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Awareness of bone strength in patients with neuromuscular disorders: ERN EURO-NMD clinician survey and European patient survey

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ABSTRACT

Bone strength is reduced In various neuromuscular disorders (NMDs). We aimed to assess the awareness and practice of bone strength management in NDMs among clinicians and patients. We performed two online surveys; among health care providers (HCPs) of the European Reference Network for Neuromuscular Disorders (ERN EURO-NMD) and among patients. The survey among 52 HCPs showed that awareness of potentially impaired bone strength in people with NMDs was reasonable to good: the vast majority of HCPs asked often or almost always about bone fractures during history-taking (81 %). Bone strength was less often assessed: often or almost always at diagnosis (50 %) and at follow-up (58 %). Medical training on this topic was considered poor to very poor in 50 % of HCPs. Prevention and treatment of reduced bone strength was variable and multidisciplinary care was sub-optimal. The survey among 581 patients provided important additional insights. Many patients were followed-up outside ERN EURO-NMD centers and treatment was variable. These parallel surveys provided a broad view on the awareness and management of bone strength in people with NMDs. The findings are expected to increase the appreciation of this important aspect of NMD care, and direct future research foci and care guidelines.

1. Introduction

Osteoporosis is a disease defined by decreased bone mass and alteration of microarchitecture which results in bone fragility and increased risk of fracture. In adults, primary osteoporosis is defined by bone loss arising from physiological changes of menopause and/or ageing. Furthermore, primary osteoporosis refers to intrinsic skeletal defects of genetic origin or idiopathic juvenile osteoporosis. Secondary osteoporosis is defined as osteoporosis arising from underlying systemic illnesses or their treatment. Factors implicated in the development of secondary osteoporosis include neuromuscular diseases (NMDs), muscle disuse, iatrogenic causes such as steroids, chronic inflammation, delayed or arrested puberty and hematological disorders.

in NMD, bone strength may be compromised due to different pathomechanisms. Modified muscle-bone "cross-talk", and loss of biomechanical loading due to sub-normal mobility are likely to play a pivotal role. Nutritional issues, including malnutrition and swallowing problems that affect caloric and micronutrient intake (including calcium and

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vitamin D) can contribute as well. Furthermore, low levels of physical activity, muscle weakness, immobility are common and might lead to overweight or obesity without appropriate, adaptive increases in bone mass and density to maintain adequate bone strength. Finally, specific disease-modifying drugs are a common cause of osteoporosis e.g., chronic glucocorticoid therapy, which is the standard of care for Duchenne muscular dystrophy (DMD), myositis, and often myasthenia gravis [1].

In 2022, we have performed a review on bone strength in congenital myopathies [1]. This scoping review showed that long bone fractures (LBFs) are common in pediatric and adult patients with a congenital myopathy. Prospective data on the prevalence of low bone mineral density (BMD) and LBFs were lacking. Subsequently, we performed a one-year prospective natural history study in 21 LAMA2-MD and 10 SELENON-RM patients. Ninety percent of the LAMA2-MD and SELENON-RM patients showed low bone mineral density. Eight (38 %) LAMA2-MD and five (50 %) SELENON-RM patients had a history of fragility LBFs. During the one-year follow-up period, one LAMA2-MD patient (female, 3 years of age) experienced a fragility LBF of the right humerus. We found no difference in BMD between baseline and one-year follow-up [1].

A limited number of studies have assessed bone strength in other hereditary NMDs, such as myasthenia gravis and inflammatory myopathies, in the last decade [2–18]. The "2018 Care Considerations" for bone strength in DMD, which have been discussed in European Neuromuscular Centre (ENMC) workshops, provided international expert- and evidence-based guidance that reflected the knowledge and understanding at the time of writing [19,20]. Now, six years later, more knowledge is needed to understand the natural history of bone strength, optimal diagnosis and monitoring approaches, best candidates for treatment, the treatment strategies that provide the most acceptable benefit-to-risk profile to patients, and the most appropriate outcome measures for gauging treatment responses in the clinical and drug trial settings. To raise awareness and to reach consensus on bone strength screening and treatment to optimize bone strength in various NMDs, we have organized an ENMC workshop ("ENMC Recommendations for Optimizing Bone Strength in Neuromuscular Disorders", January 19th – 21st 2024). To assess the awareness and practice of bone strength management in NMDs, we have performed two international online surveys. We invited HCPs for a survey through the European Reference Network (ERN) EURO-NMD (https://ern-euro-nmd.eu). In parallel, patients were invited for a patient-oriented survey via national patient organizations. The findings of both studies are expected to increase the appreciation of multidisciplinary NMD management.

