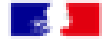


Advancement of Treatments FOR RARE DISEASES



MINISTÈRE
DE LA SANTÉ, DES FAMILLES,
DE L'AUTONOMIE
ET DES PERSONNES HANDICAPÉES

*Liberté
Égalité
Fraternité*

Dr Anne-Sophie Lapointe – Rare Diseases Mission

French Ministry of Health

Rollout of efficacious therapies: access and infrastructure considerations

Infrastructure considerations for delivering therapies at scale

France's example

Organisers:



DEPUTY MINISTRY OF
RESEARCH, INNOVATION
AND DIGITAL POLICY
REPUBLIC OF CYPRUS



CY2026.EU
CYPRUS PRESIDENCY
OF THE COUNCIL OF THE EU



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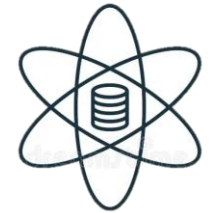
Rare Diseases, Rare Policies: A French Model of Evolving Commitment

From care to inclusion, France builds dynamic strategies shaped by evaluation, innovation, and collective action.

France has chosen **not to pursue a one-size-fits-all approach to rare diseases**, but rather to develop an **evolving policy cycle**—anchored in key pillars such as care, *diagnosis, research, support, and inclusion*.



This ambition reflects a **unique determination to design rare policies for rare diseases**—tailored to the **needs of patients**, aligned with the **expertise of healthcare professionals**, and *shaped in partnership with associations*.



This strategic vision rests on a **continuous process of assessment and renewal**: **each national plan builds on the lessons of the previous one**, ensuring that future policies are better adapted to the complex and changing realities of rare diseases.



By fostering ongoing **dialogue between all stakeholders**, France ensures that each step forward is **both evidence-based and deeply grounded in the lived experience of those affected**.

Rare diseases plan: a rare diseases voluntarist policy for an holistic approach from care to research: the frame for delivering therapies



