Addressing the unmet needs of patients and their families

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Session 1:
Rare diseases: organisational models and good practices in health and social care

Conference on Rare diseases and European Reference Networks.
How to guarantee solidary for patients?
Bilbao, 11 October 2023

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A European Action Plan for Rare Diseases

#EUAction4Rare

Progress has been made thanks to…

- Empowered patient communities and their national or European networking
- Legislation to incentivise investment in rare disease research / orphan medicinal products and national companion measures
- National Plans and Strategies setting national priorities and European collaboration between Member States
- National Centres of Expertise and European Reference Networks
- Disease registries, good practice guidelines
- National and European research programmes

2000 OMP Regulation
2006 Paediatric Use Regulation
2007 Advanced Therapies Regulation

2008 Council Recommendation
2009 Commission Communication

2011 Cross border healthcare Directive

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The rare disease story in Europe so far

Rare 2030 Foresight Study

The main recommendation called for a renewed European policy framework on rare diseases by 2023, to connect the dots between different policy areas and different countries in Europe

Rare 2030

2000
EU Regulation on Orphan Medicinal Products

incentivised investment in rare disease research

2006
EU Regulation on Paediatric Use of Medicines

2007
EU Regulation on Advanced Therapies Regulation

2008
The first Rare Disease Day

RARE DISEASE DAY

global solidarity for rare diseases

2008–2009
European Commission's Communication and EU Council Recommendation on rare diseases

rare diseases recognised as a public health priority; Member States recommended to introduce a national plan on rare diseases

2011
EU Cross-Border Healthcare Directive

European Reference Networks have a foundation

2017
Launch of 24 European Reference Networks

2019
European Court of Auditors report on EU actions for cross-border healthcare

calling for a review of rare disease strategies

Feb 2021
Rare 2030 Foresight Study Recommendations published

Dec 2021
UN Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families

The Rare Disease Population in Europe

an estimated

30 Million

people are living with a rare disease in

48 countries

in Europe.

countries

there are over

6000

distinct rare diseases.

Each rare disease affects fewer than

1 in 2000

people.

Rare diseases affect about

4%

of the population during their lifetime

lifetime

with estimates ranging from from 3.5% to 5.9%.

%

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Has vast unmet medical needs

70%
of people with rare diseases

wait more than 1 year to get a confirmed diagnosis after coming coming to medical attention.

It takes on average

5 years

for rare disease patients to get a diagnosis.

There are over

230

orphan medicines

authorised in the EU. The goal is to support support the development of

1,000

new therapies

for rare diseases by 2030.