2. Methods

2.1. Survey for health care providers (HCPs)

We conducted a survey among HCPs of the ERN EURO-NMD. These 83 centres are all specialized in the care of patients with NMDs. The survey focused on the awareness of diagnostic tests and treatments of bone strength in patients with NMDs. It was created by the organizers of the aforementioned 2024 ENMC Workshop (Drs. Leanne Ward [Canada], Antimo Moretti [Italy], David Weber [USA], and Nicol Voermans [the Netherlands]) and the patient representatives invited to this workshop (Madelon Kroneman, Ingrid de Groot [the Netherlands] and Silke Schlüter [Germany]). The scoping review and longitudinal study by Bouman et al. offered inspiration for the questions about medical bone history [1,21]. The list of inherited NMDs was based on the Genetable of NMDs (https://www.musclegenetable.fr). Questions about the frequency provided five answer categories: Almost always; Often; Sometimes; Seldom; Never. If more than one answer was possible, this was specifically reported ('≥ 1 answer possible'). After its design, the HCP survey was reviewed by T. Evangelista, ERN-EURO-NMD coordinator, and then was tested by the research group (**Supplemental Data 1**). It was approved by the local ethical committee at Radboud university medical center (*CMO 2023–16,873*).

Invitations to participate were sent by email to all HCPs on December 22nd 2023. HCPs were encouraged to discuss the questions and their associated answers among their health care team members and provide a team rather than an individual response. The email contained a link to the online survey created in Castor (https://www.castoredc.com/). Seven email addresses were corrected by the ERN-EURO-NMD office after receiving a bounce-back. Reminders were sent on January 8th and March 6th 2024 and the survey was closed on April 1, 2024. The full manuscript was sent out for approval to all participating HCPs on July 15th 2024. Reminders to review the full manuscript were sent on August 8th and August 14th 2024. All HCPs that completed the survey on behalf of their health care teams and reviewed plus approved the manuscript were included as co-authors.

To prepare for a presentation at the (aforementioned) 274th ENMC workshop in Hoofddorp, January 19th – 21st 2024, an interim analysis was performed on data available up until and including January 14th, 2024. A summary of the results of this analysis is included in the ENMC lay and full reports (https://www.enmc.org/workshops/past-workshops/) {Dittrich 2024}.

2.2. Survey for patients

The patient survey was created to collect experiences from NMD patients with regards to bone fractures, bone health screening, and treatment of osteoporosis. Participation was open to all (parents of pediatric) patients with NMDs, irrespective of their bone health history. The survey was composed in English by three patient representatives (**Supplemental Data 2**) and was translated into Dutch, German, French and Norwegian by patient representatives of these countries. The protocol was approved by the local ethical committee at Radboudumc (CMO *2024–17,129*).

The invitation to participate in the survey was tailored to maximize responses in each country (The Netherlands, Germany, France, and Norway): newsletters from the patient organizations and Facebook. The survey was programmed using the EU-Survey tool (http://ec.europa.eu/eusurvey/). The link to the survey directed the patients to their native language. It was available online from 14 December 2023 up to 15 January 2024.

Most questions were closed, except for the option "other" in some questions. The answers were translated to English with the help of the translation program DeepL Translator (http://www.deepl.com) and categorized by hand.

2.3. Statistical analysis

Standard descriptive statistics were used to summarize and report survey data. If a question was completed by less than all respondents, this was noted. If more than one answer was possible, this was reported as '≥ 1 answer possible'.

3. Results

3.1. Survey for health care providers (HCPs)

3.1.1. Introduction

Of the 83 invited HCPs, 52 responses were received from 50 different HCPs in 18 countries (HCP response rate 63 %). Fig. 1 shows the geographical locations of HCP. Italy had most participating HCPs, followed by the Netherlands and Germany. All but one HCP were from academic hospitals. The medical specialties of the HCPs were neurology ($n = 32$; 62 %), pediatric neurology ($n = 15$; 29 %), pediatrics (n = 3; 6 %), and other ($n = 2$; 4 %). The responses mentioned in the paragraphs below are of the whole group. For most questions, all 52 participants had provided an answer. If not, the number of HCPs was reported. The

