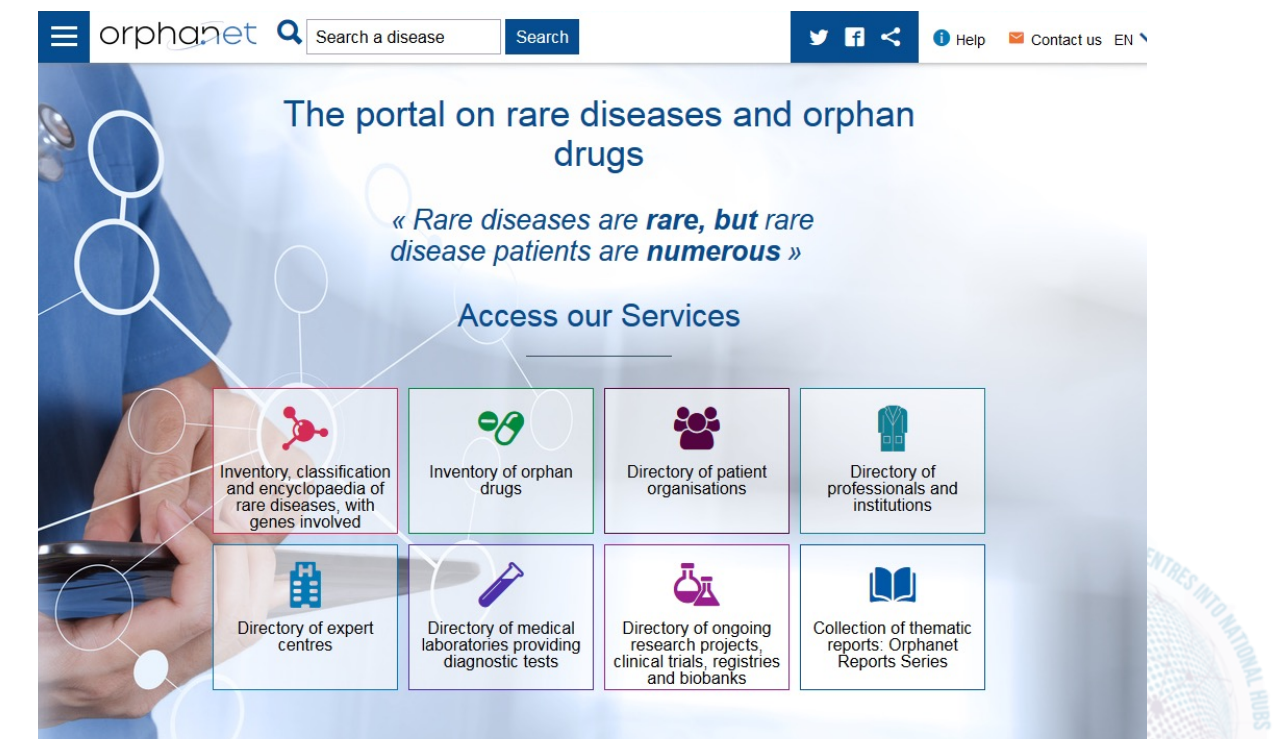
www.orpha.net

Users

Around **1,65 million visitors per month** from **238 countries**

35 % health professionals
**38 % patients, families and support groups**
As well as **students, researchers, clinical coders, industry, policy makers.**
Most appreciated products: disease summary texts on diseases, clinical signs associated to a rare disease, classifications of rare diseases, functional consequences of rare diseases (disabilities), epidemiological data*