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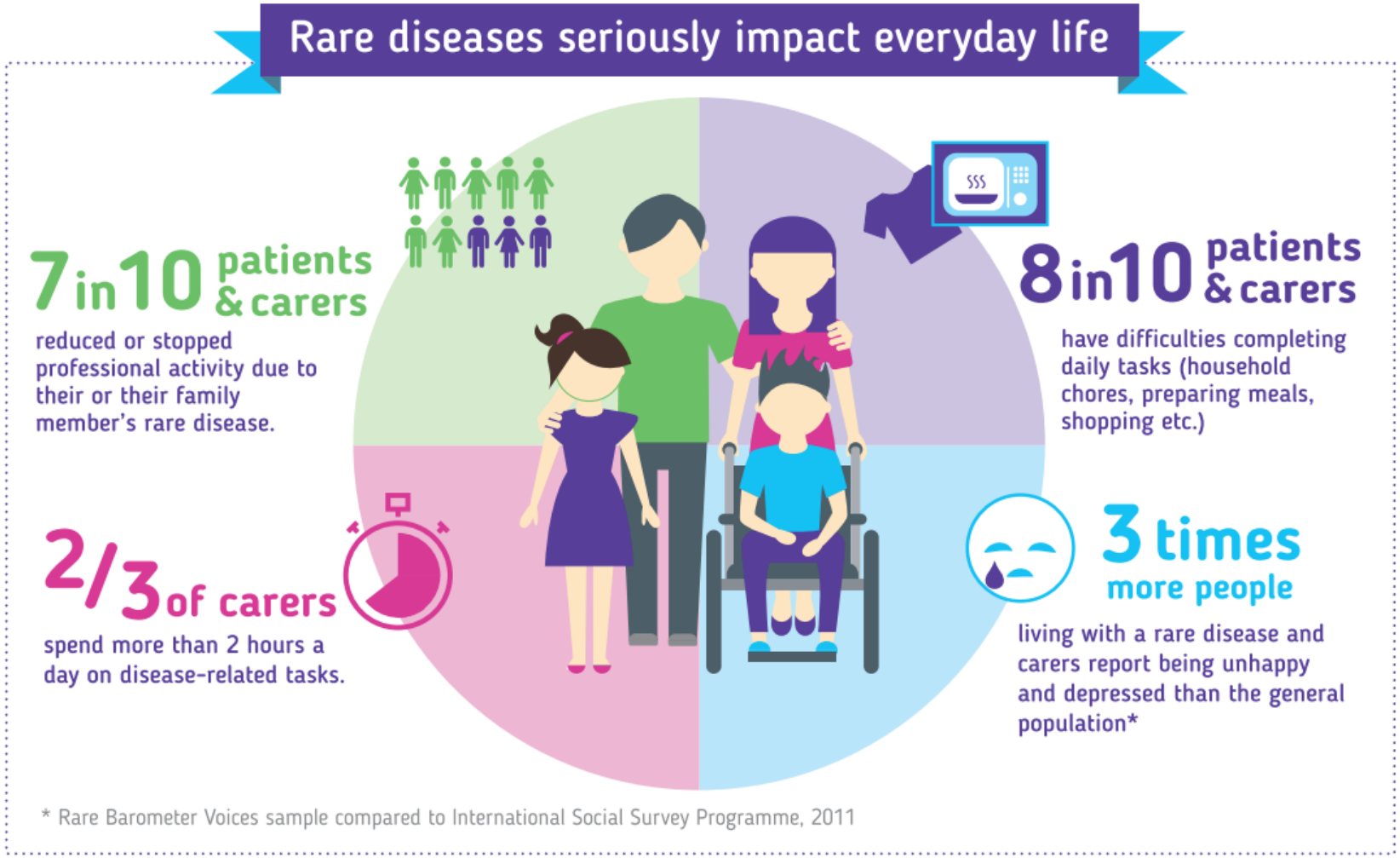
And cumulate vulnerabilities



New challenges… and new opportunities

- Knowledge & Science & Technologies
- Diagnostic tools
- Disease transformative therapies
- Data & Digital transformation
- Social and Economic impact

Rare 2030 Foresight Study (2019-2021): A roadmap for rare disease policy to 2030

- Recommendation n°1: **a new European Policy Framework driven by the needs of people living with a rare disease, to guide the implementation of consistent national plans and strategies**

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A goal-based strategy

8 Rare 2030 Recommendations

1. European/national plans and strategies
2. Diagnosis
3. Access to care
4. Person-centred care
5. Patient Partnerships
6. Research
7. Data
8. Treatment

Europe's Action Plan

GOAL 1: Ensuring healthy lives and promoting well-being

GOAL 2: Reducing inequalities

GOAL 3: Building resilient infrastructure, promoting inclusive and sustainable industry and fostering innovation

4 SDGs

SDG3: Ensure healthy lives and promote well-being for all at all ages

SDG 9: Build resilient infrastructure, promote inclusive and sustainable industrialisation and foster innovation

SDG10: Reduce inequalities within/among countries

SDG17: Revitalise the global partnership for sustainable development

Why a renewed European policy framework on rare diseases?

The **Rare 2030 Foresight Study** - initiated by the European Parliament and co-funded by the European Commission - concludes that renewed European action on rare diseases is required now to:

Address the remaining unmet needs and inequities all along the patient journey in accessing a diagnosis, treatments and care, leaving people living with a rare disease marginalised in society;

Keep pace with new technologies, new values and new expectations of Europe's citizens and give a new focus to national rare disease plans and strategies

