"I don't understand anything about the rare disease ecosystem. It's a real jungle. I'm completely lost."

"It's not a jungle. You just need the right guide."

"The right guide is right here."

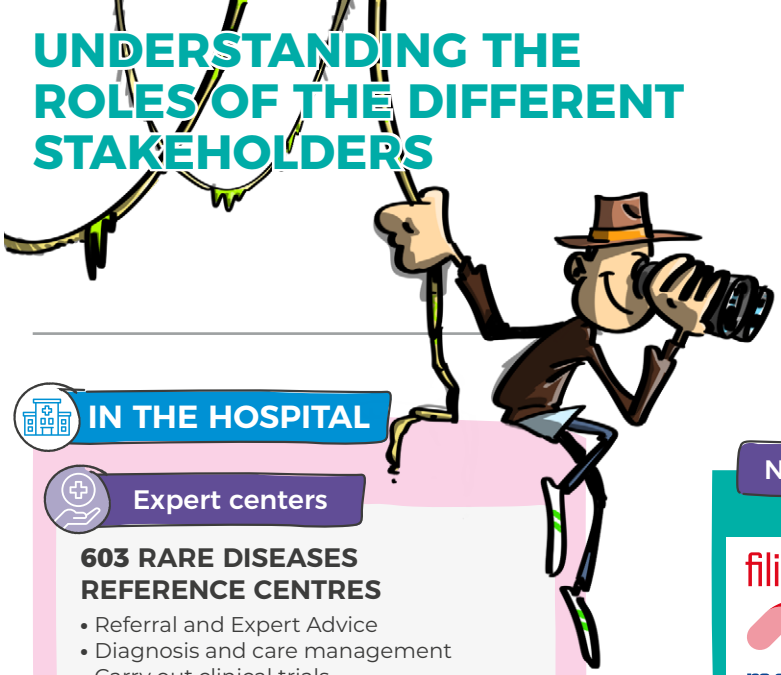
THE FRENCH RARE DISEASES **ECOSYSTEM**

Finding your way among healthcare providers and available resources

filières de santé maladies rares

CMG collège de la médecine générale

UNDERSTANDING THE ROLES OF THE DIFFERENT STAKEHOLDERS



French National Rare Disease Policy

MINISTÈRE DE LA SANTÉ ET DE LA PRÉVENTION
Liberté Égalité Fraternité

Plan national maladies rares 4

Governance

IN THE HOSPITAL

Expert centers

603 RARE DISEASES REFERENCE CENTRES

- Referral and Expert Advice
- Diagnosis and care management
- Carry out clinical trials

Referral of patients with a suspect condition

Complex cases, challenging diagnoses or highly specialised follow-up

Regional or local contact point

Close-to-home follow-up

1708 RARE DISEASES COMPETENCE CENTRES

They ensure territorial coverage across mainland France and overseas territories.

Patient referral

National Coordination

filières de santé maladies rares

23 FRENCH RARE DISEASES NETWORKS

Coordinate Rare Diseases Reference Centres (CRMR) and Rare Diseases Competence Centres (CCMR), in collaboration with patient organisations.

Structure and clarify the roles and actions within the rare disease field.

Information resources by disease group. Contact point for referral and guidance.

Coordination and support

Linking

Collaboration

Local coordination

23 RARE DISEASES EXPERTISE PLATFORMS

They organise the patient pathway between primary care and hospital services.

Sharing of expertise

European coordination

24 EUROPEAN REFERENCE NETWORKS

European networks bringing together expert centres from different countries. Expert consensus on diagnosis, care, treatment, training, etc.

Production of Clinical Consensus Statements

OUTSIDE THE HOSPITAL

Resources

ORPHANET

Reference database on rare diseases and orphan drugs.

▷ **Dissemination of reliable information and referral to expert centres.**

MALADIES RARES INFOS SERVICES

National telephone information and referral platform.

▷ **by phone (freephone number) or email, and through discussion forums.**

Family guidance

ALLIANCE MALADIES RARES

French federation bringing together 280 rare disease patient organisations.

▷ **Patient voice integrated into national actions.**

Research

FRENCH RARE DISEASES FOUNDATION

Financing and supporting scientific and medical research.

