7th ERN EURO-NMD

ANNUAL MEETING

PAB updates

21st – 23rd February 2024

François Lamy

PAB chair

European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

Funded by the European Union

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Agenda

1. Achievements
2. Ambition for 2024
3. Patients Journey

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Achievements

- EURO-NMD ERN first cycle evaluation
- Registry Hub (ad-hoc working group)
- ENMC workshop on bone health (muscle group)
- PROMs on pain and fatigue (Work Package)
- Patients Journey (first wave)

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Ambition for 2024

- Pursue ongoing activities
  - Registry Hub project
  - PROMs on pain and fatigue
  - …
- Build more Patients Journey (second wave)
- Communicate outside the network (POs)

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Ambition for 2024

Build educational material (webinars) for patients

- Registries
- Disability technical support (robotic arm, wheelchair positionning, wheelchair standing)
- Innovative drugs access
- Care (emergency, care quality, child to adulthood transition, virtual consultation)
- Very rare diseases specific information
- European Regulatory affairs

To be prioritized /ammended

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Patients Journey - what is a patient journey?

A patient journey is the chain of events people experience when they face a health issue.

Patients with rare and complex conditions often embark on a life changing odyssey including many aspects medical, social, psychological … that are incompletely known.

The aim of the Patients' Journey Program is to **improve the knowledge** of neuromuscular diseases by developing a **description of the diseases through patients' lenses.**

With this material our ambition is to lay the foundation of improved and standardized care for neuromuscular diseases in Europe.

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Patients Journey - method

- **Conception**
- Perform a mapping exercise of the needs of the disease they represent across the different phases of the disease
- Perform a literature review of existing guidelines for the disease(s) of choice

- **Construction**
- Divide the journey in different stages
- Build a graphic representation of the patients' journey
- Revision of the final Journey by patients and experts

- **Diffusion through Euro-NMD and patient organizations**
- Newsletter, webinar, conference, poster, …

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Patients Journey - organization

- Disease specific working group
  - 1 group leader
  - 3 to 4 patient representatives
  - 1 clinical contact point
  - 3 to 4 clinicians/experts
- Independent working groups
- Monthly workshops
- Coordination insured by a “support team” (Tamara / Elizabeth)
- Regular reports to the PAB
  - Synchronize with other groups work
  - Get external comments / advice

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Patients Journey
achievements

- Multifocal Motor Neuropathy
- Myathenia Gravis
- Myofibrillar Myopathy

Myofibrillar myopathy
Patient Journey

Patient Advisory Board

European Reference Network
for rare or low prevalence complex diseases
Network Neuromuscular Diseases (ERN EURO-NMD)

MEDICAL **SOCIAL**

STEP 1 Pre-diagnosis

Age of onset may vary considerably and may vary even within one family.
Red flags that may point towards MFM: when patients experience a lack of strength in the hands or feet, frequent falls or tripping, difficulties in running, climbing stairs or walking long distances, difficulties in cutting food or handling tools, a physician should be consulted.
Some cases present with heart involvement and/or respiratory involvement.

- People may see you as clumsy or lazy.
- Feeling / knowing that there is something wrong, not knowing what, makes you feel uncertain, looking for explanations.

STEP 2 Diagnosis

Myofibrillar myopathy is a group of diseases causing muscle weakness. Mutations can be found in different proteins. Symptoms may vary depending on the affected protein. The results of the following tests can point in the direction of Myofibrillar myopathy: Blood samples, including determination of CK (creatine phosphokinase) levels, EMG (electromyogram), muscle magnetic resonance (MRI), muscle biopsy and genetic testing As it is a rare disease, diagnosis is often set in a referral/expert centre (university or academic centre) by a neurologist or genetic counsellor specialized in neuro-muscular disorders.

- **It may be a relief to finally get the correct diagnosis.**
- For affected parents: It can be difficult to see that you passed the disease on to your child
- Your partner or other significant persons may need time to accept the disease as well.
- Finding information about this rare disease is often difficult.
- Your social environment might have difficulties to estimate the severity of the disease, because it is unknown.

STEP 3 First years after diagnosis

It is recommended to have regular consultations with medical specialists. At least by the neurologist or cardiologist, preferably by both. Follow-up may address cardiac problems, muscle skeletal evaluation, rehabilitation, respiratory function and swallowing problems.
Doing exercise is important. Stretching is good. Moving the muscles that are still in good condition. Keeping breathing muscles in condition is useful to remain having an effective cough. For children exercise may be going for instance to the pool and splash around. Do not compare to peers.
The diet should be varied.Supplementation of vitamins could be useful.

- This may be a good time to reconsider your professional ambitions to find a job that is possibly less strenuous and/or can be continued when the body weakens. **Having or keeping a job can contribute to the feeling of having a meaningful live.** In the early stage, the disease often is "invisible" for others.
- For others it may be difficult to understand what it does to you.

STEP 4 When symptoms get more serious

A rehabilitation physician or neurologist may help to prevent overburdening or under use of muscles.An evaluation of the respiratory function should be done regularly from day one. Be aware of symptoms such as daytime sleepiness, nightmares, morning headaches, waking up very tired after having slept all night.
A regular cardiac evaluation is recommended. Physical therapy may be helpful, especially to help prevent overburdening of healthy muscles.
Pain management may be necessary as a result of overburdening (healthy) muscles or contractures
It can be useful / necessary to have an easy accessible instruction (e.g. in your phone or written on a card) for emergency workers in case of an accident or other emergency: what to do or not to do (e.g. whether respiratory support is provided or anaesthetics that may be avoided)

- **Aids, such as a wheelchair, crutches, walking stick or mobility scooter may help to manage fatigue and keep doing things that make you happy (and thus provide energy).** The disease will become more visible. Think ahead what you will say when people (sometimes complete strangers) ask what is wrong with you.
- Car adaptations may be necessary to continue driving safely.
- **Fatigue may affect your ability to keep positive. Learning to manage your energy levels is important.**
- Those with a paid job may need to adapt their working hours or adjust their tasks to less strenuous activities.

STEP 5 When severe disability is the case

Not all mutations result in very severe disability, which is rare and usually occurring in late-disease stages.
See a physician regularly to prevent problems such as pressure sores, contractures, loosing energy due to insufficient body support/inadequate seating. Correcting or preventing contractures can be done by stretching and wearing (night)splints. Aids may be needed to help: hoist, hospital bed, arm support to keep moving your arms, power wheelchair, high toilet, shower commode. Home care may help with dressing, showering and other activities of daily living. Prevent/treat pressure sores and contractures

- **Don't get tired of visiting all different medical specialists. Healthcare is typically not yet organized around the patient, but being followed up medically is important.**
- **It is important to keep socially active and to somehow have a meaningful life. Make good use of your better days.**
- **Evaluate ways of visiting the toilet, such as a (suprapubic) catheter or belly button** stoma. **Make sure you manage to participate in society, by obtaining the right assistive devices, dealing with inaccessibility issues in society to prevent loneliness. This can be strenuous.**

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Patients Journey – lessons learned

- The final version must be a maximum of 2 pages (a larger document can be accessed through a QR code)
- It is better to use full sentences rather than bullet points to make it as clear as possible.
- Allow more space on the schedule for the design
- The patient journey must identify the medical and clinical aspects of the disease, as well as its impact on the patient's real life, and must direct patients to the best experts.

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Patients Journey – next steps

- Diffusion through Euro-NMD and patient organizations
  - Conference
  - Poster
  - Newsletter

- More patient journeys in preparation for 2024
  - Myositis
  - Amyotrophic Lateral Sclerosis (ALS)

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