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GUIDELINES

Evidence based position paper on Physical and Rehabilitation Medicine practice for people with muscular dystrophies

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ABSTRACT

Muscular dystrophies present a group of inherited degenerative disorder that are characterized by progressive muscular weakness. This evidence-based position paper represents the official position of the European Union through the UEMS PRM Section. The aim of the paper is to evaluate the role of the physical and rehabilitation medicine (PRM) physician and PRM practice for people with muscular dystrophies. A systematic review of the literature and a consensus procedure by means of a Delphi process have been performed involving the delegates of all European countries represented in the UEMS PRM Section. The systematic literature review is reported together with thirty-three recommendations resulting from the Delphi procedure. The role of the PRM physician is to assess the functional status of persons with muscular dystrophy and to plan, monitor and lead PRM program in an interdisciplinary setting within a multiprofessional team.

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KEY WORDS: Muscular dystrophies; Rehabilitation; Consensus.

Neuromuscular diseases (NMD) form a heterogenous group of disorders affecting the peripheral nervous system and/or muscles, and differs in disease onset its course time and clinical expression.[1] They are characterized by numerous signs and symptoms including muscle weakness, joint and skeletal deformities, respiratory dysfunctions as well as dynamic impairments (*i.e.* exercise intolerance, etc.).[2] Despite the heterogenicity in expression, most of the patients with NMD have similar requirements for diagnostic access and treatment procedures.[3] Furthermore, advances in genetic testing for individuals with NMD, enable not only more accurate diagnostics and treatment of such pathology, but introduce the possibility of genetic counseling.[4] The burden of NMDs are reflected in the fact that some of them (Becker muscular dystrophy, facioscapulohumeral dystrophy, myotonic dystrophy and Charcot-Marie-Tooth disease) increased over time, while others like amyotrophic lateral sclerosis, Duchenne muscular dystrophy had stable prevalence.[5]

Muscular dystrophies

Muscular dystrophies present a group of inherited degenerative disorders that are characterized by progressive muscular weakness.[6, 7] They are caused by mutations on numerous genes, with dominant, recessive, X-linked expression or can be as de novo mutation.[7] In the diagnosis, special attention should be referred to the pattern of weak-

ness distribution, age at the onset of symptoms, family history and specific characteristics that can distinguish certain types of muscular dystrophies.[6] The Duchenne Muscular Dystrophy is reported to be among most common types of muscular dystrophies with incidence of 1 in 3500 males.[6]

It should be stressed that muscular dystrophies lead to the loss of muscle strength and reduction in individuals' physical activity, causing various degrees of disability that affect patients' quality of life.[8] Considering the care of these patients, particular attention needs to be addressed to cardiac and pulmonary systems, especially in certain types of muscular dystrophies.[9, 10] The necessity for participation of family in the care of individuals with muscular dystrophy is important and challenging, since there is a need for a long-term involvement. Previous reports stated that family involvement has positive effects on individuals with long-term illness with favorable adaptation to the disease, positive impact on one's participation in treatment as well as improved clinical response.[11] Therefore, the importance of standards of care implementation refers to the fact that optimal care can increase survival and anticipatory guidance as well as promote proactive management.[12]

Physical and rehabilitation medicine (PRM) physicians are trained in postgraduate specialist studies to treat and implement rehabilitation programs for patients with neuromuscular diseases. However, it should be noticed that uniformed standards across the Europe haven't been established so far. The emerging need for optimal, up-to-date and evidence-based treatment of patients with muscular dystrophy at all levels of medical care is the cornerstone of successful management of the entire plethora of challenges that both patients and their families along with society are involved in. Therefore, the main goals of PRM physicians along with the multiprofessional team participating in the treatment of these patients should be to preserve and/or improve functioning, enabling the reintegration into society and improve the quality of life of one bearing in mind the needs of family and/or caregivers.

The aim of this paper is to evaluate the role of the physical and rehabilitation medicine (PRM) physician and PRM practice for people with muscular dystrophies.