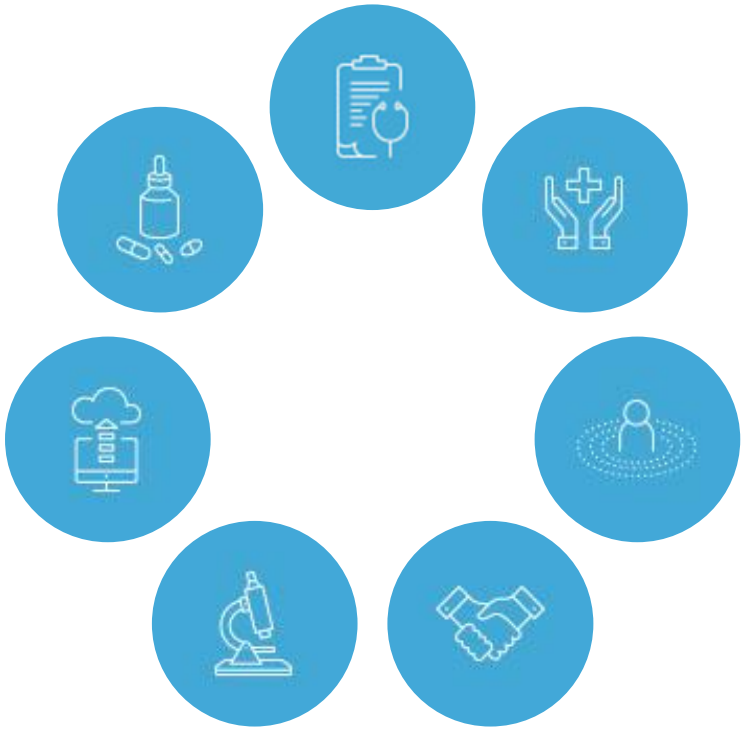
Sustain the European Commission's strategic approach in addressing a **distinctive domain of high European added-value**

Bring together existing and upcoming actions:
- **across countries**
- **across sectors and policy areas**
- **across the rare disease pathway**

where the EU can add the most value under **one interconnected framework**.
- **within international perspective**

A comprehensive, integrated, goals based European policy framework

- **Common objectives and measurable goals to set a common direction**
- The person's journey: **A holistic and life-long view**
- **Bridging the gaps between national and EU initiatives**
- **Bridging the gaps between different legislative pieces** on data, research, treatment, healthcare, social care



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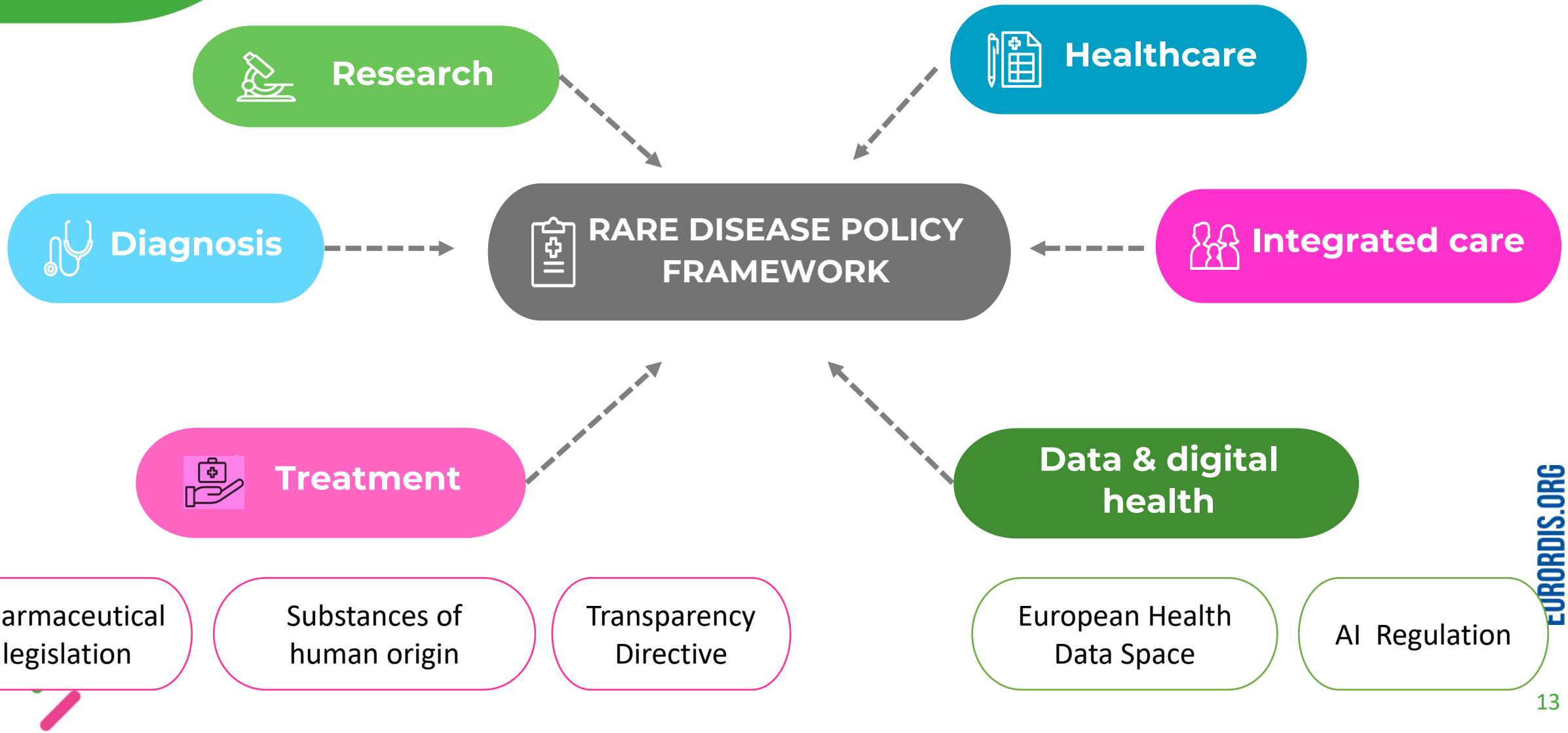
Our challenge in Europe: achieving a truly integrated ecosystem



