



29 November 2024

European Economic
and Social Committee

Leaving No One Behind : European Commitment to Tackling Rare Diseases

French Rare Disease National Plan –
Model and Impact

Jean-Philippe Plançon
Vice-President



RD National plans are supported by



MINISTÈRE
DE L'ENSEIGNEMENT
SUPÉRIEUR
ET DE LA RECHERCHE



BUDAPEST



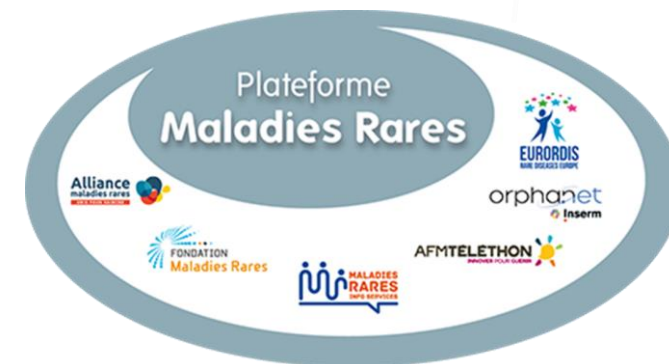
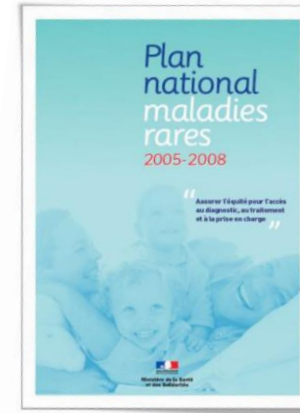
Nothing about us without us !

Strong engagement from patient associations

Major political involvement from the Parliament and the French Republic Presidents

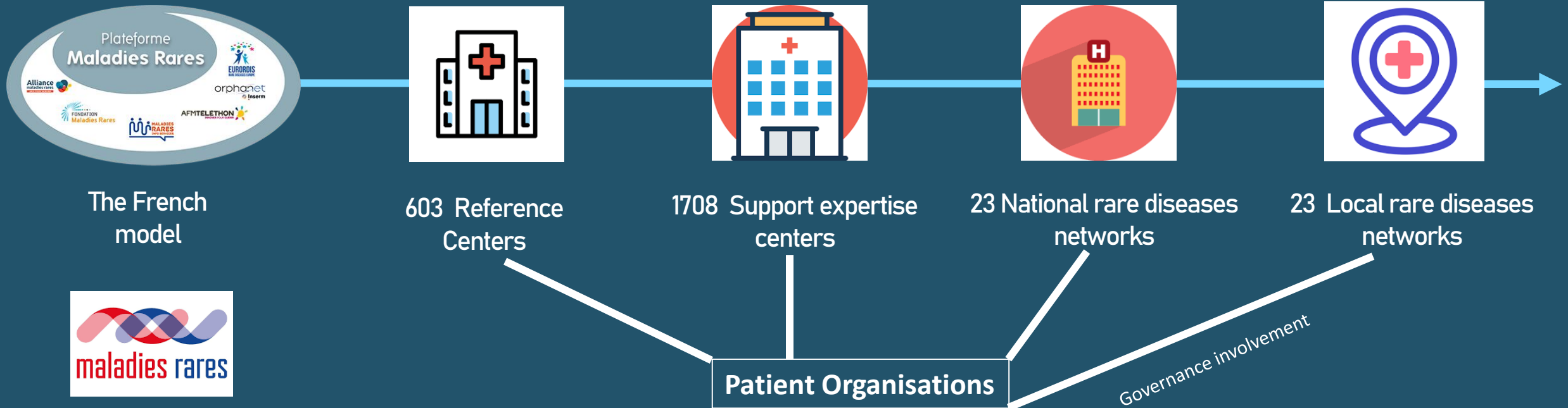
A global approach to the rare diseases cause, tackling all issues simultaneously and in a coordinated way, regardless of any institutional silos

Strong building of public policies with all stakeholders, in particular patient associations, through numerous consultations and working groups.