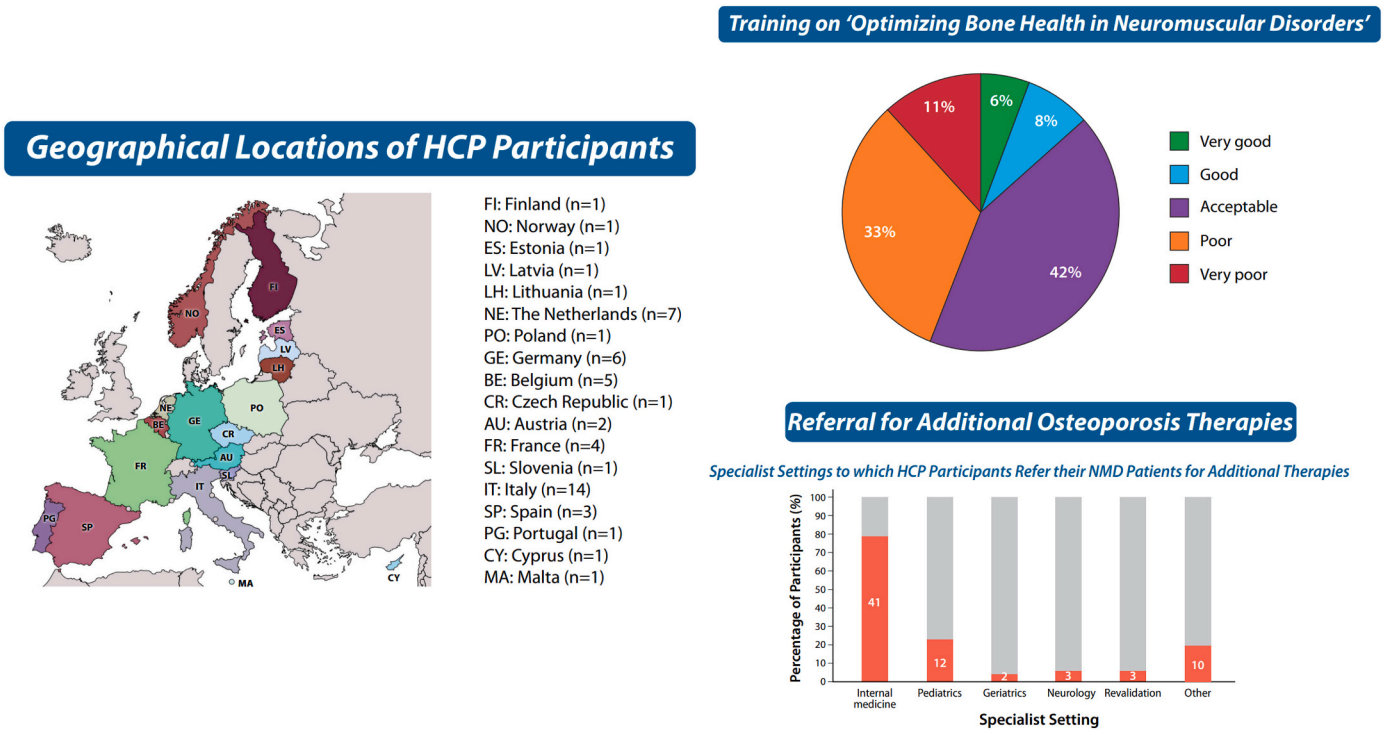

Fig. 1. Geographical locations. Fig. 1 shows the geographical locations of HCP participants and the number of HCP participants per country.
Training on 'optimizing bone health in neuromuscular disorders'. Figure showing how HCP participants would rate their training on 'optimizing bone health in neuromuscular disorders'. Total $n = 52$.
Referral for additional therapy. Figure showing to which specialist settings HCP participants refer NMD patients for additional therapy. Total n = 52. More than one answer possible.

overview of the responses to all questions in the survey is available as **Supplemental Data 3.**

3.1.2. Training on optimizing bone health

The HCPs were asked about how they rated their medical training on 'optimizing bone health in neuromuscular disorders'. Only three HCPs rated this as very good (6 %) and four as good (8 %); the others rated this as acceptable, poor or very poor ($n = 45$; 87 %; Fig. 1).

3.1.3. Monitoring bone strength in the diagnostic phase and during follow-up of the neuromuscular disorder

HCPs were asked to report the clinical scenarios in which they initiated bone strength assessment(s) in patients with NMDs (≥ 1 answer possible), as follows: as part of primary prophylaxis before a first fracture ($n = 37$), as part of secondary prophylaxis after one fracture ($n = 21$), or as part of secondary prophylaxis after two or more fractures (n = 2).

Table 1 shows the frequency with which specific features of bone strength was assessed during medical histories and ancillary investigations in the diagnostic and follow-up phases of bone health care in NMD; history of bone fractures, the mechanism of injury in the event of a fracture, and the history of vertebral fractures. Overall, the vast majority of HCPs asked often or almost always about bone fractures during history-taking ($n = 42$; 81 %). Bone strength was less often assessed: often or almost always at diagnosis ($n = 26$; 50 %) and at follow-up ($n = 30$; 58 %).

Fig. 2 shows the wide variation in criteria used by HCPs to define osteoporosis (≥ 1 answer possible), in children and in adults. This variability was underscored by the answers provided in the category 'other', including DXA reports from the radiologist, referral to the rheumatologist or endocrinologist and laboratory testing (high alkaline phosphate, low vitamin D).

Bone mass, structure and density assessment were also variable (both in the diagnostic and follow-up phases; (≥ 1 answer possible): DXA was used most commonly to assess BMD ($n = 50$; n = 50), followed by digital X-ray radiogrammetry (n = 5; $n = 4$) and peripheral quantitative computed tomography (pQCT: 3D assessment of bone structure, composition and geometry at the radius and tibia; $n = 2$; n = 2). Other techniques were X-ray of the spine for vertebral fracture evaluations, and consultation with a rheumatologist. The next question concerned the skeletal sites that were assessed with DXA scan (≥ 1 answer possible; Fig. 2), showing similar diagnostic and follow-up practices. The spine, lateral distal femur, and (total) hip were most frequently assessed. Fig. 2 shows which additional tests HCPs perform in the diagnostic process and during follow-up visits, with a similar range of tests at both time points (≥ 1 answer possible). The most common serum measurements were vitamin D, calcium, phosphate, alkaline phosphate and creatine. Other tests were parathyroid hormone, calcitonin, cortisol and referral to an endocrinologist.

