

## **Report from the first workshop of the ERN EURO-NMD Multidisciplinary management and care specialty group**

Patients with neuromuscular disorders (NMDs) commonly grapple with the diverse and manifold consequences of muscle weakness, and many disorders also involve other organ systems. As a result, a multidisciplinary approach is considered the standard of care for patients, both pediatric and adult, with NMDs.

The Norwegian initiative to develop a multidisciplinary management and care specialty group within the ERN EURO-NMD was supported by the member representatives at the board meeting in Brussels in July 2022. The speciality group was founded at the annual ERN EURO-NMD meeting in Porto in February 2023.

This report provides a summary of the main topics presented and discussed at the first workshop of the multidisciplinary management and care group, arranged at Frambu Resource Centre for Rare Disorders, outside of Oslo in Norway on 4<sup>th</sup> and 5<sup>th</sup> of May 2023. The workshop gathered speakers with extensive experience in NMDs and specialty group members with the aim of sharing knowledge and developing activities within the multidisciplinary group. The workshop was a hybrid event with the possibility to participate at the center or remotely for those not able to travel. In total, 36 participants from eight different European countries participated.

The following main topics was addressed at the workshop.

- Models and status of multidisciplinary management and care
- Needs in palliative care
- Pain, fatigue and rehabilitation interventions
- Use of biomarkers as outcome measures in clinical practice and research

More information about topics and presentations are found in the workshop program.

### **Models and status of multidisciplinary management and care**

It is well documented in multiple diagnosis-specific international consensus care guidelines, that a multidisciplinary approach is essential in treatment and care of NMDs. However, there seems to be a discrepancy between the structures and systems proposed in the literature, and actual implementation in healthcare-systems across Europe.

Advances in diagnostics and treatment have increased the complexity of patient monitoring and care. In this new treatment era patients are surviving longer and, in most cases showing improvement in motor function. New patient phenotypes are emerging and a more proactive approach to multidisciplinary care is even more relevant.

Standardization is a prerequisite for quality and consistency of health care services. To ensure this there is an urgent need of more research to develop reliable procedures and evidence-based measures for all relevant care aspects. In addition, the heterogeneity in symptom manifestation and prognosis implies that the multidisciplinary approach needs to be adapted to the individual patient needs, and no truly effective multidisciplinary care can be developed without patient involvement.

Apart from description of important care areas, the evidence of the effect of multidisciplinary teams is scarce. Some have proposed an ideal scenario where healthcare services within hospitals are

structured as an interdisciplinary core team, equipped with the capacity to access supplementary resources as required. There is currently an ongoing study investigating status of the multidisciplinary care offered to patients with NMDs in Europe. The study with some of its intermediate results were presented at the workshop.

#### *Discussion and future perspective*

Sharing of experiences and discussions between workshop participants, revealed variability between centers and countries in professionals involved, organization of- and health care services offered to patients with NMDs. However, the multidisciplinary approach to management and care in general seems more developed in the pediatric compared to the adult field. In most cases, the main barriers are lack of time and resources, and obstacles in organization and coordination of health care services.

Dissemination of the importance of implementing multidisciplinary management and care for NMDs will be a priority of the specialty group. In addition, enhancement of visibility and recruitment of more members into the specialty group were deemed important for developing networks within the multidisciplinary management and care group. A description of the specialty group was agreed developed and published at the ERN EURO-NMD website. As a fundament for development of profession specific networks within the specialty group, appointment of area representatives within the specialty group was discussed and agreed.

#### **Needs in palliative care**

Palliative care is defined as the prevention and relief of suffering of pediatric and adult patients and their families facing the problems associated with life-limiting illness. It is relevant for a range of chronic progressive life-limiting NMDs and offers a support system to help patients and families to cope and live as actively as possible until death. Palliative care is a holistic approach addressing clinical, psychological, social, and spiritual needs and include advanced care planning where the individuals' values, beliefs, wishes about treatment and goals of care should be assessed. This structured model of care planning enables the establishment of a mutual understanding of priorities, hopes, fears, goals for future treatment, end-of-life care and death. It is recommended to be a systematic and continuous process.

Pediatric palliative care differs in certain aspects from palliative care for adults, and should be offered to the child and family members from time of diagnosis of a life-limiting condition and be continued through to end-of-life. It also includes bereavement care of family members after the child's death. The care services need to be adjusted to the child's communication skills, physical, cognitive, and social development and should be offered concurrently with, and not instead of, disease directed treatment. Introducing pediatric palliative care early helps avoid disruptive transition to new care team at the worst possible time. It also contributes to improved symptom management throughout the disease trajectory, improved quality of life for the ill child and family members, and improved psychosocial care at end-of-life.

