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Direction générale de l'offre de soins

French Ministry of Health and Prevention

A new goal-based and coordinated strategy for rare diseases

How to bring together existing EU strategies and actions in a comprehensive framework?

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ERN BOARD OF MEMBER STATES, Fr Representative

Why could patients benefit from the data collected by the ERNs and the ERN registries ?

Population

- 30 million people across Europe
- 6000 diseases

Since *2009 Council Recommendation on an action in the field of rare diseases :*

→ Improvement of diagnosis, treatment and care for rare diseases through technological and scientific progresses ;

→ Creation of 24 ERNs.

High unmet needs

- An average of 5 years needed to obtain a diagnosis
- 6% of rare diseases benefits from a specific treatment

Major issues

- Scarcity of knowledge and expertise
- Psychological, emotional and financial burden
- Vulnerabilities of patients exacerbated by the COVID-19 pandemic
- Pace of change unequal across the European continent

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1er janvier 2022 – 30 juin 2022

The Ministerial Conference organised by the French EU Presidency on rare diseases on 28 February 2022 underlined the willingness shared by several Member States, patient organisations and industry to go further than what has already been done.

A number of ideas were put forward to ensure that all European patients receive prompt and fair treatment:

Proposal for a policy framework for rare diseases: best practices and to steer innovations

→ **Necessity to introduce a new comprehensive policy framework for rare diseases: a European Action Plan for rare diseases.**

Objective 1

- **Drive change through innovative European collaboration** : patient needs-led research, rare disease innovation and works for accessibility, affordability and sustainability of the lifecycle of medicines.

Objective 2

- I**mproving patient and families' care pathways:** by prioritising earlier, better and more accurate diagnosis, full inclusion in society and delivery of highly specialised healthcare and integrated person-centered care.

Opportunities to reach these objectives

- **Data sharing** : adopting common quality standards to guarantee and facilitate data sharing, introducing artificial intelligence…
- **Multi-stakeholder partnerships** : between European countries, European experts, patient organisation…
- Consolidation of **European Reference Networks (ERNs)** and integration into national healthcare systems

A plan structured around measurable goals

The plan would reach out and guarantee appropriate and effective care for the vulnerable population of 30 million people in Europe living with a rare disease.

A longer life …

- **Ensuring earlier, faster and more accurate diagnosis** through better use and accessibility of diagnostic technologies and best practices ;
- **Promoting universal access to healthcare** through more efficient patient pathways and cross-border healthcare in order to ensure access to appropriate expertise ;
- **Introducing a holistic approach to care**, considering all aspects of the person's medical and social care.

Rare diseases often cause premature death: 70% of rare diseases affect children, and delayed diagnoses prevent access to timely interventions.

… and a better life …

- **Reducing** psychological, social and economic **vulnerability** ;
- Ensuring 50% of all persons living with a rare disease have **access to transformative therapies** ;
- Supporting **better access to both education and employment** for people living with a rare disease, as well as their families and carers.

Every person living with a rare disease, has the right to modern and comprehensive care achieving not just the absence of disease, but a state of complete physical, mental and social well-being.

Europe's Action Plan for rare diseases would introduce measurable goals to reduce inequalities faced by people living with a rare disease within countries, and between countries

Holistic approach to the person's needs through improved access to high quality, integrated medical and social care is the basis of the new policy framework.

- Reduce psychological, social and economic vulnerability of people living with a rare disease and their families by ⅓,:
  - favouring a more integrated approach to allow families to get support, including financial and emotional support, including through ERNs. The entire family is impacted by the challenges of living with a rare disease, increasing isolation and impoverishment (SDG 1);

- Ensure 50% of all persons living with a rare disease have access to transformative therapies, which could significantly improve their quality of life (SDG 3);

- Support better access to both education and employment for people living with a rare disease, as well as their families and carers.

The rights of quality education (SDG 4) for children, teenagers, and young adults affected by a rare disease are not respected and inequalities in terms of social inclusion and equal opportunities strike very early in life.

Young adults experience difficulties with every step of independent living, from finding, keeping or returning to a decent work (SDG8, 10).

How could patients benefit from coordinated plans at national level ?