The four disorders in which HCP participants most frequently performed bone strength screening at follow-up visits (≥ 1 answer possible) were muscular dystrophies ($n = 48$), congenital muscular dystrophies ($n = 37$), congenital myopathies ($n = 35$), inherited motor neuron diseases ($n = 33$) and metabolic myopathies ($n = 24$). Acquired disorders for which steroids are commonly prescribed were less often mentioned: inflammatory myopathies ($n = 21$), myasthenia gravis ($n = 19$), and inflammatory neuropathies ($n = 9$).

HCPs reported checking for vertebral fractures during follow-up visits (≥ 1 answer possible) according to the following scenarios (≥ 1 answer possible): once the neuromuscular disorder was diagnosed ($n = 43$) or at the initiation of glucocorticoid therapy or during follow-up visits ($n = 31$). Other scenarios where HCPs reported checking for vertebral fractures were the following: 1. in patients with DMD, 2. routinely every year after initiation of glucocorticoid therapy, 3. according to the clinical condition (back ache, history of long-bone fractures, poor dietary status, severe spinal deformities, loss of ambulation),

Table 1
The components of a comprehensive bone health assessment and their associated frequencies during the diagnostic and follow-up phases of care.

Do you take a history of bone fractures (e.g. history of long bone fractures)?					
	Almost always	Often	Sometimes	Seldom	Never
Diagnostic process (n = 52), %	34 (65,4 %)	8 (15,4 %)	8 (15,4 %)	1 (1,9 %)	1 (1,9 %)
Follow-up (*n* = 52), %	26 (50,0 %)	18 (34,6 %)	5 (9,6 %)	3 (5,8 %)	0 (0,0 %)
When taking a fracture history: do you obtain a history about the mechanism of injury?					
	Almost always	Often	Sometimes	Seldom	Never
Diagnostic process (n = 51), %	32 (62,7 %)	13 (25,5 %)	5 (9,8 %)	1 (2,0 %)	0 (0,0 %)
Follow-up (n = 52), %	32 (61,5 %)	14 (26,9 %)	5 (9,6 %)	0 (0,0 %)	1 (1,9 %)
Do you assess vertebral fractures?					
	Almost always	Often	Sometimes	Seldom	Never
Diagnostic process (n = 52), %	16 (30,8 %)	17 (32,7 %)	10 (19,2 %)	7 (13,5 %)	2 (3,8 %)
Follow-up (n = 52), %	12 (23,1 %)	19 (36,5 %)	13 (25,0 %)	6 (11,5 %)	2 (3,8 %)
Do you take a history of dietary intake of calcium?					
	Almost always	Often	Sometimes	Seldom	Never
Diagnostic process (n = 52), %	20 (38,5 %)	10 (19,2 %)	12 (23,1 %)	8 (15,4 %)	2 (3,8 %)
Follow-up (n = 52), %	15 (28,8 %)	17 (32,7 %)	11 (21,2 %)	8 (15,4 %)	1 (1,9 %)
Do you check for bone mineral density?					
	Almost always	Often	Sometimes	Seldom	Never
Diagnostic process (n = 52), %	15 (28,8 %)	11 (21,2 %)	17 (32,7 %)	7 (13,5 %)	2 (3,8 %)
Follow-up (n = 52), %	11 (21,2 %)	19 (36,5 %)	14 (26,9 %)	6 (11,5 %)	2 (3,8 %)

4. in case of pain or other fractures, 5. depending on the severity of the weakness and ambulation, and 6. in case of a fall. Fig. 2 shows how frequently HCPs assess vertebral fractures and perform DXA scans at follow-up visits (≥ 1 answer possible), highlighting a wide range of answers.

3.1.4. Treatment, consultation and guidelines

The next part of the survey was on treatment, consultation and guidelines. The range of treatments prescribed for patients diagnosed with a NMD and osteopenia or osteoporosis was very wide (≥ 1 answer possible): bisphosphonates (oral or iv; *n* = 27 and *n* = 25, respectively), vitamin D supplementation (*n* = 51), and calcium supplementation (*n* = 36). Other responses included referral of patients to a rheumatologist, endocrinologist or bone health specialist otherwise.

Table 2 shows the recommended treatments. Optimizing physical activity was almost always recommended (n = 36; 69 %) or often (*n* = 14; 27 %). In addition, many HCPs referred their patients to a physiatrist for rehabilitation approaches (almost always *n* = 34; 65 %; often *n* = 13; 25 %). The majority of HCPs referred patients to adapted sports programs if they were not able to optimize physical activity on their own. The rehabilitation-specific approaches prescribed by HCPs were (≥ 1 answer possible): strengthening exercises (*n* = 40), aerobic training (n = 36), (supported) standing programs (*n* = 33) and balance training (n = 25).

A majority of HCPs recommended vitamin D supplementation (almost always: *n* = 27; 52 %, often: *n* = 18; 35 %) and dietary calcium intake (almost always: n = 18; 35 %, often: n = 18; 35 %) Referral to a dietician was considered less often (almost always: *n* = 6; 12 %, often: *n* = 11; 21 %). The dose of vitamin D was adjusted according to (≥ 1 answer possible): the patient's previously measured vitamin D status (n = 40), the patient's age (*n* = 30), sex (n = 13), the amount of sun exposure (*n* = 12), skin colour (*n* = 10) and pregnancy status (*n* = 7). The recommended calcium dose was adjusted according to (≥ 1 answer possible): the patient's previously measured calcium status (n = 30), age (*n* = 28), sex (*n* = 17), and pregnancy status (*n* = 8).