Methodology

This evidence-based position paper (EBPP) has been developed in accordance with the methodology proposed by the Professional Practice Committee of the Section of Physical and Rehabilitation Medicine of the Union of European Medical Specialists (UEMS-PRM Section).[13] It consists of two parts: "systematic review of the literature" and "consensus with Delphi procedure among UEMS PRM Section delegates."

Literature search

The systematic review of the literature included articles relevant to the PRM profession. The search relevant to muscular dystrophies was conducted using the search terms/strings and included articles extracted from the PubMed/MEDLINE. Additionally, we included grey literature for other guidelines that are not published in Journals (online sources), as well as representative articles/reports/position papers in the searched field by major relevant international bodies (*e.g.* the WHO, the UEMS PRM Section and others). The time period of searched articles was between 2006-2019 years. For the ICF classification, the WHO reference from 2001 year was cited in recommendations. Our priorities in the systematic literature search were systematic reviews, meta-analyses, randomized controlled trials, evidence-based papers and guidelines, however other relevant papers were included as well.

Search strings in PubMed/MEDLINE: muscular dystrophy[All Fields] AND rehabilitation[All Fields] (String 1) #1033; muscular dystrophy [All Fields] AND physical therapy[All Fields] (String 2) #524; muscular dystrophy [All Fields] AND exercise[All Fields] (String 3) #833; muscular dystrophy [All Fields] AND electrodiagnostics [All Fields] (String 4) #34; muscular dystrophy [All Fields] AND telemedicine[All Fields] (String 5) #6; muscular dystrophy [All Fields] AND pain[All Fields] (String 6) #369; muscular dystrophy [All Fields] AND epidemiology[All Fields] AND intervention[All Fields] (String 7) #505.

We also included additional articles in PubMed/MEDLINE from White Book on Physical and Rehabilitation Medicine (PRM) in Europe (#3), from PubMed/MEDLINE of interest (#1), ICF core set (#1) and grey literature (#3) (Supplementary Digital Material 1: Supplementary Text File 1).

TABLE I.—*Results of the consensus procedure.*

Round	Number of recommendations	Accept	Accept with changes	Reject
1	32	34.4%	65.6%	0%
2	33*	27.3%	72.7%	0%
3	33	100%		0%
4	33	100%		0%
5	33	100%		0%

*1 added after 1st Round.

TABLE II.—*Overall view of the recommendations.*

Content	Number of recommendations	Strength of recommendations				Strength of evidence			
	N.	A	B	C	D	I	II	III	IV
Overall general recommendation	1	100%	0	0	0	0	0	100%	0
Recommendations on PRM physician's role in Medical diagnosis according to ICD	3	0	100%	0	0	0	0	33.33%	66.67%
Recommendations on PRM physician's role in PRM diagnosis and assessment according to ICF	4	75%	25%	0	0	0	0	25%	75%
Recommendations on PRM management and process	23	69.57%	30.43%	0	0	0	0	26.09%	73.91%
Recommendations on future research on PRM professional practice Project definition	2	50%	50%	0	0	50%	0	0	50%
Total	33	63.64%	36.36%	0	0	3.03%		27.27%	69.70%

The recommendations draft and consensus followed the five steps Delphi procedure (Table I) that was proposed by the Methodology paper.[13] Strength of Recommendations (SoR) grading, and Strength of Evidence (SoE) grading was described as well in the Methodology paper.[13] Overall view of the recommendations is presented in Table II.

Results

Systematic review

The electronic search of the literature identified 3309 papers and 3 references from grey literature, from which 949 abstracts and 3 grey literature articles were selected and finally 82 articles of them as well as 3 from grey literature were considered to produce this paper. Flow chart of the selection process is presented in Figure 1.