National Plans: access to care and expertise for all

Visibility

PNMR 1

2004 — **2008**

the RDs became a public health issue
- 131 labelled CRMR in 120 French Hospitals

Coordination

PNMR 2

2011 (16) — **2014**

Structuring of FSMR
Work on the establishment of the BNDMR
Rare Disease Foundation

Sharing

PNMR 3

2018 — **2023**

4th, July 2018
Third Rare Disease Plan (PNMR3)

RD Policy: Integration into national healthcare systems

The 3rd national plan for rare diseases in France: expectations and actions regarding the data collected by the ERNs and the ERN registries

- To create a dynamic observatory for the monitoring of patients “without a diagnosis”

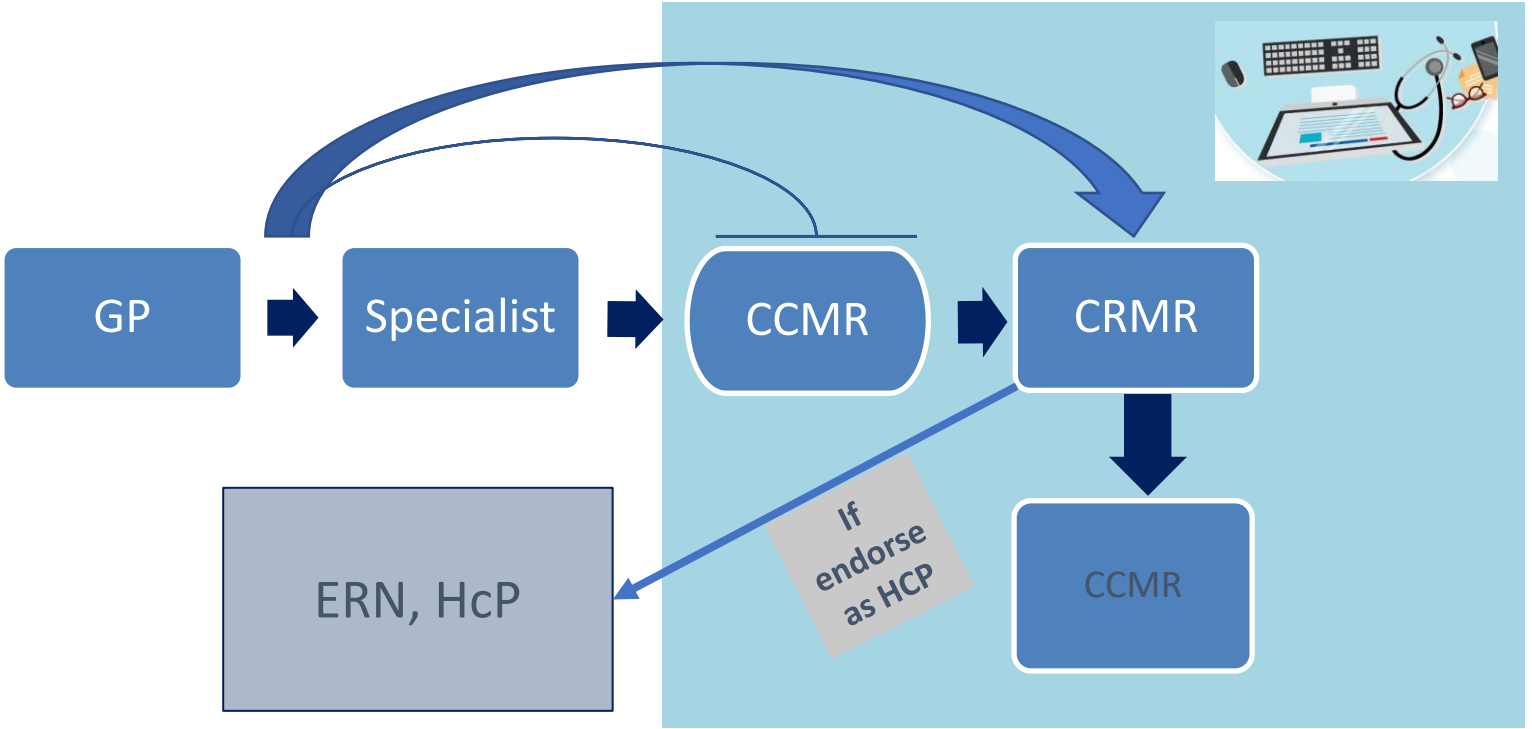
- To boost innovation: more efficient access to treatments, with disease-based approaches instead of drug-based ones and amplification of the drug repositioning process

- To reinforce education and training: better sharing of teaching tools and the development of patient education in the field of rare diseases

- To encourage patient support: the promotion of quality for key moments in the care pathway, personalised medical-social, education and professional mecanism

- The plan also reflects an ambition to significantly improve the care of patients with rare diseases in French overseas regions: question of equity

Rare diseases stakeholders collaboration to ensure that the ERNs meet a clear patient pathway
Need to integrate and sustain EU and national plans and strategies for rare diseases on a long-term basis



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Drafting of the Road Map : a plan structured around measurable goals

The plan would reach out and guarantee appropriate and effective care for the vulnerable population of 30 million people in Europe living with a rare disease

… thanks to innovation

- Foster the **approval of approximately 1000 innovative treatments** for people living with a rare disease ;
- **Increase investment** in rare disease innovation by 200%, across basic, clinical, social and translation research ;
- **Ensure a better regulatory environment and improved market access**, through strengthened involvement of the regulator, evaluator and payer in clinical investigation at an early stage, and data collection networks for rare diseases.