▷ **Identification of research priorities and networking of researchers.**

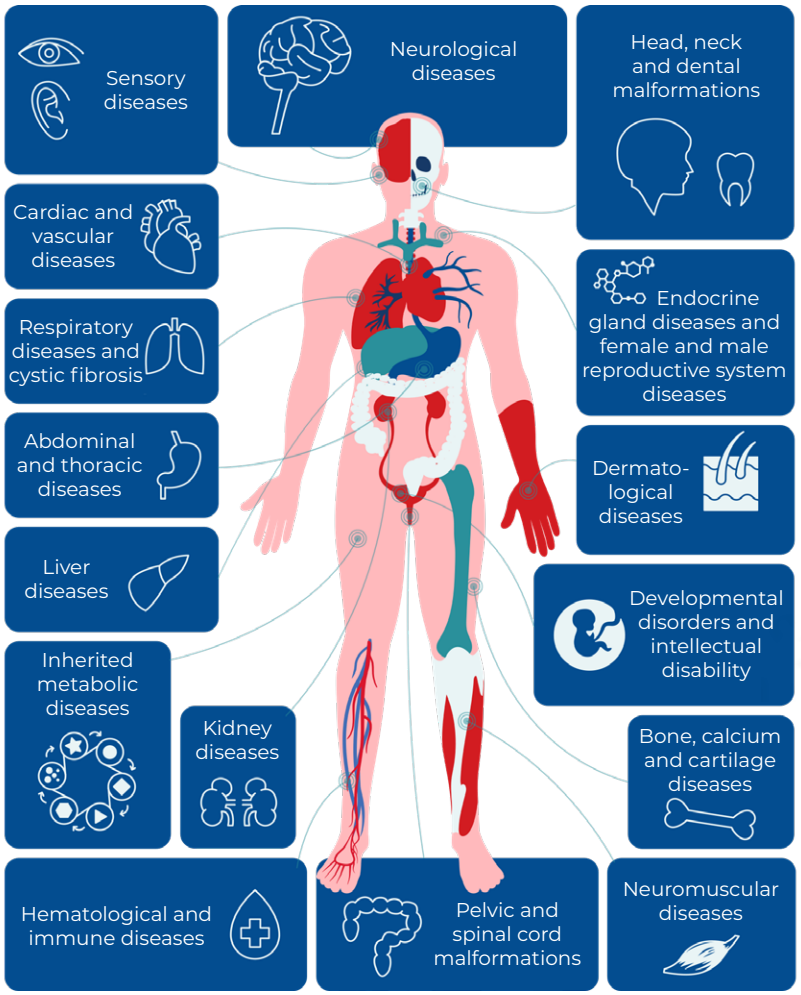
23 FRENCH RARE DISEASES NETWORKS

National coordination and resources

filières de santé maladies rares

List of FSMR and contacts

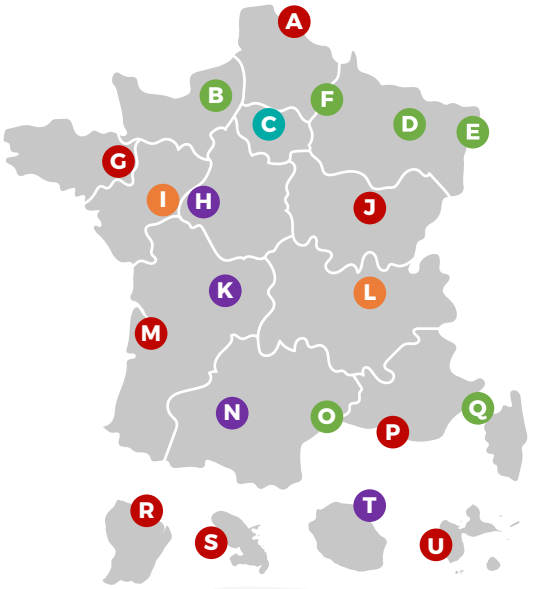
The French Rare Disease Networks coordinate the implementation of the French National Rare Disease Plans at country level. They are care and medical expertise networks organised by major disease groups (e.g. neuromuscular, dermatological, metabolic diseases, etc.).
The networks develop and provide **practical tools and resources** such as national guidelines and protocols, resource directories, diagnostic support tools, training programmes, and more.



Source : filieresmaladiesrares.fr

23 RARE DISEASES EXPERTISE PLATFORMS

Reception, guidance, and integrated hospital support



Rare Disease Expertise Platforms in mainland France and overseas territories (PCOM, Overseas Coordination Platform) are hospital-based structures that complement the network of the reference centres (CRMR) and the rare diseases networks (FSMR). They strengthen coordination between care and healthcare professionals, adapting to local specificities.

Their aim is to bring together **all expertise and services useful for patients with rare diseases** within a given territory: guidance, links with community-based care, support for medico-social procedures, and rapid referral.