**AXIS 1: IMPROVE THE LIFE
AND CARE PROCESS**



**AXIS 3: PROMOTE ACCESS
TO TREATMENT FOR
RARE DISEASES**



RARE DISEASES



**AXIS 2: FACILITATE AND
ACCELERATE
DIAGNOSTICS**



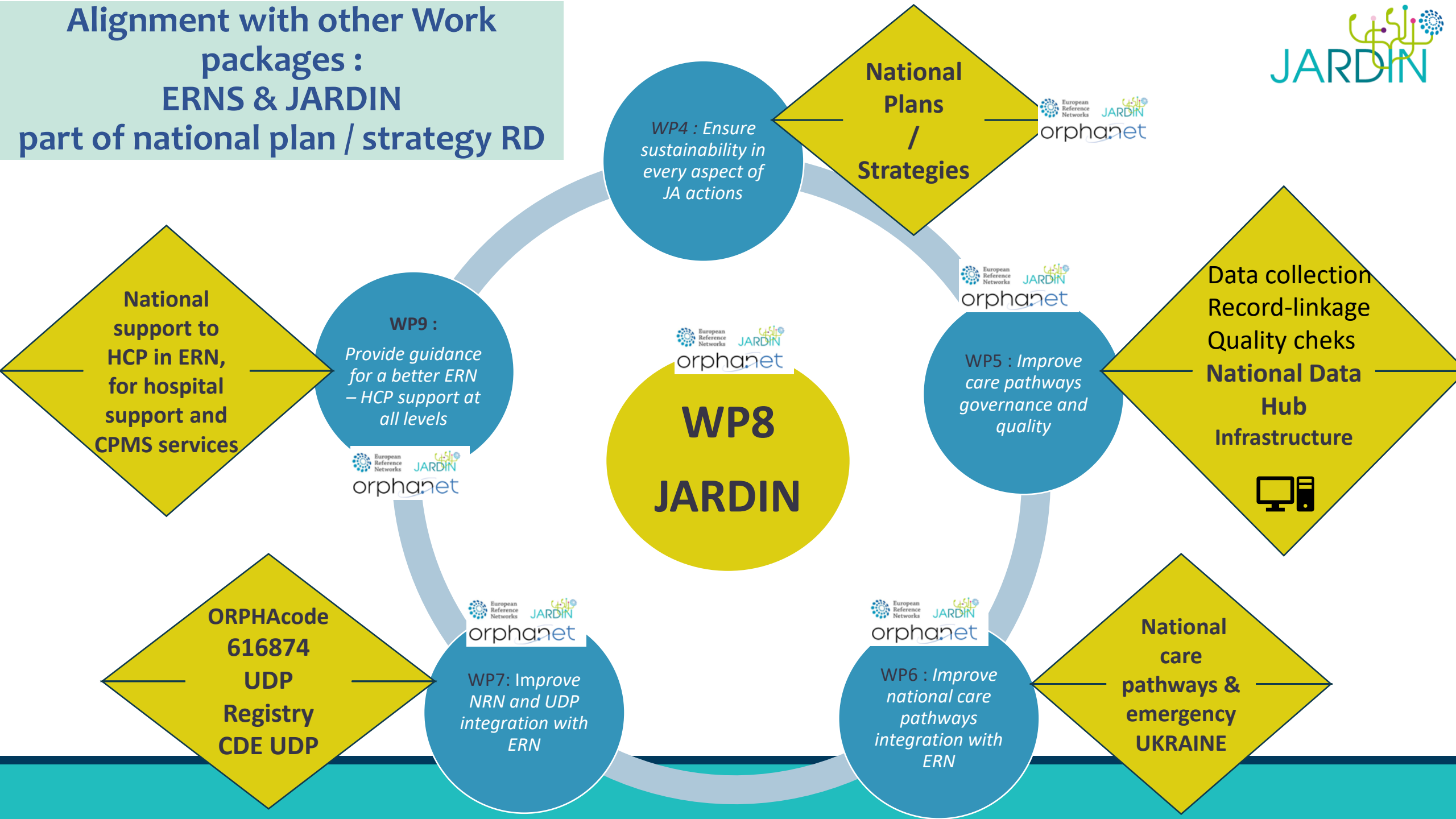
**AXIS 4: DATABASES AND
BIOBANKS:**



**IMPROVE THE LIFE AND
CARE PROCESS**

European Reference Networks  

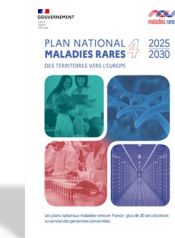
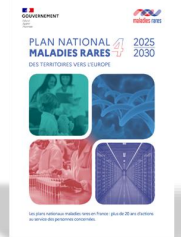
Alignment with other Work packages :
ERNS & JARDIN
 part of national plan / strategy RD



IMPROVE THE LIFE AND CARE PROCESS:
clear care pathway contribute to enhance the recruitment for clinical trials

➤ **Sustainability of ERNs and JARDIN perspectives**

- Improving access to care in expert centers.
- Enhancing coordination between hospital structures and outpatient care.
- Optimizing healthcare pathways for patients.



▪ This axis includes measures to:

- Strengthen therapeutic education** for patients.
- Improve key transition periods**, including **adolescence, aging, and pregnancy**.
- Leverage digital tools** to support **care coordination** and enhance patient management :