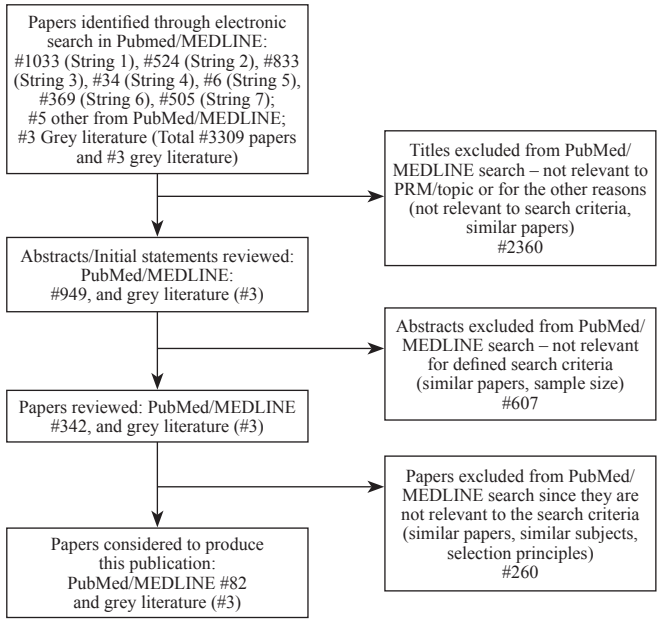

Figure 1.—Flow chart of the study.

Recommendations were prepared according to the chapters proposed in the Methodology paper (h) as defined by the Professional Practice Committee of the UEMS-PRM Section:

A. Overall general recommendation;

B. Recommendations on PRM physicians' role in Medical Diagnosis according to ICD;

C. Recommendations on PRM physicians' role in PRM diagnosis and assessment according to ICF;

D. Recommendations on PRM management and process;

E. Recommendations on future research about best PRM professional practice in persons with muscular dystrophies.

Recommendations

A. Overall general recommendation

1. The professional role of the PRM physician as a part of the multiprofessional and/or interdisciplinary team for patients with muscular dystrophy, is to propose physical and rehabilitation medicine treatment with regard to the age, existing comorbidities, recognize present functional capacity and optimally improve it, prevent and reduce complications` severity degree, as well as to enable optimal integration into community.[14-16] The PRM physicians should apply evidence-based interventions and guidelines concerning rehabilitation treatment for patients with muscular dystrophy [SoR: A; SoE: III].

B. Recommendations on the PRM physician's role in Medical diagnosis according to ICD

2. It is recommended that PRM physicians obtain specific expertise regarding muscular dystrophy diagnosis along with optimal and most appropriate treatment approaches.

They should collaborate with neurologists, pediatricians and/or other medical specialists relevant to the specific condition. PRM physician is the specialist competent to assess the functional consequences of damage to muscles or other associated organs. Regular monitoring of secondary conditions, comorbidities and complications is advised since they can lead to the additional progressive impairments [SoR: B; SoE: IV].

3. It is recommended that PRM physicians consider individualized functioning of the patients with muscular dystrophy whatever the age. Some age groups have specific conditions (older and children) and therefore (less true with advances in genetics) modified treatment responses.[17, 18] The proper examinations bearing in mind the age of patients will enable optimal assessment and prevent high risk patient groups from potentially negative outcomes (*e.g.* type of muscular dystrophies, specific complications, rapid functional decline, and reduced survival) [SoR: B; SoE: IV].

4. It is recommended that PRM physicians have knowledge of and are able to interpret electrodiagnostic tests (electromyoneurography-EMNG) and establish the severity degree of myopathic pathology.[19-21] PRM physicians should gain expertise for this diagnostic method [SoR: B; SoE: III].

C. Recommendations on PRM physicians' role in PRM diagnosis and assessment according to ICF

5. It is recommended that PRM physicians perform a complete assessment and define medical component of individual rehabilitation plan for patients with muscular dystrophy with particular attention to the identification of present symptoms and clinical signs as well as impairments including: muscular and respiratory dysfunction (mobility and upper limb suppliance), cardiac dysfunction, pain, contractures, postural control/balance function, spine deformities, presence of fatigue and dysphagia, diet problems[15, 22-25] [SoR: A; SoE: IV].