Fig. 1 shows the referral to other medical specialists and additional therapies (≥ 1 answer possible). Most referrals were to internal medicine (*n* = 41) and pediatrics (n = 12). Other options included rheumatology (n = 7), cardiology, orthopedics and pediatric endocrinology. The majority of HCPs reported that their centre had a bone clinic (*n* = 32; 62 %). Fifteen HCPs (29 %) reported that there was no bone clinic, and five (10 %) reported that they were not aware of this. Medical specialists included in bone clinics were (≥ 1 answer possible): endocrinologist (*n* = 21), pediatrician (n = 10), internist (*n* = 9), rehabilitation physician (n = 9), neurologist (*n* = 5), pediatric neurologist (n = 4), nurse practitioner (n = 4) and osteologist (n = 4). Other responses included rheumatologist (n = 4), nephrologist and geriatrician.

One third of the HCPs reported that their centre had a local guideline on bone strength monitoring (n = 17). The majority of HCPs (*n* = 31; 60 %) reported that there were no local guidelines, and four HCPs (8 %) did not know whether guidelines existed, or not, at their centre. The most frequently used published guideline (≥ 1 answer possible) was on bone health and osteoporosis therapy in DMD (*n* = 38) [22] and diagnosis, rehabilitation, orthopedic and nutritional care in SMA (*n* = 29} [23].

3.2. Survey for patients

3.2.1. Characteristics of the respondents

In total, 581 patients with a wide range of NMDs responded to the survey. The largest groups were diagnosed with hereditary motor sensory neuropathy (HMSN) / Charcot-Marie-Tooth Disease (CMT; *n* = 87; 15 %), (facioscapulohumeral muscular dystrophy (FSHD; *n* = 81; 14 %), limb girdle muscular dystrophy (LGMD; *n* = 52; 9 %), spinal muscular atrophy (SMA; *n* = 41; 7 %), myotonic dystrophy (MD; n = 41; 7 %) and myositis (*n* = 35; 6 %). Forty-two percent had another diagnosis (*n* = 244). Of the respondents, 66 % (*n* = 383) were able to walk (with or without walking aids), 15 % (n = 87) were part-time wheelchair users and 18 % (*n* = 105) were full-time wheelchair users. Eighteen percent (n 105) of the respondents used corticosteroids for at least one year or more. An overview of the responses to all questions in the survey is available in **Supplemental Data 4.**

3.2.2. Experiences with fractures

About one third of the respondents (*n* = 173; 30 %) experienced a fall with a fracture in the past five years. This was not specified for location. As a result, 46 % (*n* = 80) experienced reduced mobility afterwards and 10 % (n = 17) lost their ability to walk. A fracture of the arm, hand, or finger was also reported to impact mobility, since these fractures restricted the use crutches or a rollator.

3.2.3. Diagnosed with osteoporosis/osteopenia

A minority of participants was diagnosed with osteoporosis (*n* = 95; 16 %) or osteopenia (*n* = 16; 5 %). The majority (*n* = 345; 59 %) had no previous bone strength assessments or diagnosis, and 19 % did not know