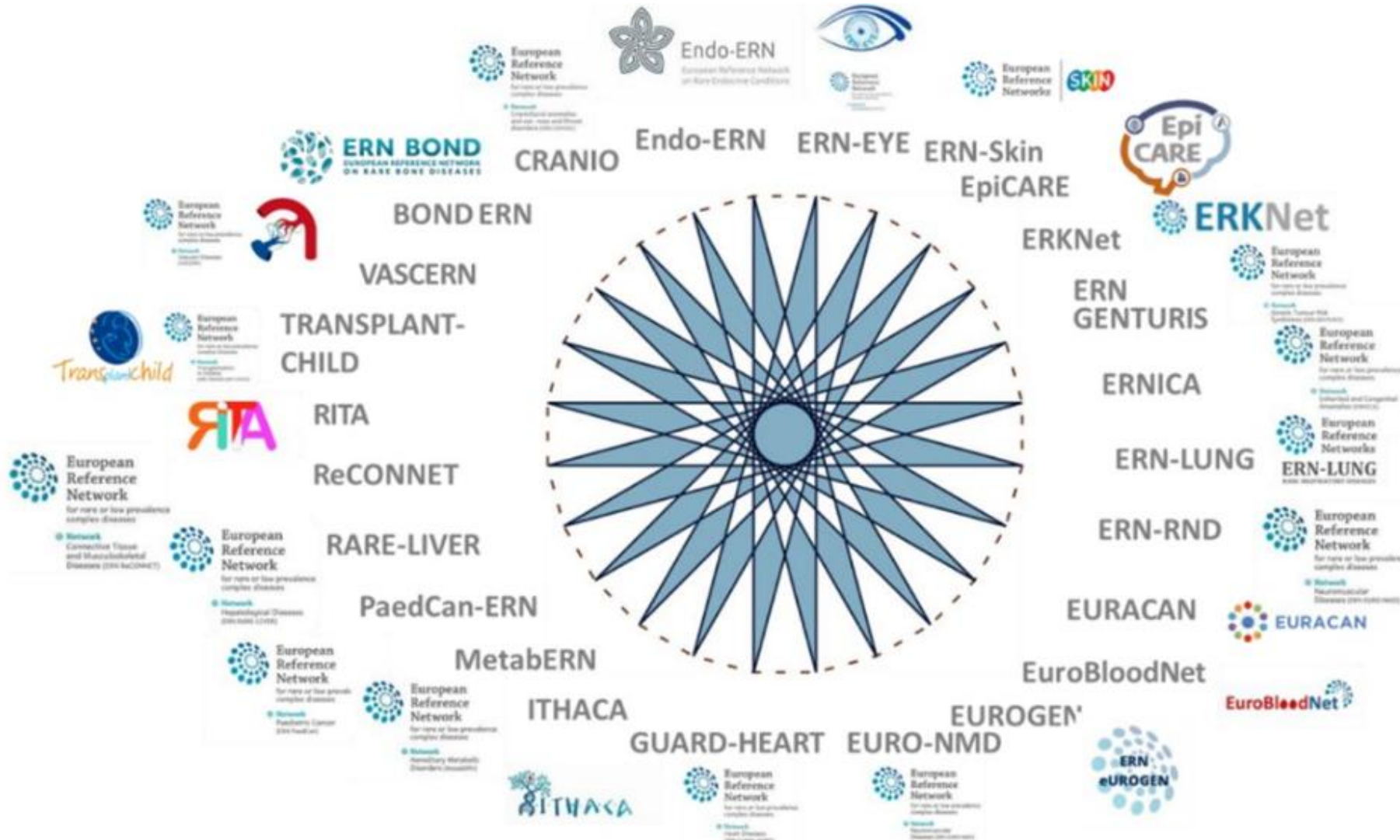
PROMS



Strong EU Rare Diseases network

24 ERNS which cover 90% of rare Diseases :

a « game changer » with these EU coordination



IMPROVE THE LIFE AND CARE PROCESS: clear care pathway contribute to enhance the recruitment for clinical trials : alignment at national and european levels



Action 8.1: Supporting European Reference Networks (ERNs) and Their Integration into the National Healthcare System



- Strengthen cooperation between ERNs, national rare disease networks, and European expert centers (Health Care Providers - HcP). **Where is the patient ?**
- Implement data collection mechanisms at national level to contribute to ERN registries. **Which data ? How is the natural history of the diseases ?**

Action 8.2: Integrating National hub for rare diseases into European Strategies and Recommendations

- Develop specific, interoperable datasets aligned with ERN standards within national data base for rare diseases. : **Sufficient amount of data at European level**
- Encourage the participation of national hub for rare diseases in European research programs.
- Simplify administrative processes and provide training on European tools to enhance engagement.



Strong focus on 3 elements :

- PNDS - National Diagnostics and **Care Protocols** (Work Package 6)
- Care Pathways (Work Package 6)
- CPMS (Work Package 8 & 9)



Launch of the Open-source repository

Goal

Allow external stakeholders to use and adapt CPMS 2.0
Create a community around CPMS 2.0

Ongoing

Repository is ready to go online. Waiting communication decision.
Preparatory steps for the creation of the community



**FACILITATE AND
ACCELERATE
DIAGNOSTICS**



FACILITATE AND ACCELERATE DIAGNOSTICS



Improving Diagnosis to better have access to treatment

the **National Plan / Strategy** strengthens **diagnostic observatories** and promotes the **integration of new diagnostic methods, particularly in genetics in experts centers.**



According to **Philippe Baptiste**, Minister of Higher Education and Research:

“Technological innovation, particularly through the use of genomic sequencing tools, plays a key role in reducing diagnostic wandering for patients.”

As part of this effort, the rare diseases plan also supports:

- ✓ Acceleration of diagnostic procedures.
- ✓ Precision Medicine and Genomic therapeutics
- ✓ Training of healthcare professionals in this field

FACILITATE AND ACCELERATE DIAGNOSIS

Focus on links with JARDIN

■ Reinforce coordination with Rare Diseases National Hub



- Implementation of a Mirror JARDIN working group for all the 23 national Hubs , in duo with JARDIN project management

■ Work in close collaboration with the Undiagnosed Diseases Program in Rare Diseases (Work Package 7)

- Strong links with Task 7.2 : undiagnosed programs in rare diseases



- Ex : dissemination of specific OrphaCode for undiagnosed people in national and European registries

ORPHA:616874 Rare disorder without a determined diagnosis after full investigation



PROMOTE ACCESS TO TREATMENT FOR RARE DISEASES

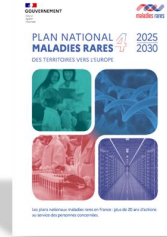


PROMOTE ACCESS TO TREATMENT FOR RARE DISEASES

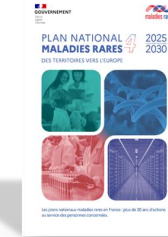
➤ For national plan and strategy scientific research at the heart of its strategy.