6. It is recommended that PRM physicians use the International Classification of Functioning, Disability and Health (ICF) framework for collecting information on functioning, including activity and participation domains emphasizing impairments in body structure and function in patients with muscular dystrophy, within their environments and taking into account personal factors of these patients.[26-28] The assessment of functioning based on the ICF should include: problem identification, goal setting, intervention and re-assessment. This assessment should be 3 to 6 months for children, 1 to 2 years for adults (except in case of pulmonary or cardiac dysfunction not stabilized), with even more frequent assessment in cases of concern, status change, etc. Such tool should be recommended for proposing, planning, and implementing the optimal rehabilitation treatment[29] [SoR: A; SoE: III].

7. It is recommended that PRM physicians use ICF Core Sets for neuromuscular diseases for patients with muscular dystrophy to target PRM interventions[30] [SoR: A; SoE: IV].

8. It is recommended that PRM physicians pay particular attention to lifestyles and participation (*e.g.* environmental domains) of patients with muscular dystrophy in order to identify barriers and facilitators that could influence the involvement in numerous life situations[15] [SoR: B; SoE: IV].

D. Recommendations on PRM management and process

Inclusion criteria (**e.g.** when and why to prescribe PRM interventions)

9. It is recommended that PRM physicians prescribe an optimal and adequate rehabilitation program whenever rehabilitation is needed in close collaboration with a neurologist, internal medicine specialist, pneumologist, cardiologist, pediatrician, orthopedic surgeon and/or other physicians, within the multiprofessional and interdisciplinary team in every stage of disease[16, 25, 31, 32] [SoR: A; SoE: III].

10. It is recommended that PRM physicians timely prescribe PRM interventions where there are indications throughout every step of care for persons with muscular dystrophy, thus reducing impairments in body structures and functions, maintaining and/or facilitating individuals' activities and participation in their environments[31, 33, 34] [SoR: B; SoE: III].

Project definition (definition of the overall aims and strategy of PRM interventions)

11. It is recommended that PRM physicians along with the multiprofessional team define overall aims and strategy for rehabilitation interventions. The PRM physician`s role is to: manage and coordinate the rehabilitation goal setting process, plan, monitor and evaluate proposed PRM programs with special consideration of the individual's needs. Additionally, PRM physicians should educate and advise patients as well as family members and/or caregivers to a self-management[35, 36] [SoR: A; SoE: IV].

12. It is recommended that PRM interventions are implemented depending on the phase of the health condition: in acute care settings including intensive care units, in postacute care settings, in specialized rehabilitation cen-

ters, at home or in the community, with the main purpose of reducing activity limitations and participation restrictions in specific environments as well as preventing further functional decline.[37, 38] Moreover, the introduction of telemedicine as a supplementary tool in multidisciplinary care of patients with muscular dystrophy might be of certain advantage[39, 40] [SoR: A; SoE: IV].

13. It is recommended that PRM physicians participate in every step of the transition of patients with muscular dystrophy to adulthood[41, 42] [SoR: A; SoE: III].

14. It is recommended that PRM physicians participate in discussions, and propose recommendations on policies, interventions and programs, as well as good practice statements to health-policy makers, stakeholders, academic institutions and international organizations in order to meet the needs of persons with muscular dystrophy[41-44] [SoR: A; SoE: III].

Teamwork (professionals involved and specific modalities of team work)

15. It is recommended that rehabilitation for patients with muscular dystrophy is performed by a multiprofessional rehabilitation team working in multi-, inter- or trans-disciplinary way in all relevant rehabilitation settings[34, 45] [SoR: A; SoE: IV].

16. The multidisciplinary team should include: expert PRM physicians, neurologists, orthopedic surgeons, psychiatrists, internal medicine specialists, cardiologists, pneumologists, pediatricians and other medical specialists together with other rehabilitation professionals: physiotherapists, exercise physiologists, occupational therapists, speech and language therapist, nurses, social workers, prosthetists, orthotists, rehabilitation engineers, vocational counsellors, behavioral therapists, and other health professionals: dieticians, nutritionists, and social care services or community-based workers, with mandatory inclusion of family members or caregivers[39, 46-48] [SoR: A; SoE: III].