A call for a European Action Plan for Rare Diseases from across the community

- **2019 European Court of Auditors**' Report
- All stakeholders: patients & families, clinicians (eg ERNs), researchers, pharma & biotech, national policy makers : **Foresight Rare 2030 Recommendation n°1**
- The 2000 rare diseases patient organisations across Europe, all national alliances, all European federations: **#30 Millionreasons, a call for the families and persons living with a rare disease**
- **European Parliament** debate, 45 MEPs call upon the Commissioner for Health, mention in resolutions on building back better
- **Member States** support through the Slovenian, French and Czech EU Council Presidencies July 2022 - 50+ partners of the **European Conference on Rare Diseases** (ECRD)
- **European Economic and Social Committee** opinion on rare diseases
- **22 Member States endorsed** the **Call to Action of the Czech EU Council Presidencies**
- Rare Diseases Day 2023 - **50 Members of the European Parliament** call on the President of the Commission Ursula von der Leyen and the Health Commissioner Kyriakides

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The voice of the EESC - European Economic and Social Committee

"Having heard evidence from people right across the patient community and civil society, we learned that there is huge, unrealised potential for EU policy to make sure scientific, technological, clinical and social advances reach every European citizen living with a rare disease"
Alain Coheur, EESC Rapporteur.

What the EESC called for:

- A European Action for RDs based on measurable goals
- Optimising the use of health data
- Speeding up diagnoses
- Support for – and beyond – the European Reference Networks
- Improving access to orphan drugs
- Greater support for informal carers

The voice of the European Parliament: 50 MEPs signed a letter to the President of the European Commission on Rare Disease Day

European Parliament

To:
Ms. Ursula von der Leyen
President of the European Commission
Rue de la Loi 200
1049 Brussels

Copy to :
Ms Stella Kyriakides
Commissioner for Health and Food Safety

Mr. Nicolas Schmit
Commissioner for Jobs and Social Rights

Brussels, 28th February 2023

Rare Disease Day call for a European strategy on rare diseases

Dear Madam President,

28 February 2023 marks the 16th Rare Disease Day, a global awareness day celebrated in over 100 countries around the world. This year, Rare Disease Day puts the focus on equity.

As Members of the European Parliament and on behalf of the Network of Parliamentary Advocates for Rare Diseases, we have the honour to once again call on you to introduce a comprehensive European strategy on rare diseases to better meet the needs of the 30 million European citizens living with a rare disease.

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Rare 2030
Foresight in Rare Disease Policy

Feb 2021

Rare 2030 Reco n°1

May 2021

Launch of #30million reasons campaign

Nov 2021

European Parliament Debate

June 2022

ECRD 2022

March 2022

Conseil de l'Union européenne

EPSCO Council

Feb 2022

French Ministerial conference

Czech Presidency Expert Conference — **Oct 2022**

EESC Opinion on Solidarity in RDs — **Oct 2022**

Czech Call to Action + EPSCO

MEP letter to EC President — **Feb 2023**

TODAY's CONFERENCE — **Oct 2023**

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What's next?