Structuring therapeutic research

- Encouraging the repositioning of existing drugs
- Organizing real-world data collection to better evaluate treatment effectiveness
- It includes measures to:



- ✓ Optimize information systems
- ✓ Develop artificial intelligence to enhance data utilization



An opportunity to foster closer collaborations between public research and industry, transforming scientific discoveries into concrete solutions for patients."

AXIS 3: PROMOTE ACCESS TO TREATMENT FOR RARE DISEASES

Focus on links with JARDIN : access in the future to integration and innovation with new treatment



➤ Focus on Work Package 8 : Data Management

• Task 8.1

Identify current barriers to RD data sharing and inventory of existing solutions



• Task 8.2

Implement solutions to improve semantic accuracy and interoperability of RD health data



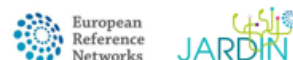
• Task 8.3

Propose and develop implementable solutions to overcome organizational, technical and legal barriers to integration of national health systems and ERN data management



• Task 8.4

Test and implement solutions in agile mode



• Task 8.5

Visualize RD Expert centres with ERN & NRN





DATABASES AND BIOBANKS



DATABASES AND BIOBANKS: the infrastructures needed to reinforce the proof of the benefit of new treatments

➤ Strengthening a Territorial and European Approach

Expanding territorial healthcare coverage aims to ensure that all patients have access to high-quality expertise, regardless of their place of residence.

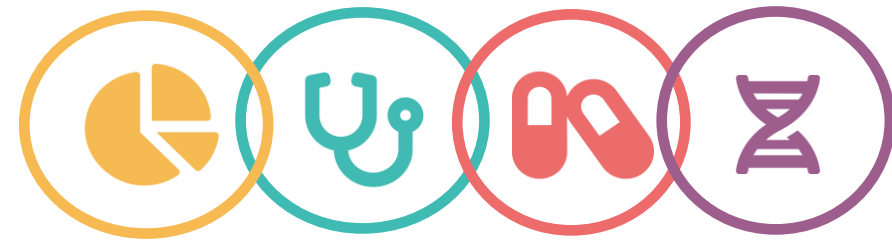
- At the European level, 27 EU countries + Norway need to coordinate initiatives to:

Improve rare disease monitoring
Harmonize best practices
Optimize resource-sharing for patient care



This includes efforts to collect, use, and consolidate health data while leveraging artificial intelligence for better analysis and decision-making.

National plan for rare diseases: European approach to enhance data sharing and utilization : some solutions through the JARDIN Joint Action, launched in February 2024, which aims to boost health data exchange among experts centers.

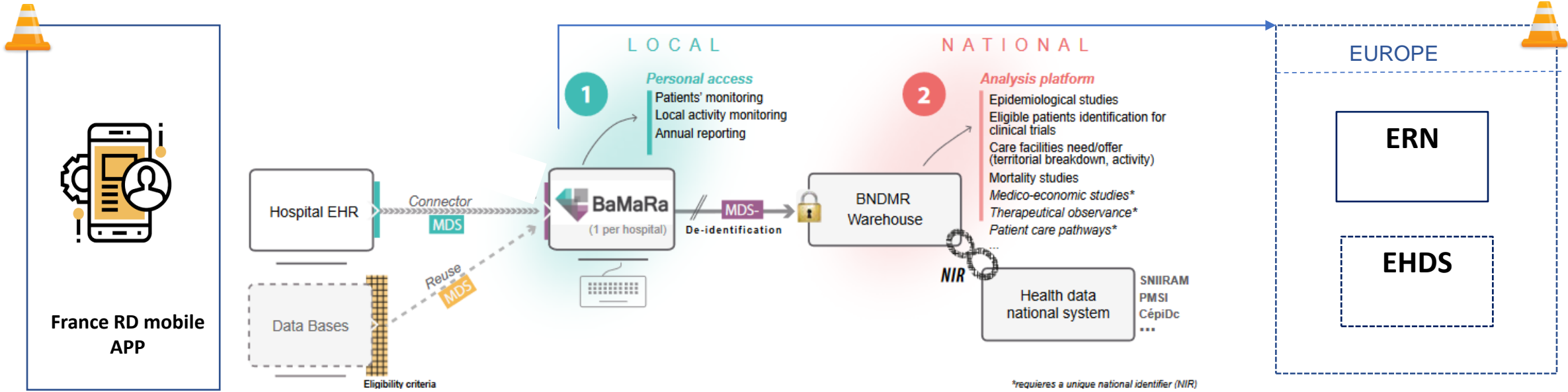


BNDMR

Banque Nationale de Données
Maladies Rares

Organization & Minimum Datasets Layers Strategy @ BNDMR

French National Rare Disease Data Registry (BNDMR)



Patient use

Local level

Hospital (Data Controller)- APHP (Data Processor) contract.