A rare diseases care ecosystem



Reference centers at the heart of patient care



- 70% of patients say they found it easy to contact their center.
- Over 60% waited an average of less than three months for an appointment.
- Only 6% waited over a year.
- 77% were satisfied with the quality of care offered, and 68% with the speed of delivery of test results and hospitalization reports. 71% of respondents were satisfied with the services offered by expert centers.



- Coordination between GP and expert center.
- Regarding the “child-adult” transition, 54% of associations said they were satisfied with the support offered between a pediatric service and an adult service.
- Finally, only 33% of centers offer psychological support, 20% the services of a social worker and 30% full information on transport costs, long-term conditions and other more technical aspects.

TO IMPROVE

Rare diseases health networks: For a change of scale



- 80% of patient organizations said they are "satisfied" with the Rare Diseases health networks.

This high level of overall satisfaction is due to a systematic collaboration that works, from the point of view of both the associations and the medical professional networks.



Major successes of the national reference networks includes : decisive support in creating guidelines, monitoring the re-labeling of expert centers, and help in designing and monitoring patient health education programs.



TO IMPROVE

Supporting human and social sciences programs, financial support for associations, legal and psychological support, telehealth and raising awareness of rare diseases among general practitioners.

Raising awareness of rare diseases among professionals appears to be a priority issue for all stakeholders.

Concrete example



**European
Reference
Network**

for rare or low prevalence
complex diseases



Network

Neuromuscular
Diseases (ERN EURO-NMD)



Education / Information

Ex. Dysimmune and inflammatory neuropathies

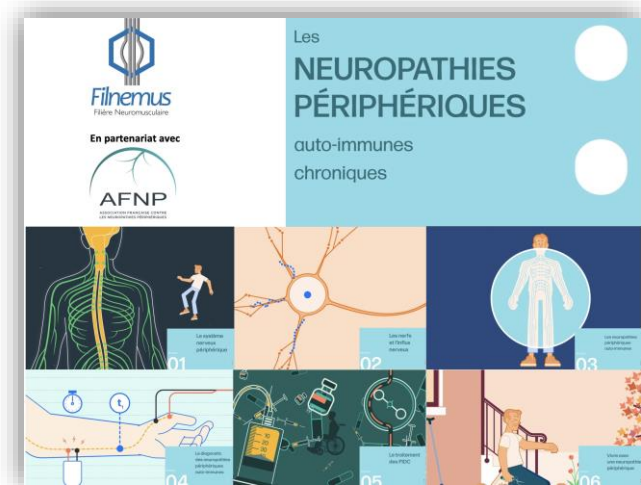
- .National call for projects, dedicated to patient organizations
- .Specific WG « Improve patient journey» / Treatment observatories
- .Patient therapeutic education program : to support patient autonomy

- . Supporting relevant projects for patients / Education; information...
- . In line with the missions of the national reference networks (improving care - research - education, training and information - Europe and international).



Interactive information sheets online

Videos



6 épisodes



11 episodes



European
Reference
Network

for rare or low prevalence
complex diseases

 **Network**
Neuromuscular
Diseases (ERN EURO-NMD)


**COLLABORATION ERNs – NRNs +++
Supported by POs involvement**


Education / Training

<https://ern-euro-nmd.eu>




Chronic dysimmune PERIPHERAL NEUROPATHIES

 Filnemus
Filière Neuromusculaire

 European Reference Network
for rare or low prevalence complex diseases

 AFNP
ASSOCIATION FRANÇAISE CONTRE LES NEUROPATHIES PÉRIPHÉRIQUES

 EPODIN

Multifocal motor neuropathy Patient Journey

STEP 1 Main symptoms and impact

- Asymmetric multifocal muscle weakness and atrophy
- Usually in the upper limbs, but it can also affect the lower limbs
- Onset can be from hands (buttons, glass, etc.)
- Difficulties dressing
- Choreoform
- No sensory disturbance
- Feeling
- Usually little or no pain (except postural pain)
- Symptoms generally worsen over time and all limbs may be affected

STEP 2 Diagnosis

The diagnosis is most of the time chaotic and lengthy. The diagnosis is most of the time chaotic and lengthy. It is mainly based on clinical history, physical examination, nerve conduction studies (electromyography) and blood test (in particular to look for CMV antibodies). Diagnosis sometimes can take years.