* Annual Orphanet Users' Survey 2022

Country	Users	New Users
	1,291,191 % of Total: 100.00% (1,291,191)	1,154,084 % of Total: 100.06% (1,153,379)
1. Italy	**256,027** (19.85%)	227,398 (19.70%)
2. France	**210,665** (16.33%)	180,414 (15.63%)
3. Germany	**114,526** (8.88%)	106,611 (9.24%)
4. Spain	**77,129** (5.98%)	68,119 (5.90%)
5. United States	**76,670** (5.94%)	73,879 (6.40%)
6. Mexico	**71,114** (5.51%)	62,258 (5.39%)
7. Brazil	**59,881** (4.64%)	56,346 (4.88%)
8. Colombia	**32,781** (2.54%)	29,219 (2.53%)
9. Netherlands	**29,137** (2.26%)	27,438 (2.38%)
10. Belgium	**27,498** (2.13%)	24,057 (2.08%)
11. Argentina	**23,259** (1.80%)	21,058 (1.82%)
12. India	**23,001** (1.78%)	21,571 (1.87%)
13. Switzerland	**22,060** (1.71%)	20,037 (1.74%)
14. Canada	**18,295** (1.42%)	17,039 (1.48%)
15. Peru	**18,139** (1.41%)	16,171 (1.40%)
16. Poland	**17,186** (1.33%)	15,746 (1.36%)
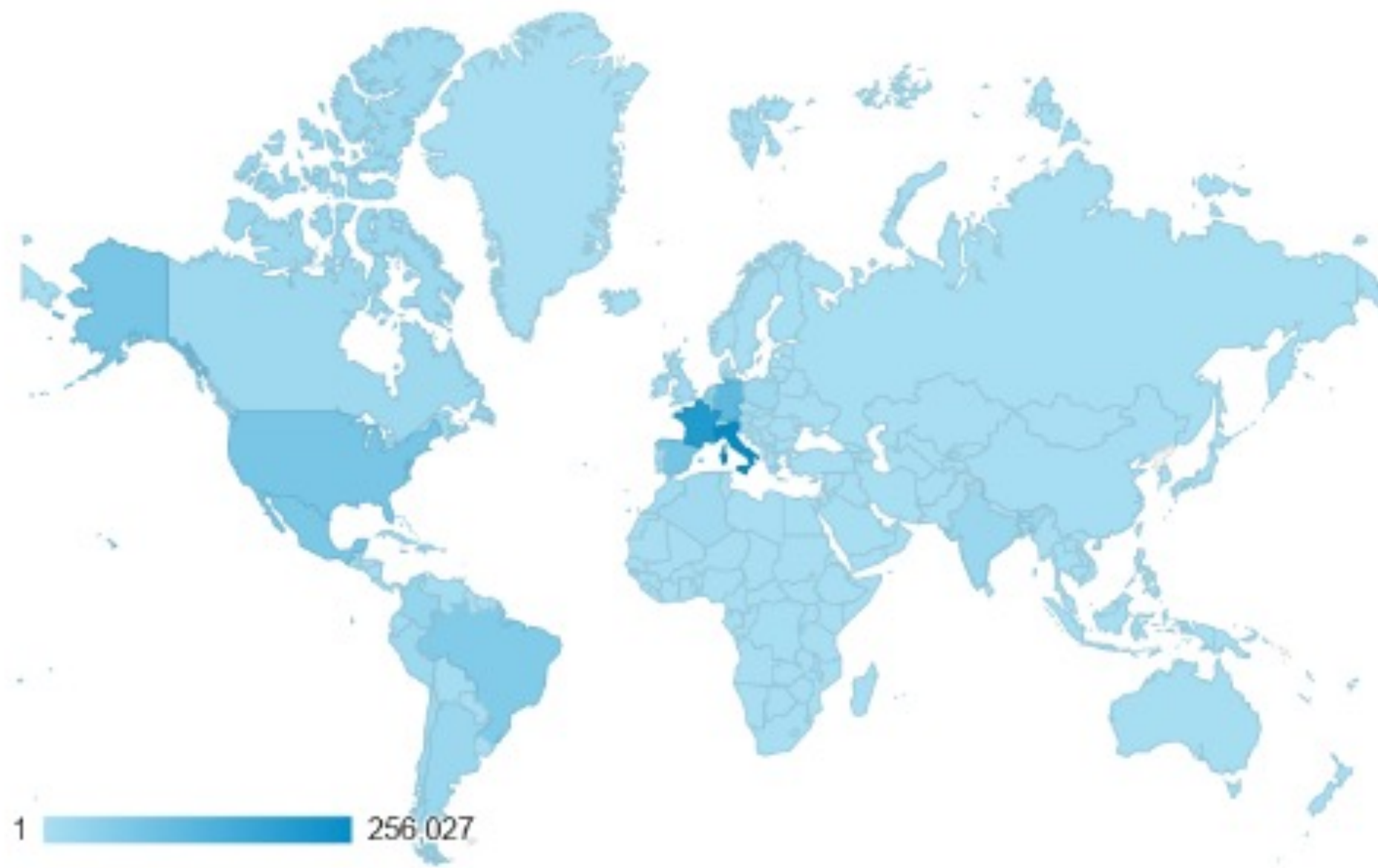
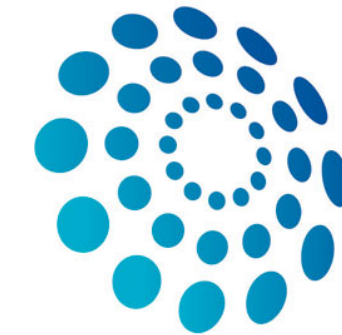1　256 027