Scope of the contract: RD labelisation centres by the DGOS

- Primary use
- Health Data Hosting (HDS) by APHP

Patient information: public wall display by each hospital

National Level

Data Sharing Agreement Hospital-APHP (APHP as data controller)

Secondary use

BNDMR Charter

IRB for research

Individual information notice for patients- Opt-Out

Transparency portal

French DPA (CNIL) autorisation or CNIL reference framework for Health Data Warehouses (EDS) Compliance

ERN-HCP (Joint Controllers)
Data Sharing agreement

ERN patient informed consent (OPT-in)

RD.CDE transfert from BaMaRa by each HCP

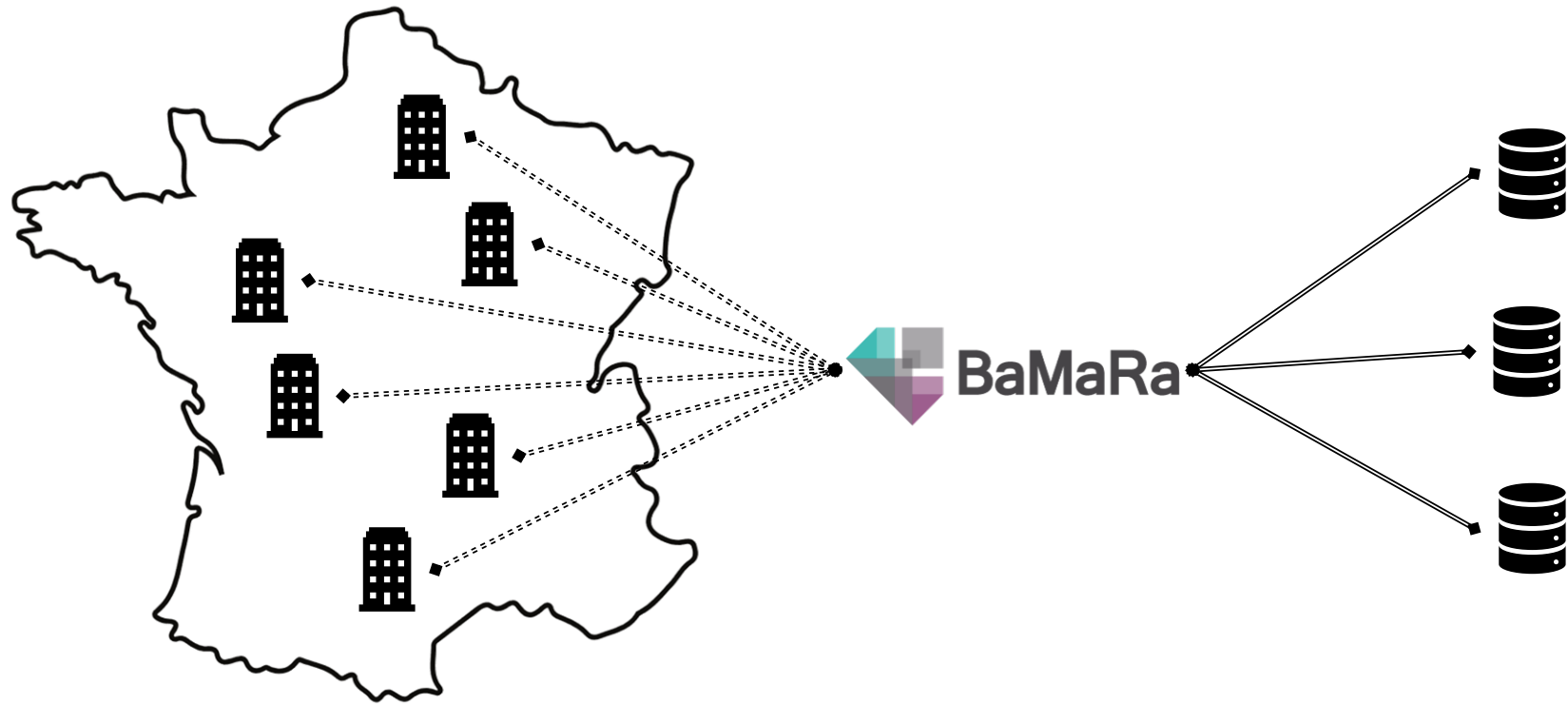


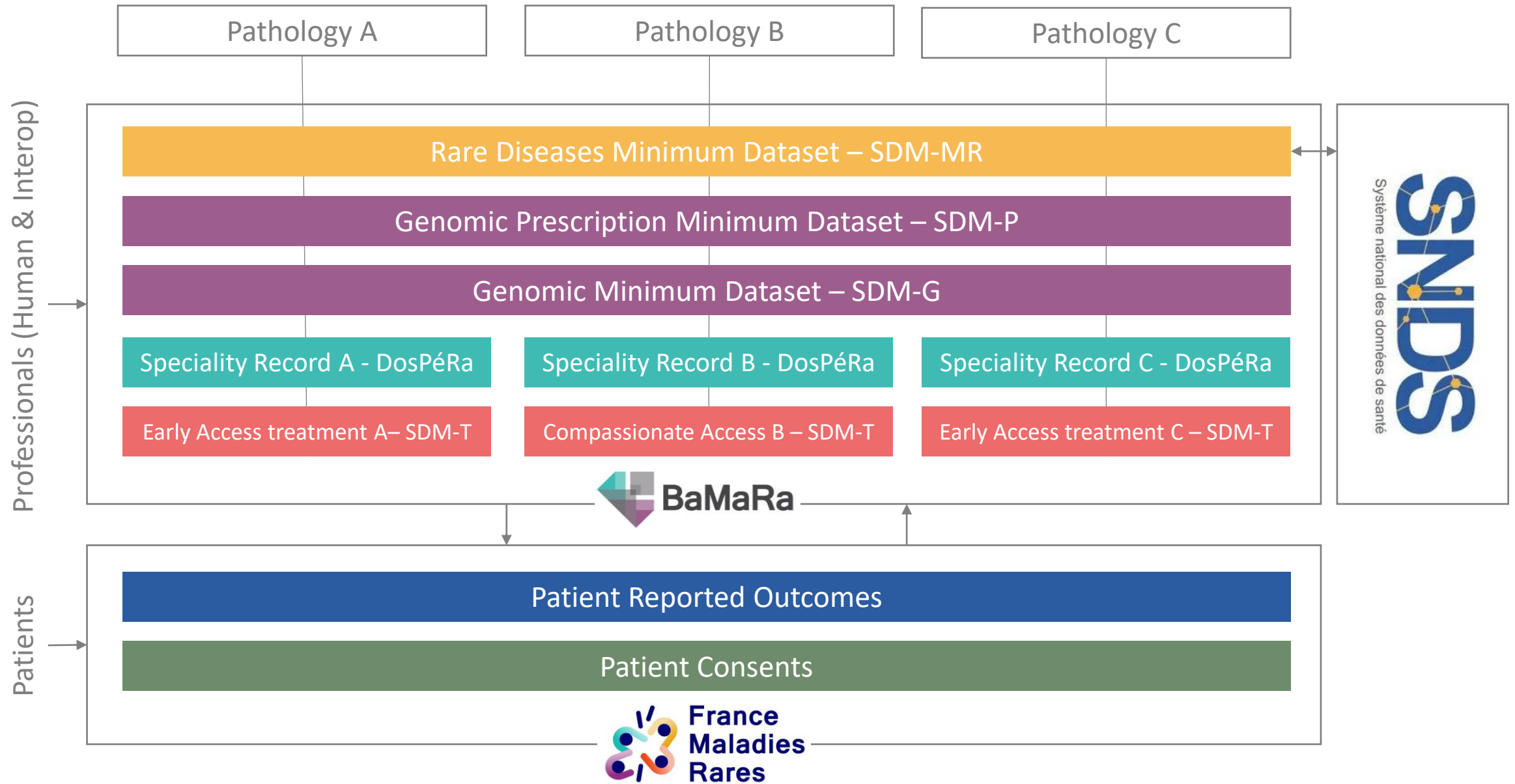
Centralized and standardized process for data sharing agreement

- In France, there is already 1 agreement between each institution (hospital) and BaMaRa, which will only need to be amended to cover the ERNs.
- There will then be “only” 1 “agreement” per ERN
- The DSA signed between the AP-HP (legal entity of the BNDMR) and the legal entity of the ERN.

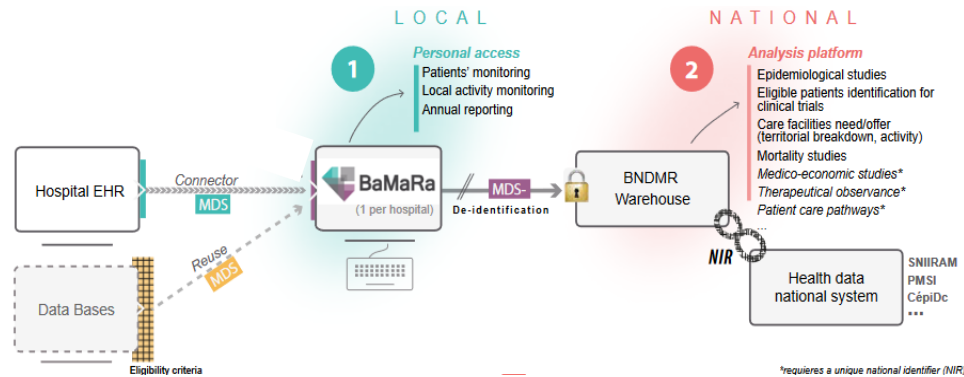
Hospital-BaMaRa agreements (APHP)
(Already exist: simple amendment)