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A EU “whole system” that “leaves no one behind” living with rare and complex conditions

An EU whole system for rare diseases

In **Rare 2030** this community recommended a multilevel support (European, national and local) to create

"a **mature highly specialised healthcare ecosystem** that **leaves no person living with a rare disease behind"**

a "strategy of **future ERNs targeted towards all rare disease patients** in Europe, and not only those attending ERN HCPs or 'affiliated' centres: ERN operations should always target this wider population, wherever possible"

Where are we today with this ambition?

- With the **ERNs and their integration in national healthcare systems we are leading the way** to reduce the time to early, better and more accurate diagnosis for all people living with rare and complex conditions in Europe, to secure high quality healthcare delivery
- Yet we need **continued commitment & an upgrade of the system** for **real impact on lives of people with RDs**
- Progressively we must will cover **all RD, in all countries in Europe**, eventually **measure health and social outcomes.**

An Open Letter from the RD community

The rare disease community comes together in calling on the EU institutions and our national governments to stand by the European Reference Networks.

Dear Mrs von der Leyen,
President of the European Commission,

Dear Mrs Metsola,
President of the European Parliament,

Dear Heads of Governments of EU Member States,

We, the undersigned, representing the European Reference Networks (ERNs) and people living with a rare disease across Europe, **call on the EU institutions and EU Member States' governments to stand by the European Reference Networks** and to uphold their commitment to enable long-lasting impact in people's lives and, fundamentally, give all people living with a rare or complex condition in Europe the same opportunities to access timely and adequate specialised healthcare.

ERNs and their integration in national healthcare systems & into the EU cross-border healthcare system

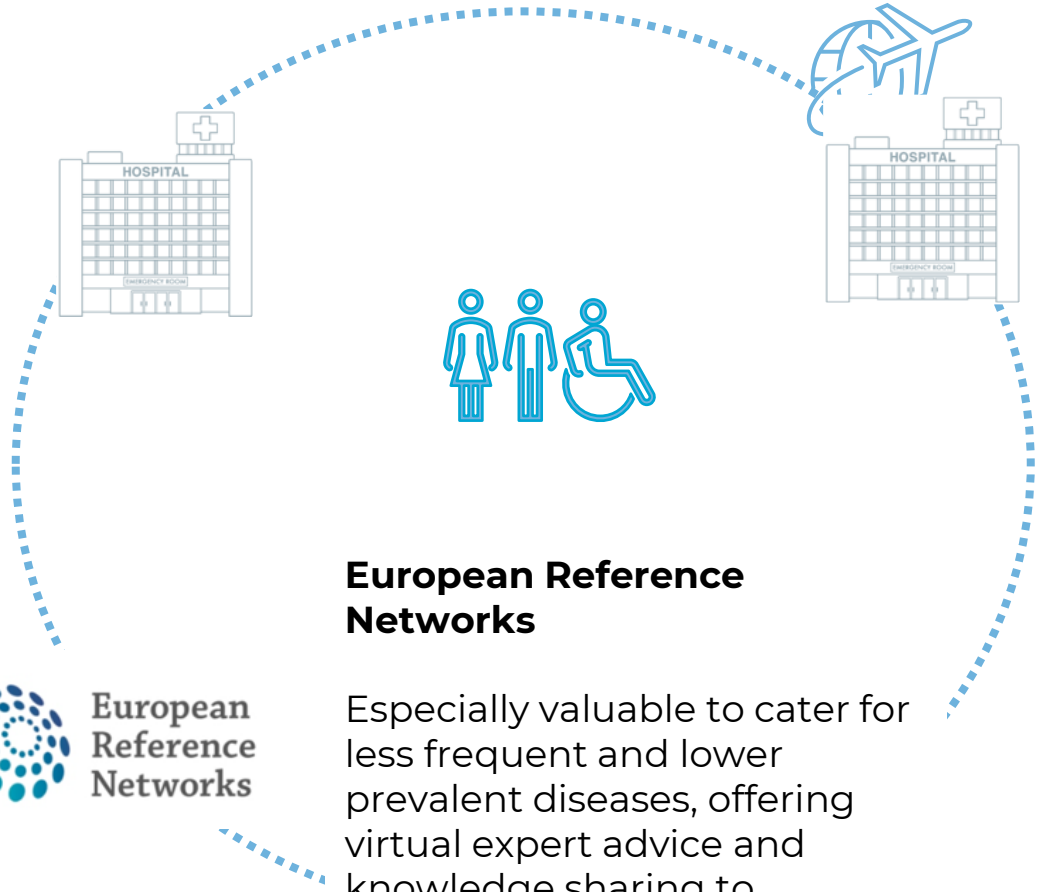
- The ambition is to have national health systems and ERNs **operating together as elements of ONE integrated European system** under a "**whole system**" approach and **European care pathways, that connect seamlessly regional, national and cross-border healthcare services and infrastructures.**