Only 6% of rare diseases have a dedicated treatment, it is far below the huge potential of scientific advancements. The rare disease field suffers from the scarcity of knowledge and expertise. Europe's Action Plan for Rare Diseases would foster resilient infrastructure, promote inclusive and sustainable industrialization and encourage innovation.

Specific objectives JA ERN Coordination

Infrastructure for data sharing across the EU with the development of European-wide patient registries

ERNs

- An extension of national healthcare systems
- To provide an additional dimension of EU-wide networked care in addition to that provided at national level
- A paradigm shift in their respective fields of clinical excellence but also made important steps towards a sharing of know-how around rare diseases between a large number of medical specialities across the EU in a short time
- Concerted actions from the very first days of the COVID pandemic are probably the best proof of how this has worked.

A vision on how access to ERNs data should be organised to address our needs:
With the ERNs, the need for a real-life data collection system at the EU level = attractivity of the European R&D ecosystem for rare diseases

New therapies are arriving on the market earlier and earlier:

- Need to assess their value **early** on and then **on an ongoing basis**;
- Clinical data **not yet mature enough** to clearly demonstrate therapeutic value;
- **Difficult to recruit patients** for orphan indications: small numbers, geographical dispersion;
- **Long-term clinical benefit** difficult to measure, e.g. gene therapies;

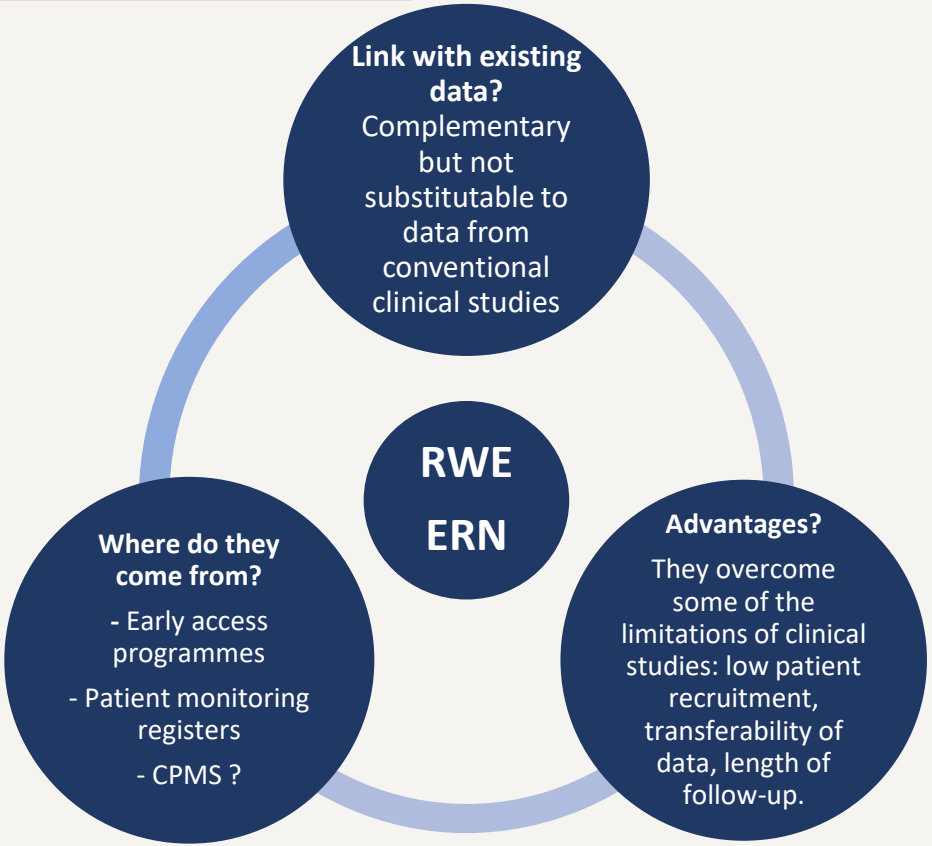
Consequence: difficulties in medico-technical evaluation and then in pricing

→ ***Need to strengthen existing mechanisms for rare and ultra-rare diseases, in particular concerning the collection of real-life data at a European level***

Advantages of introducing such measures :

- Establishing EU-wide real-life monitoring of treatments;
- Facilitating data collection;
- Improving the quality of this data;
- Strengthening research and development in rare diseases.

Real World Evidence - RWE



RWE allows health authorities and payers to

- Obtain **additional information** after bringing to market;
- **Re-evaluate the benefit** of a product, clarify its place in the therapeutic strategy;
- Enable the implementation **of innovative financing models** or the **product price revision** ;
- Obtain a marketing authorisation in a niche indication in the case of a **use outside the scope of the marketing authorisation** ;
- Have a **more global understanding** of the care pathway of rare disease patients.

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