DSA APHP-ERN
(Only one to sign by ERN)



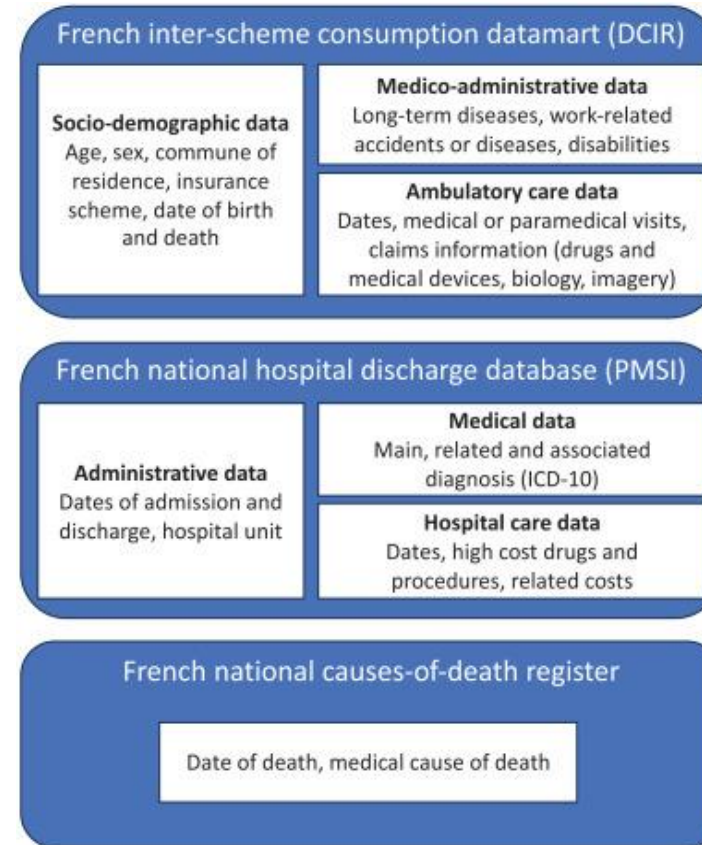


A further step for data reuse of rare disease data: a complete follow-up over 15 years using national claims data



How to link the two databases?

- Direct linkage: national social security number
- Indirect linkage: using their common variables
 - Birthdate (M/Y) and sex
 - City of residence (city/department)
 - Activity dates (D/M/Y) and context (hospitalization, consultation or other);
 - Hospital Number (FINESS system)