17. The PRM physician should organize and conduct regular team meetings in order to propose, discuss and update the individual rehabilitation plan [SoR: B; SoE: IV].

PRM interventions

18. It is recommended that PRM physicians adopt adequate and effective communication skills with patients and family members or caregivers and adequately respond to patients` demands regarding concerns and important information. Furthermore, PRM physicians should take active involvement in continuous medical education in order to develop adequate theoretical knowledge, evidence based clinical skills and therapeutic skills (pharmacological and rehabilitation) and adopt guidelines for best optimal planning and performing PRM treatment through all stages of rehabilitation in patients with muscular dystrophy[49, 50] [SoR: A; SoE: IV].

19. It is recommended that PRM physicians participate in the multidisciplinary treatment in every stage of muscular dystrophy, with individually designed rehabilitation programs that are oriented to the individual's needs, response and tolerance to treatment, prevention of complications and functioning improvement[30, 51, 52] [SoR: A; SoE: IV].

20. It is recommended that PRM physicians apply treatment modules (physical medicine and rehabilitation medicine strategies, pharmacological and adjunctive treatments) in patients with muscular dystrophy taking into consideration the presence and degree of expression of clinical signs and symptoms[53-55] [SoR: A; SoE: IV].

21. It is recommended that rehabilitation assessment by the PRM physician in the multidisciplinary team of patients with muscular dystrophy is performed when needed by the patient, keeping in mind that follow-up in children and adults might be different in frequency due to the specifics in these age populations. Additionally, assessment of pulmonary function is recommended for ambulatory or non-ambulatory patients at least yearly or as much as necessary (for those age of 6 years and above) especially if vital capacity is less than 40% or the ability to cough is impaired or the patient complains of signs of hypercapnia or if the patient is already ventilated. It is recommended to monitor for scoliosis once a year in ambulatory patients and twice a year in non-ambulatory patients during growth and less frequent in adult age[32, 33, 56] [SoR: A; SoE: IV].

22. It is recommended that PRM physicians deliver care standards oriented to preventive measures in the early presymptomatic stage of disease in patients with muscular dystrophy[56] [SoR: B; SoE: IV].

23. It is recommended that patients with facioscapulohumeral muscular dystrophy (FSHD) participate in aerobic exercise training and cognitive-behavioral therapy to ameliorate chronic fatigue and pain. Additionally, balance training should be considered as well[57, 58] [SoR: B; SoE: IV].

24. It is recommended for PRM physicians to consider that lip strengthening exercises are shown effective for improvement of lip strengthening in children with myotonic dystrophy type 1. Furthermore, lip strengthening exercises should be considered as a complement and not a replace-

ment for speech therapy and dysphagia treatment[59] [SoR: B; SoE: IV].

25. It is recommended that pain be addressed in multidisciplinary setting individually for patients with muscular dystrophy at all ages. The rehabilitation interventions should include physical therapy, orthotic interventions with postural control, assistive devices (*e.g.* wheelchair), psychological – behavioral therapy and pharmacological therapy. Additionally, specific attention should be given to back pain in patients with Duchenne muscular dystrophy due to the possible iatrogenic vertebral fractures, particularly if patients are on glucocorticoid treatment.[33, 36, 60] Other sites of fracture and falls should also be taken into consideration [SoR: A; SoE: IV].

26. It is recommended that PRM physicians use validated instruments for the evaluation of activity levels of patients with muscular dystrophy[61-64] [SoR: A; SoE: IV].

Outcome criteria

27. It is recommended that PRM physicians decide on the outcome criteria during the assessment and goal-setting processes using physical examination and suitable disability scales as minimal set of assessments within the ICF framework.[30, 31, 65-69]

1. ICF based measures for neuromuscular disease related disabilities:
- ICF core set neuromuscular disease NMD;
- Motor Function Measure (MFM) and Revised Upper Limb Module (RULM).