- If they are not well connected with the existing healthcare infrastructures in each country, people with rare and complex conditions will never be able to fully benefit from this infrastructure.

One fully integrated system with connected, infrastructures to ensure timely access to specialised healthcare

National Health Systems

In the majority of countries, Expert Centres may provide adequate healthcare to the patient population affected by the more prevalent rare diseases, either at regional or national level (<390 rare diseases, affecting 98% of the RD population).



European Reference Networks

Especially valuable to cater for less frequent and lower prevalent diseases, offering virtual expert advice and knowledge sharing to diagnose and treat persons with a rare disease.

In person CBCH where needed

When a patient needs to travel to another country.

RD patients report delays in access, mostly due to financial, language, mobility barriers and difficulties to secure prior authorization given the lack of expertise in a country. Ref: EC evaluation CBHC Directive, staff working document

Leaving no one behind: what's missing?

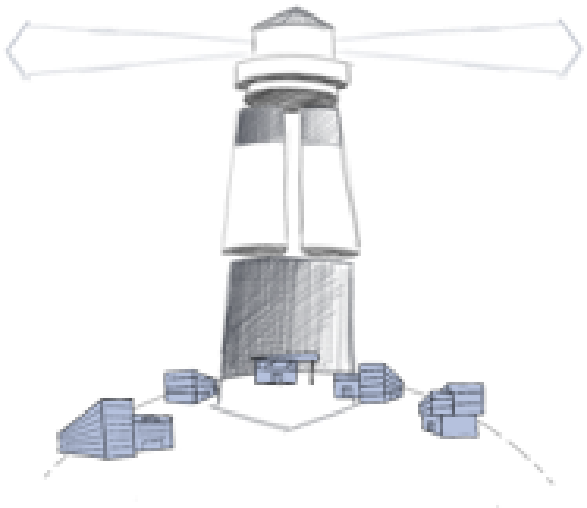
Difficulties to access highly specialised CBHC are further exacerbated when one or more of the following factors emerge - the 3 are usually interlinked:

1. **Very small numbers of patients** and/or low incidence across the EU.
2. **Safety & clinical viability are compromised** because there are very few expert teams across the EU with the clinical competency required to secure sufficient experience to provide a safe and sustainable highly specialised healthcare service or centres qualified to administer an innovative therapy.
3. **Limited financial capacity of individual MS**: Individual MS lack the financial and innovation capacities required to provide and maintain high-cost, highly specialised and innovative healthcare services, including workforce training & adequate replacement rates.

Keeping in mind the fundamentals of healthcare services planning and the aspiration of Universal Health Coverage

Healthcare services are best organised as close to the population as possible, where decision makers are best positioned to understand and meet local population needs …

… this principle is also true for rare diseases, but healthcare planning is more efficiently organised at **national or supra-national** level where there is a sufficient number of cases to understand the needs of this patient population.

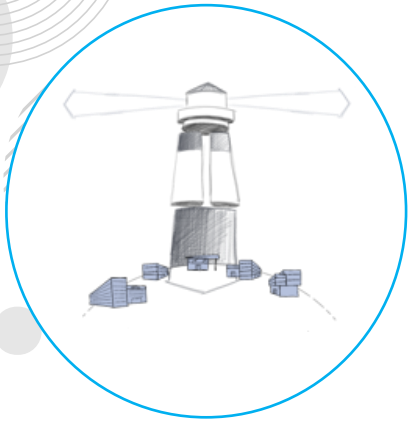


Different approaches, infrastructures and organisational arrangements are required to address rare disease healthcare needs, depending on 3 factors:

1. Prevalence and incidence rate of a given disease or a given intervention
2. Number of expert teams available to provide the service
3. Financial implications for countries to arrange services at a national or sub-national level and the innovative capacity of individual health systems

Closing the gap: European commissioning of highly specialised healthcare services for very rare & complex

By establishing an **EU system to plan, including work force planning, fund, commission and contract highly specialised healthcare services** from leading expert centres recognised as “European Rare Disease Lighthouses”, connected to national, ERNs, and International networks to deliver highly specialised interventions for:



- people living with a disease that affects fewer than 500 people across the EU
- people that require complex interventions where the number of procedures performed each year at EU level is below the above threshold
- people that require innovative treatments that are/will be typically delivered in a few centres across the EU.