A common language
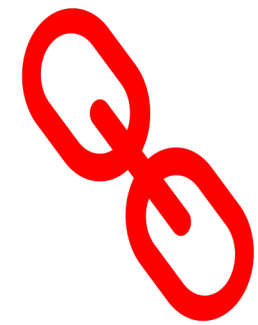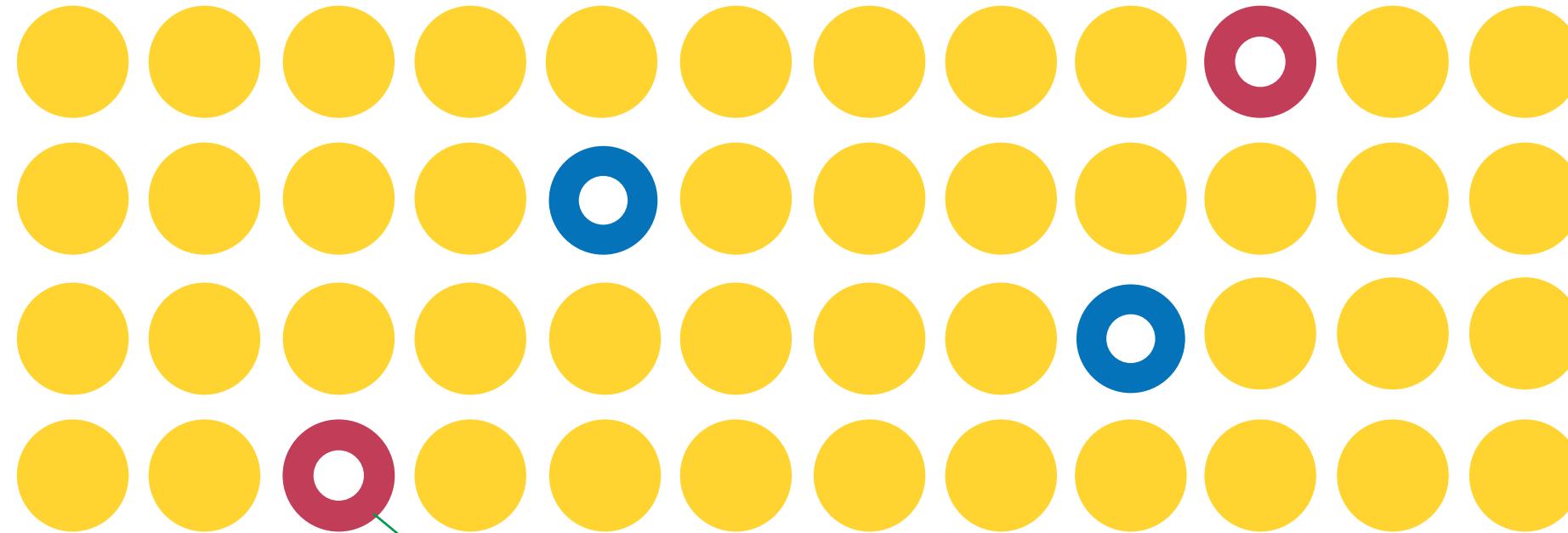
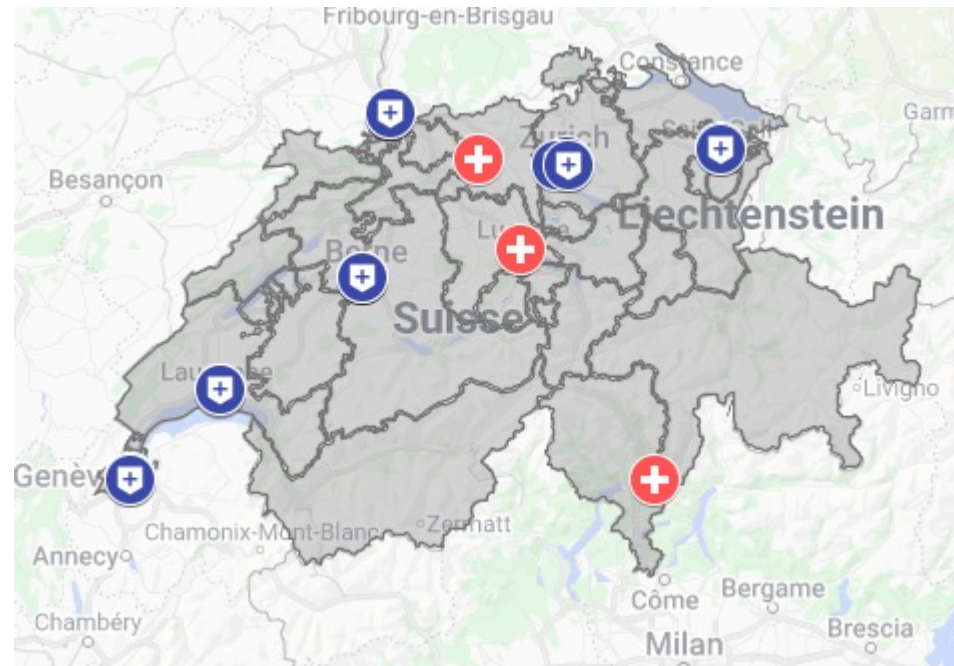# Reinforce the national level to add international value

Non European countries ← orphanet ← **European Reference Networks**

National nodes

- Care and research activities
- ORPHAcode implementation

National hubs

- Information
- ORPHAcode implementation support

National nodes

- Care and research activities
- CPGs
- ORPHAcode implementation

Pilot Readiness?

1. Identification of lighthouse – Orphanet connection point

2. Funding - WHO EC collaboration?
   EU4health and other programmes to stimulate Orphanet and ERN international collaboration?

Thank you for your attention