2. Generic health measures:
- Gross motor function scale;
- SF-36;
- World Health Organization WHOQOL-BREF quality of life assessment;
- Groningen Activity Restriction Scale (GARS);
- Impact on Participation and Autonomy Questionnaire (IPAQ);
- WHO Disability Assessment Schedule (WHODAS 2.0);

3. Specific patient reported questionnaire (Rasch validation):
- QoL-gNMD (quality of life);
- ACTIVLIM (activity) [SoR: B; SoE: IV]

28. It is recommended that PRM physicians follow updated consensus statements, guidelines and protocols for optimal rehabilitation and management of patients with muscular dystrophy in order to make evidence-based decision for best possible outcome criteria[26, 45, 70-75] [SoR: A; SoE: III].

Length/duration/intensity of treatment (overall practical PRM approach)

29. It is recommended that the PRM physician plan the length, duration and intensity of the rehabilitation treatment within multidisciplinary care, taking into consideration specific needs and desired goals, along with the physical and psychological conditions of the patient with muscular dystrophy[33, 45, 53] [SoR: A; SoE: IV].

Discharge criteria (*e.g.* when and why to end PRM interventions)

30. It is recommended that patients with muscular dystrophy are being followed up by a multiprofessional rehabilitation team that is led and coordinated by the PRM physician together with the pediatrician in young age from first onset of symptoms, during transition to adulthood and later on, taking into consideration the patients` physical and psychological conditions and their needs including but not limited to: nursing and medical requirements, rehabilitation goal attainment, home and community settings and family or caregiver situation[76, 77] [SoR: B; SoE: IV].

Follow-up criteria and agenda

31. It is recommended that PRM physicians manage the postdischarge follow-up plan on regular basis keeping in mind physical, cognitive, psychosocial and functional status in patients with muscular dystrophy[78] [SoR: A; SoE: IV].

E. Recommendations on future research about best PRM professional practice in persons with muscular dystrophies

32. It is recommended that PRM physicians participate in multidisciplinary projects within systematic research regarding the effects and benefits of certain rehabilitation modalities and interventions in patients with muscular dystrophy. Better understanding of epidemiology, pathophysiology, risk and prognostic factors along with survival rates will help in better optimization of rehabilitation resources and more effective short- and long-term treatment planning. Further research in technical aids, along with advancements in disability surgery will have significant impact on quality-of-life improvement in these patients. Interventions to provide the necessary assistance to caregivers taking into account the risk to their health, or preventive screening of possible disorders in transmitting mothers (ie. Duchenne muscular dystrophy)[79-82] [SoR: A; SoE: I].

33. With the advancement of gene therapy both ex vivo and in vivo, it is important to avoid diagnostic errors, since at this stage of medical technology it is possible to carry out full genome searches (or DNA microarrays depending on clinical presentations). The PRM physician participates in avoiding a wandering of the genetic diagnosis and in screening heterozygous people or transmitters with the help of the geneticist[53, 83-85] [SoR: B; SoE: IV].

Limitations of the study

The article presents the role of the PRM physician in the diagnosis and treatment of patients with muscular dystrophies and is not considered a complete guideline. Furthermore, considering the time-consuming process of literature search and inclusion in recommendations, along with the Delphi voting procedure, additional new literature has not been added.

Conclusions

The role of the PRM physician is to assess the functional status of persons with muscular dystrophy and, to plan, monitor and lead PRM program in an interdisciplinary setting within a multiprofessional team, to lead the Individual Rehabilitation Project defining the goals and monitor the outcome. Furthermore, the PRM physician should focus to promote functioning to the maximum possible level and integration in the community, while taking care of the musculoskeletal and psychomotor development of the person, prevent complications and provide education and counselling to patients and caregivers. Moreover, the PRM physician should avoid diagnostic errors in the event of muscular dystrophies, given genetic progress (diagnostic and therapy), foster child-adult rehabilitation transitions and support the new assessments necessary for new treatments. This EBPP represents the official position of the the UEMS PRM section and describes the professional role of PRM physicians in muscular dystrophy.

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