Only through greater solidarity and enhanced cooperation in this area, EU countries will be able to organise and manage certain highly specialised healthcare services on an optimal population size, to ensure timely, safe, affordable, accessible and sustainable high-quality care for all.

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How to close the gap?

Eligibility criteria

People living with a disease that affects <500 people across the EU

People that require complex interventions <500 procedures/year performed in the EU level

People that require innovative treatments that are/will be typically delivered in a few centres across the EU.

European planning, funding and commissioning

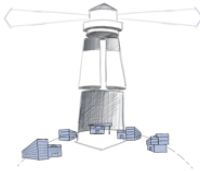
Joint service planning

European fund for highly specialised healthcare services

European procurement to contract healthcare services from European Lighthouses

Coordinated evaluation and monitoring system

Service delivery



European Expert Centres/Lighthouses

Shared agreements with national HCPs for follow-up

Treatment eligibility panels

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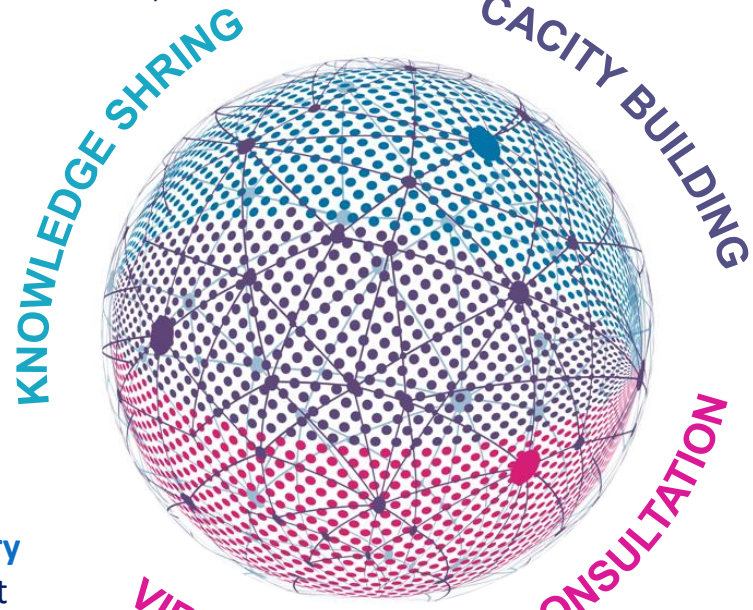
WHO-hosted Global Network for Rare Diseases (GNRD)

To improve the rare disease **patient journey**, ensuring equitable access to diagnosis, healthcare services, and psycho-social care for PLWRD.

To promote **capacity building**, **knowledge sharing**, and **collaboration** among stakeholders to strengthen services for PLWRD at national level.

To provide technical **support to primary care** and frontline services and support access to specialist advice, diagnosis, and treatment to ensure that the expertise travels rather than the patient



WHO & RDI

Collaborating to establish a unified vision for the development of GNRD, Rare Diseases International (RDI) and **WHO** have formalized their commitment by signing a Memorandum of Understanding, thereby enhancing their collaborative efforts.

PARTICIPANTS

- Actively and internationally working in the field of RDs
- Existing **networks** or collaborations of **expert centres**
- Member States, intergovernmental organizations, NGOs, patient associations, hospitals and academic institutions, private sector and philanthropic foundations

WORKING GROUPS

- Coordinate joint activities
- Initially established according to most relevant **RD disease clusters**: **Need-led** Dimension; **Clinical-led** Dimension

REGIONAL & GLOBAL HUB(s)

Hubs of Expert Centres & Patient Organisations to connect with healthcare systems under a **hub model** to support strengthening of local systems in rare diseases.

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Thank you!