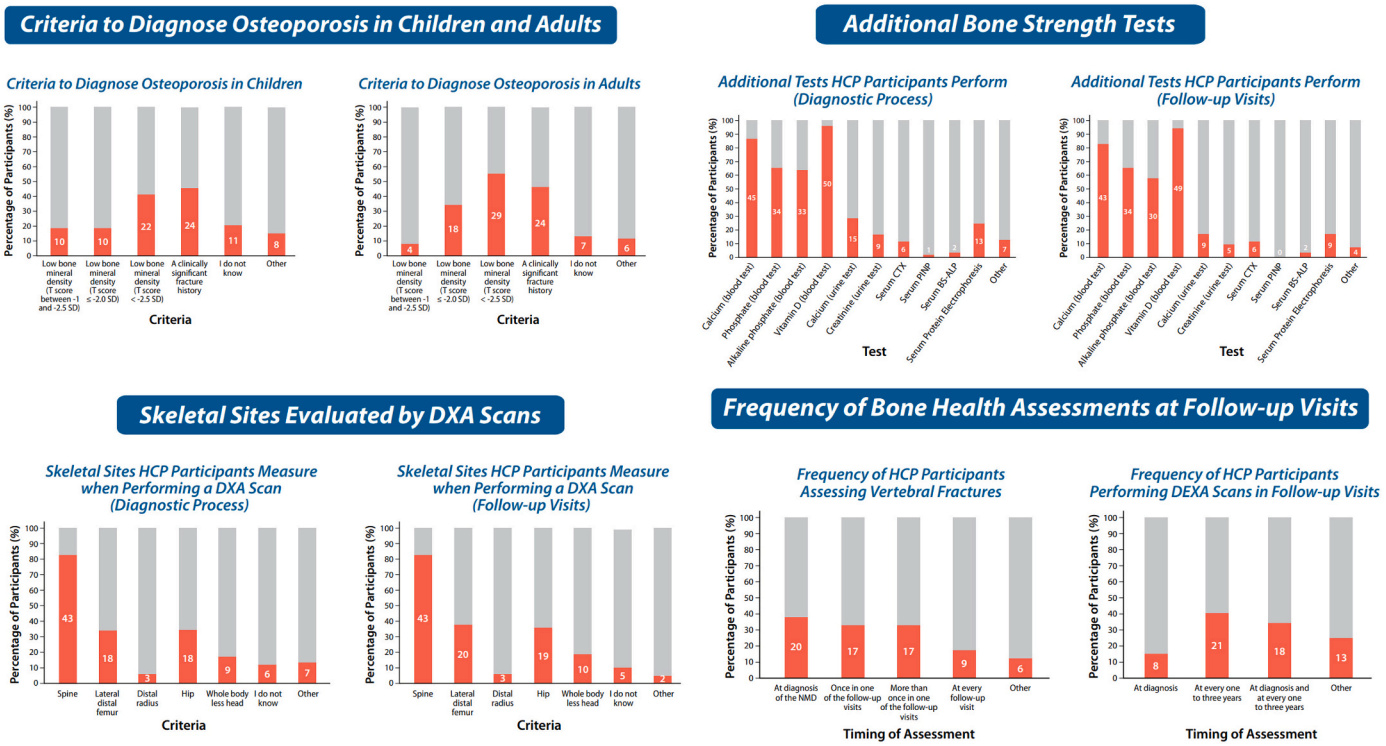

Fig. 2. Criteria to diagnosis of osteoporosis in children and adults. This figure shows which criteria need to be met to define osteoporosis in children and adults. More than one answer possible; Total $n = 50$.
Skeletal sites DXA scan. Figure showing which skeletal sites HCP participants measure when performing a DXA-scan in the diagnostic process and in follow-up visits. Total n = 52. More than one answer possible.
Additional tests. Figure showing which additional tests HCP participants perform in the diagnostic process and at follow-up visits. Total n = 52. More than one answer possible.

Table 2
Treatments recommended by Health Care Professionals to patients with NMDs.

If possible, do you recommend the NMD patient to optimize physical activity? (n = 52), %				
Almost always	Often	Sometimes	Seldom	Never
36 (69,2 %)	14 (26,9 %)	2 (3,8 %)	0 (0,0 %)	0 (0,0 %)
Do you recommend the NMD patient to refer to a physiatrist for rehabilitation approaches?				
Almost always	Often	Sometimes	Seldom	Never
34 (65,4 %)	13 (25,0 %)	3 (5,8 %)	2 (3,8 %)	0 (0,0 %)
If the NMD patient is not able to optimize physical activity on their own, do you refer this patient to an adapted sports program?				
Almost always	Often	Sometimes	Seldom	Never
12 (23,1 %)	24 (46,2 %)	9 (17,3 %)	5 (9,6 %)	2 (3,8 %)
Do you recommend vitamin D supplementation to the NMD patient?				
Almost always	Often	Sometimes	Seldom	Never
27 (51,9 %)	18 (34,6 %)	6 (11,5 %)	1 (1,9 %)	0 (0,0 %)
Do you recommend sufficient dietary calcium intake to the NMD patient?				
Almost always	Often	Sometimes	Seldom	Never
18 (34,6 %)	18 (34,6 %)	14 (26,9 %)	2 (3,8 %)	0 (0,0 %)
Do you refer patients to a dietician to optimize the NMD patient's dietary calcium intake?				
Almost always	Often	Sometimes	Seldom	Never
6 (11,5 %)	11 (21,2 %)	19 (36,5 %)	10 (19,2 %)	6 (11,5 %)

NMD = neuromuscular disorders.

whether they had such assessments, or not. Over half of the respondents ($n = 69$; 55 %) with a diagnosis of osteoporosis/osteopenia received this diagnosis after the first fracture.

Osteoporosis was not limited to patients in wheelchairs. One out of five walkers and one out of five part-time wheelchair users were diagnosed with osteoporosis or osteopenia. Only one third of the patients received their diagnosis in an academic medical centre, the others received their diagnosis either in local or regional hospitals ($n = 52$; 42 %) or from an ambulatory medical specialist practices (mainly in Germany, where medical specialists have independent ambulatory practices not connected to a hospital). One out of four corticosteroid users was diagnosed with osteoporosis or osteopenia.

3.2.4. *Treatment*

Patients with a diagnosis of osteoporosis or osteopenia were predominantly treated with vitamin D ($n = 68$; 54 % prescribed; $n = 26$; 21 % patient's initiative). About half of the respondents took calcium supplementation (53 %) and/or were treated with osteoporosis medications such as bisphosphonates. One out of 10 was not treated for this condition, while 3 % ($n = 4$) reported other treatments, such as denosumab injections. Among those treated with bisphosphonates, one out of five ($n = 22$; 20 %) experienced side effects. For 36 % ($n = 8$) of the respondents who experienced side effects these were severe enough to stop the treatment. The most frequently mentioned side effects (≥ 1 answer possible) were intestinal problems (n = 8), muscle or joint pain ($n = 7$), feeling ill (n = 5) and fever (n = 4). Those who stopped treatment mostly mentioned muscle or joint pain. Those who did not stop treatment due to side effects mostly reported intestinal problems.