Palliative care is underutilized in most relevant NMDs, except in amyotrophic lateral sclerosis. Barriers are described to be unawareness of the possible benefits, misperception of palliative care being synonymous to hospice or end-of life care, and lack of collaboration between neurologists and palliative care physicians. Challenges also include the broad spectrum of symptoms and progression, and unpredictable disease trajectories in NMDs. The literature on palliative care for NMD is limited, and more research is needed on how to best integrate palliative care in care pathways, including the transition from pediatric to adult care.

### *Discussion and future perspective*

Palliative care is still a young field with large variations between countries. Limited access to multidisciplinary teams with education and skills required for delivery of palliative care was described as a shortcoming in many countries.

Few of the participants had experience from palliative care and few were in regular contact with palliative care teams. Several participants expressed an insecurity on when to introduce and how to communicate about palliative care to patients and their next of kin, as the term palliative care still easily refers to end-of life. This makes the phrase palliative care difficult to use in clinical practice. Advanced care planning or comprehensive care was alternatives used at some centers.

Early introduction to palliative care is recommended but it is difficult to explain to some parents that new treatments and palliation goes together. Providing palliative care isn't about giving up hope, rather it focuses on enhancing quality of life. In some diagnoses, e.g. Myotonic dystrophy type I it is important to have conversations about care planning and document future wishes before the patients get cognitive affected.

### **Pain, fatigue and rehabilitation interventions**

It is well known that many patients with NMDs experience chronic pain and fatigue, and these aspects can have severe consequences for quality of life and participation. Pain can be caused by overuse due to weakness, static loading, muscle tension, compensation mechanisms and neuropathic pain, and are influenced by psychosocial and physical reactions to pain. Chronic fatigue is characterized by early exhaustion that isn't directly related to effort, and it is not typically ameliorated by rest. Many instruments for measures of pain and fatigue are developed, but there is currently no standard validated patient reported outcome measures used in clinical practice or research for NMDs. This limits comparisons between centers and studies.

Pain and fatigue in NMDs are multidimensional problems and therefore may require multicomponent interventions adjusted to the individual patient's personality and needs. Exercise interventions in NMDs should begin with low intensity and progress gradually. To prevent chronic pain and fatigue it is important that the patient listen to the signals from their body, and uses aids when required to avoid overuse. To cope and understand the signs and symptoms of their NMD, patients need education. Controlled intervention studies indicate that the combination of cognitive behavioral therapy and individually tailored exercise can alleviate chronic fatigue and enhance muscle mass in NMD patients.

### *Discussions and future perspectives*

Discussion revealed that much work remains before effective multicomponent rehabilitation interventions are implemented in care services and offered as part of routine practice to patients with NMD in all European countries. As a basis for this multidisciplinary rehabilitation clinics with knowledge and experience in NMDs is required, and are established in some but not all countries.

### **Use of biomarkers as outcome measures in clinical practice and research**

Muscle magnetic resonance imaging (MRI) and muscle ultrasound have become useful tools in the diagnosis and follow-up of patients with NMDs. Both techniques measures levels of fat infiltration and muscle volume, and may be used for description of muscle pathology and muscle involvement, as well as disease progression, in various NMDs. MRI may include the whole body whereas ultrasound is typically applied to a specific muscle or a set of muscles. MRI have been used to detect

and quantify changes in muscles following treatment interventions. In the literature, significant correlation between MRI measures and 6-minute walk test results have been documented.

Technology for quantifying gait in patients with NMDs, e.g. sensor and/or video-based gait analysis measures, are available on the market. Such technologies may be used to describe typical movement patterns in different disorders, and measure effect of interventions. Use of rehabilitation technology devices is an emerging field, but more research is needed to investigate the effect in patients with NMDs.

#### *Discussion and future perspective*

Fewer participants at the workshop used robotics or gait analysis as an outcome measure to evaluate patients in clinical practice and research compared to MRI and ultrasound.

Functional test instruments are widely used, and a large variety of instruments are regularly used in clinical practice and research. Likewise, variable instruments for assessment of pain and fatigue are used. It was agreed that the specialty group should work towards harmonization and standardization of outcome measures across ERN EURO-NMD centers. It was suggested to arrange a new separate workshop to discuss and address this further.