PEMR/PCOM serve as practical entry points for general practitioners by providing **a single hospital-based contact person** to facilitate referral, offer specialist and expert opinions, and support care coordination, among other functions.

plateformes d'expertise et de coordination maladies rares

List of PEMR and contacts

- **A** **PLEMaRa** *(Hauts de France, Lille)*
- **B** **NorMaRare** *(Normandie, Rouen)*
- **C** **Île de France**
  - **PEMR Paris Centre** *(Necker)*
  - **ESMARA** *(Henri Mondor)*
  - **PEMR Paris Nord** *(Robert Debré)*
  - **PEPS** *(Bicêtre)*
  - **PEMR Sorbonne Université** *(Armand Trousseau)*

Plateforme Grand Est

- **D** **LARA** *(Nancy)*
- **E** **PARA** *(Strasbourg)*
- **F** **PARC** *(Reims)*
- **G** **RARES BREIZH** *(Bretagne, Rennes-Brest)*
- **H** **PEMR VL** *(Centre-Val de Loire, Tours)*
- **I** **PRIOR Maladies Rares** *(Pays de la Loire, Angers)*
- **J** **PEMR BFC** *(Bourgogne-Franche Comté, Dijon)*
- **K** **NAN** *(Nouvelle-Aquitaine Nord, Limoges)*
- **L** **PEMR AuRA** *(Auvergne-Rhône-Alpes, Lyon)*
- **M** **BexMaRa** *(Nouvelle-Aquitaine, Bordeaux)*
- **N** **MaRaTo** *(Occitanie, Toulouse)*
- **O** **PEMR Montpellier-Nîmes** *(Occitanie, Montpellier)*
- **P** **PEMR AP-HM** *(PACA- Corse du sud, Marseille)*
- **Q** **RESILIENCE** *(PACA- Haute Corse, Nice)*
- **R** **COMARG** *(Guyane, Cayenne)*
- **S** **ALLO Maladies Rares** *(Martinique, Fort de France)*
- **T** **RE-MA-RARES** *(Réunion-Mayotte, St Pierre de la Réunion)*
- **U** **KARUKERARES** *(Guadeloupe, Pointe à Pitre)*

FOR MORE INFO

Rare Diseases Reference Centres (CRMR)

The CRMR provide medical and scientific expertise on one or more specific rare diseases. They ensure diagnosis, specialised care, clinical research, and training.

Referral of the patient for specialist opinion, diagnostic confirmation, and follow-up in complex clinical cases.

centre de référence maladies rares

Rare Diseases Competence Centres (CCMR)

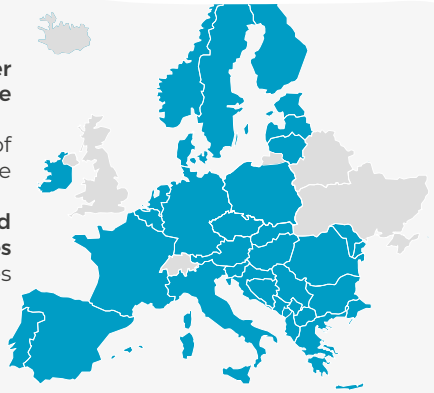
Local partners of CRMR, the CCMR provide routine follow-up and local coordination of the patient care pathway.

Local medical team providing specialised follow-up care.

centre de compétence maladies rares

European Reference Network (ERN)

ERNs are cross-border networks that bring together European hospital centres of expertise to tackle rare diseases.
ERNs enable specialists in Europe to discuss cases of patients, providing advice on the most appropriate diagnosis and the best treatment available.
There are currently 24 ERNs across the EU and Norway. ERNs include **1,606 specialised centres** located in 375 hospitals across 27 Member States and Norway.



European Reference Networks

DID YOU KNOW IT?

"Brèves de doc" : practical information sheets designed for general practitioners

- **A guide dedicated to patient referral,** for cases with suggestive symptoms or an unknown diagnosis.
- **A guide presenting and reiterating the importance of French National Diagnosis and Care Protocols (PNDS),** recommendations, and related guidance.

"Les clés du diagnostic" : tool to support the diagnosis of rare diseases.

Online resource dedicated to physicians to help identify a rare disease from a common clinical warning sign in the general population.
The tool also helps identify the appropriate expert centre to refer the patient or request a specialist opinion.

Information and resources

orphanet

International database of expert resources for rare diseases.

Comprehensive directory of more than 6,000 rare diseases.

Directory of reference/comptence centres

Classification used for diagnosis

List of orphan drugs

Registry of research projects

Clinical trials, registries, and biobanks

Directory of diagnostic laboratories

Collection of thematic reports

orpha.net/fr

MALADIES RARES INFO SERVICES

French information and support service dedicated to rare diseases. It also assists healthcare professionals in identifying an expert centre.

Free helpline number : 0 800 40 40 43

maladiesraresinfo.org

Alliance maladies rares
UNIS POUR VAINCRE

Collective spokesperson for patients and rare disease associations in France.
The federation informs and supports patients and their families.

alliance-maladies-rares.org

To facilitate the care pathway of patients living with rare diseases, the Collège de la Médecine Générale and the French Rare Disease Networks (Filières de Santé Maladies Rares) provide **practical tools developed by general practitioners for general practitioners.**

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in digital format

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