4. Discussion

4.1. Multiperspective view

These parallel surveys among ERN-EURO-NMD clinicians and patients with a NMD provided a multiperspective view on the awareness and management of bone strength in NMDs. The survey among HCPs showed that the reported awareness about reduced bone strength in NMDs is reasonable to good, both at diagnosis and at follow up. However, medical training on this topic was considered moderate to poor, an observation which identifies a potentially modifiable barrier to effective bone health care. The approach to bone strength assessments and the prevention plus treatment of reduced bone strength were both highly variable. The survey among patients provided important additional insights, including the fact that fractures traditionally considered trivial, such as finger fractures, can significantly interfere with patients' abilities to use mobility devices. Overall, the reported awareness among patients for bone strength was poor to moderate. Osteoporosis and osteopenia were not limited to full-time wheelchair users or patients using glucocorticoids. The main findings are discussed below.

4.2. Survey for HCPs

The first remarkable finding was the poor rating of the received medical training on bone strength in NMD. These responses were not stratified for years of working experience. Enhanced training and awareness is expected to improve attention to management of bone strength in the clinical setting. A recent study evaluating the impact of educational intervention on primary care providers' knowledge, confidence and frequency of patient counselling with respect to strength training and BMD also showed that provider knowledge about and frequency of counselling patients about strength training were low. Brief, interactive, small group educational sessions with specific recommendations were shown to increase provider-reported confidence and rates of strength training advice to their patients. It was suggested that training HCPs in this way could be easily duplicated and studied in other health systems. In addition, making educational materials on muscle strength training and bone health widely available to patients and providers was expected to encourage providers to increase counselling on this topic [24].

Many HCPs reported that they initiate bone strength assessment(s) in NMD patients as primary prophylaxis and that they often assess the history of various bone fractures. This was neither in line with the patients' survey nor with the results of our recent scoping review on bone quality in patients with a congenital myopathy [1,21]. This review included many patients with long bone fragility fractures and concluded that unselected cross-sectional studies are needed to carry out prevalence estimations on decreased bone quality in patients with (congenital) myopathies. Further, long-term follow-up studies are expected to reveal the evolution of bone strength over time to optimize recommendations for optimizing BMD in specific groups of NMDs.

Assessments of bone mass, structure and density and the sets of criteria used by HCPs to define osteoporosis in children and in adults were variable. DXA scan was used by most of HCPs to evaluate BMD. That is in line with available general guidelines for the assessment of BMD in children and adults. The definition of osteoporosis is general: the condition characterized by compromised bone strength, leading to an increased risk of fragility fractures. However, the diagnostic criteria for low BMD, osteopenia, and osteoporosis in children and adults (according to the International Society for Clinical Densitometry (ISCD) [25]) were moderately to poorly adhered, suggesting that knowledge implementation is required. Notably, the ISCD recommendations for the acquisition and analysis of DXA scans differs for children versus adults. The survey did not specifically ask respondents to differentiate between children and adults for questions related to DXA utilization, limiting the interpretation of these data. Nevertheless, the spine is one of the skeletal sites recommended by the ISCD in both pediatric and adult patients [26].

4.3. Survey for patients

The survey among patients showed additional insights. About one third of the respondents had experienced a fracture in the past five years. Approximately half experienced reduced mobility afterwards and 10 % lost their ability to walk. One fifth of participants was diagnosed with osteoporosis or osteopenia, most often after the first fracture. Osteoporosis and osteopenia were not limited to full-time wheelchairs users or those receiving corticosteroids. A majority of patients was not diagnosed or did not know about the status of their bone strength. The prevalence of osteoporosis and osteopenia in this cohort of people was therefore likely underestimated based on this part of the study. The prevalence of fractures in this cohort of people with various NMDs (30 % in past 5 years) was in line with what we have observed in a one-year prospective natural history study of patients with LAMA2-muscular dystrophy (n = 21) or SELENON-related myopathy (n = 10). Eight (38 %) LAMA2-MD and five (50 %) SELENON-RM patients had a history of a low-trauma LBF [21]. Remarkably, prospective assessment showed low BMD in 90 % of the participants [21]. This is much higher than the prevalence reported in the current survey. This is most likely related to the severity of muscle weakness in these conditions and the prospective assessment of bone strength in an unselected cohort [1].

Since a majority of patients was diagnosed in non-specialized centres, awareness of the impact of fractures on patients' lives and the need for early screening needs to be communicated with local hospitals and practices as well. This is an important mandate of HCPs with sufficient knowledge of and experience with bone strength issues in people with NMDs. Another important aspect unveiled by this survey was the clinically significant impact of upper limb fractures (www.enmc.org/download/enmc-recommendations-for-optimizing-bone-strength-in-neuromuscular-disorders/), including complete, premature loss of ambulation, inability to walk with the assistance of mobility aids, and loss of the ability to navigate an electric wheelchair.