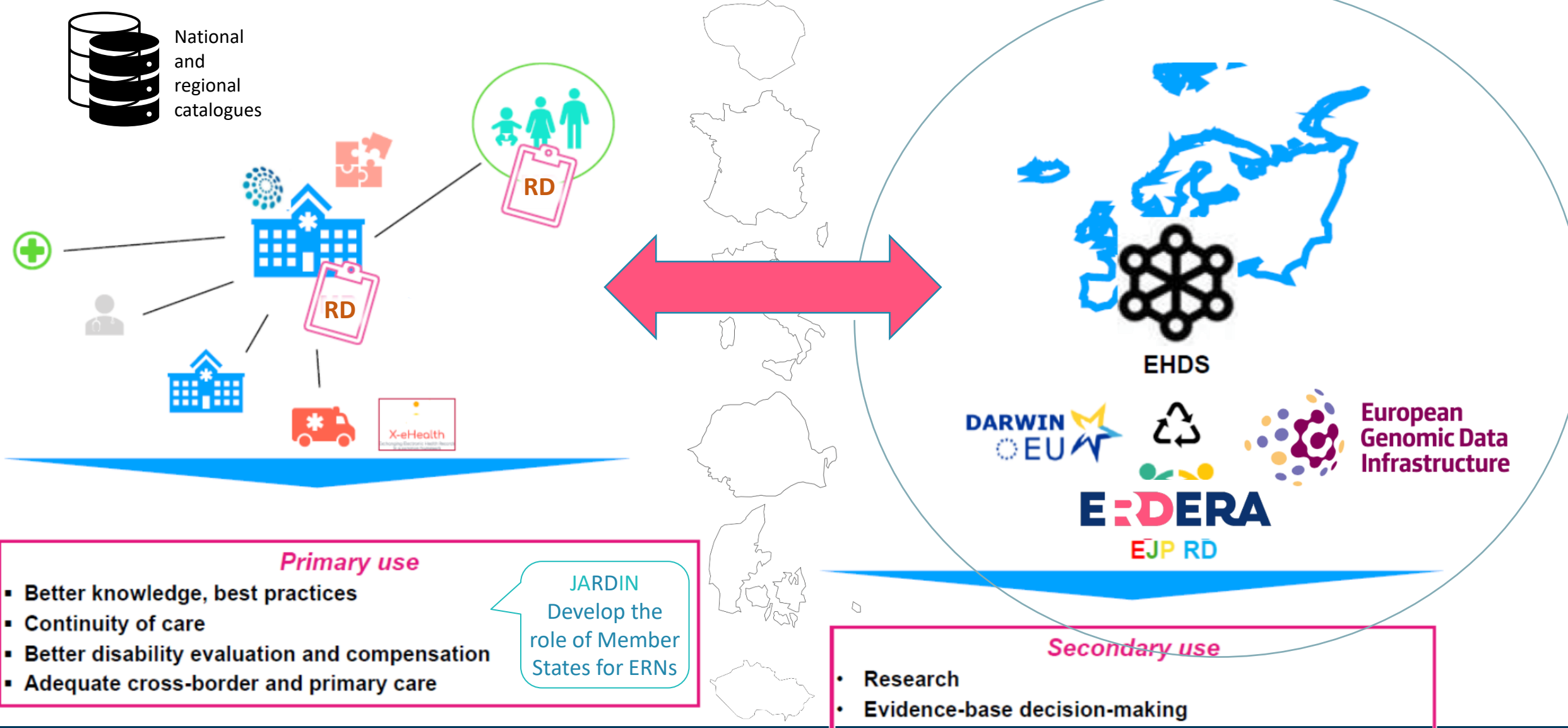


ICD-10: International Classification of Diseases-10th revision





The future we want: RD in the data ecosystem



EMA reflections on how to use real world data for evidence generation



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> Development of a reflection paper on the use of external controls for evidence generation in regulatory decision-making guideline

Establishing efficacy based on single-arm trials submitted as pivotal evidence in a marketing authorisation

Development of a reflection paper on the use of external controls for evidence generation in regulatory decision-making - Scientific guideline

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ICH M15 guideline on general principles for model-informed drug development - Step 2b - Scientific guideline

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This [guideline](#) defines an harmonised framework for evaluating MIDD evidence and provides recommendations for related planning and regulatory interactions, implementation, reporting, and submission.

JARDIN : what's next ?

Why JARDIN 2.0 is strategic for the future of ERNs, the integration in national rare diseases plans and rare cancer policies

Strategic bridge between national plan and ERNs

After ? A new European Plan for rare Diseases to better integrate innovation in care and research pathways?



European
Reference
Networks

Challenge : Coordinate rare disease policies with stakeholders in biotechnology and health data in conjunction with eHDS, DARWIN, the implementation of the Biotech Act, pharmaceutical legislation and medical device legislation in order to support the development of new treatments, particularly when no commercial model exists.

Solution : JARDIN integration mechanism

- Align ERN participation with national governance / plan & strategy rare diseases and cancers
- Embed ERN pathways into national referral systems
- Structure interoperable data flows
- Ensure sustainability beyond EU project funding

MFF 2028–2034 – Health challenges

Findings

- Health is a pillar of resilience, sovereignty, and cohesion
- The 2028–2034 MFF redefines the architecture of European funding
- Need to preserve a clear and visible health ambition



Challenges

- **How we work together ?** in a way to demonstrate that the 2028–2034 MFF consolidate the European Health Union as a pillar of resilience
- Crises have demonstrated the value of coordinated European action
- Health is becoming a matter of sovereignty and cohesion : European plan for rare disease ? **What we want after JARDIN ? In a way of maintaining continuity of effective policies**
- Rare diseases offer a consensual framework for action
- France supports a clear and visible health ambition : WP8 Jardin cp-coordination with NL, 7 ERNS, European HTA work, biotech act, ...

Key message

- Rare diseases are a showcase of the European Health Union: an asset for international clinical research and care pathways based on strong foundations with 24 ERNs:
 - Data structuring
 - Organization of clinical trials
 - Close collaboration with EMA and HTAs
 - Patient involvement in European public policies
 - Facilitated transition from research to clinical practice
 - Economic challenges: production autonomy, academic bioproduction, joint price negotiation
- They combine patient impact, innovation, and sovereignty
- Their funding must be clear and sustainable within the MFF
- **Members from the ERN Board have to work closely with their national point contact who participate at the negotiations for the budget of the future MFF**





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DES TERRITOIRES VERS L'EUROPE



Thanks !