STEP 3 Treatment

The first line treatment for MMN consists of immunoglobulins (plasma-derived medicinal products), which can be administered intravenously (IVI) or subcutaneously (SC). The administration of immunoglobulins can be quite intense and, in some countries exclusively conducted within a hospital setting. Patients with MMN often require long-term treatment that may extend throughout their lifetime, emphasizing the chronic nature of the condition. Even if IVIG treatment is very effective and in spite of continuous treatment symptoms may worsen. Other treatments are under investigation and may prove to be effective. Maintaining regular, adapted physical activity, physiotherapy and occupational therapy, aim to preserve the patient's physical autonomy.

STEP 4 Patient needs

One significant challenge is the lack of expert centers specialized in MMN and the difficulties in referring patients to these highly specialized facilities for optimal follow-up. Patients may face obstacles in accessing treatment, which can result in lasting negative consequences for their health and well-being. The lack of a dedicated support team, including options for hospital or home care, can complicate the delivery of consistent and comprehensive care to MMN patients.

MMN is classified as a very rare disease, there is a crucial need for patient education to enhance understanding of the disease (pathophysiology, available treatments, potential outcomes, support...). Providing access to rehabilitation services and promoting patient autonomy are vital aspects of MMN care, helping individuals maintain their quality of life and independence. In the event of a decline in autonomy, patients may require support in adapting their homes and daily routines to accommodate their changing needs.

Updated European guidelines are important for healthcare professionals to stay informed about the latest best practices in MMN management. Training programs for general practitioners and non-expert specialists are essential to recognize MMN patients. Raising awareness about the existence of MMN is also important, as participation in research studies can contribute to the development of new treatments and therapies for MMN.

STEP 5 Supportive care to be developed

Implementing adapted physical activity programs can help individuals with MMN improve their physical health and mental well-being. Encouraging appropriate physical activity recommendations tailored to each patient's specific condition is crucial for maintaining their quality of life. Regular monitoring and support, addressing mental health and emotional well-being of MMN patients, effective pain management strategies, with a focus on preventing, compensating, and providing care can improve discomfort and enhance the daily lives of MMN patients dealing with MMN.

Follow-up

The organization of follow-up care for MMN can vary from one patient to another, often involving a combination of healthcare providers. Many individuals with MMN can receive periodic follow-up from their general practitioner or primary care physician, who helps managing their routine healthcare needs. In addition to general follow-up, patients with MMN often benefit from specialist care provided by a neurologist who specializes in neuromuscular disorders, ensuring that they receive specialized treatment and monitoring for their condition.

6 Videos
6 Episodes

Patient Journeys program (2023-2024)

- . Multifocal Motor Neuropathies
- . Myasthenia gravis
- . Myofibrillar Myopathy

NEUROPATHIES REHABILITATION SUMMER SCHOOL

18TH - 21ST SEPTEMBER 2023
HOTEL EXCEL ROMA MONTMARIO

 European Reference Network
for rare or low prevalence complex diseases

 Network
Neuromuscular Diseases (ERN EURO-NMD)

06 congress

SUMMER SCHOOL

12th - 14th September 2024

PATIENTS JOURNEY

PATIENTS PERSPECTIVE

 EPODIN

 EUROPEAN FEDERATION

 EAMDA

 Patient Advisory Board

 European Reference Network
for rare or low prevalence complex diseases

 Network
Neuromuscular Diseases (ERN EURO-NMD)

 Filnemus

MARSEILLE
LE VIEUX PORT

"Mais alors,
dit Alice, si
le monde n'a
absolument
aucun sens
qui nous
empêche d'en
inventer un ?"



MERCI

« But then says Alice, if the world
makes absolutely no sense, who's
stopping us from inventing one? »

Quotation attributed to
Lewis Carroll

THANK YOU