4.4. Comparison of survey among HCPs and survey among patients

The differences in responses of clinicians and patients provide guiding insights, although it is important to note that the survey questions were different so direct comparisons were not possible. These differences might be partly related since the clinicians were all part of the EURO-NMD centres, whereas patients reported being followed up both in academic and other centres. Despite that, the parallel surveys shed light on the suspected ascertainment bias from both the clinician and patient perspective, but it also pointed to aspects that would generally not receive attention by one or the other the groups in isolation. For example, hand and foot fractures were noted to negatively impact quality of life by patients (as discussed earlier), whereas these are typically not considered clinically significant fractures by clinicians. Finally, the patient survey reached a very high number of responses in a short time frame. Although we could not assess the response rate directly, we think that this underscores the relevance of this topic for patients with NMDs.

4.5. Interdisciplinary topic

Several parts of the survey showed that the diagnosis and management of compromised bone strength is a truly multidisciplinary field. The expertise needed to administer medical treatments for reduced bone strength is generally considered to be highest among specialized (pediatric) endocrinologist and rheumatologists. However, patients receive long-term follow-up by rehabilitation specialists and neurologists. The medical training of HCPs does not generally include this special knowledge, and the topic is not included in further training activities.

This was reflected in the moderate to poor quality of training available on bone strength in NMD. As such, the topic of bone strength in NMDs falls between the working fields of the endocrinologist, rheumatologist and neuromuscular specialist and might therefore be classified as interdisciplinary. We think that awareness among pediatric and adult neurologists about bone strength should be amplified, and that multidisciplinary guidelines can provide a basis for what part of management can be done by which clinicians.

Promotion of bone strength is important in the general population and in various groups of patients with chronic disorders, especially for those who do not fit the normal osteoporosis risk profile [27,28]. Remarkably, the majority of HCPs reported that their centre had a specialized multidisciplinary bone clinic, including a wide range of medical specialists. While promising, only one third of HCPs reported the presence of a local guideline for bone strength management in NMDs. The study among patients illustrated the importance of guidelines since many patients were diagnosed with reduced bone strength in a centre other than a specialized EURO-NMD centre.

4.6. Attention for bone strength in NMDs in existing guidelines

The limited information in guidelines beyond DMD and SMA, and the lack of specific considerations for these vulnerable groups in terms general osteoporosis guidelines, reveal critical gaps fueling the patients' unmet needs. Most osteoporosis guidelines focus on postmenopausal osteoporosis, providing little relevant guidance for patients with secondary osteoporosis, including those with NMDs. In parallel, many disease specific guidelines provide only general guidance on optimizing BMD, e.g. in Pompe disease [29], congenital myopathies [30], and congenital myasthenic syndrome [31]. Furthermore, patients with NMDs are may be excluded from the development and/or validation of novel techniques to evaluate bone strength; one such example is The Trabecular Bone Score, a tool that has been developed to refine identification of patients at risk for osteoporotic fractures [32]. This observation highlights a significant shortfall in the inclusivity of current medical guidelines, indicating a pressing need for more comprehensive, but at the same time specific, guidance to address the unique needs of people with NMD.

4.7. Strength and limitations

This combined survey has a number of strengths and limitations. First, conducting the study among the two groups collaboratively and in parallel is a strength. The patients contributed to the questionnaire for the HCPs and the clinicians commented on the patients' survey. The patient-researcher team originated from the Muscle working group of ERN-EURO-NMD and was multinational. The clinician-researcher team was multidisciplinary (combining expertise on bone strength with expertise on NMDs) and multinational. This process was a co-creation between patients and clinicians worldwide, clearly illustrating the strength of patient participation in EURO-NMD activities. Second, both surveys had high response rates in a short period. The patient survey resulted in 581 responses in a single month. A precise frequency of responses over a defined time period could not be calculated because the invitation for the survey was sent out and placed online at the same time. The HCP survey reached a response rate of 60 % in four months.

This bias among clinicians affiliated with ERN EURO-NMD centers could be considered a limitation. By inviting the coordinator of each EURO-NMD site, we created an ascertainment bias in the cohort of clinicians taking care of patients with NMD in Europe. However, we deliberately performed this study among EURO NMD centres since they are considered experts in the diagnosis and management of patients with all NMDs, and we were interested in the extent to which bone strength awareness was present and management was provided in the EURO-NMD centres. Furthermore, HCPs were encouraged to discuss among their team members and provide a team rather than an individual response. We did not specifically verify whether this took place, which is another potential limitation of this study. Finally, we cannot rule out the possibility that HCPs were likely to present what they think that they should do rather than what they actually carry out in current clinical practice, a phenomenon that is inherent to any potential survey. A similar bias is present for the patient survey: patients who have experienced bone health issues might have been more likely to participate to the survey, although the call for participation did deliberately invite all patients with a NMD to participate.

5. Conclusions

This combined survey underscores significant need for improved bone strength management strategies and implementation in patients with various hereditary and acquired NMDs. Multi-disciplinary prevention and treatment of bone strength loss should be considered the cornerstone of timely and effective bone health management; effective management should also be considered so important that is should be embedded in routine, day-to-day clinical practice. This was also discussed during the recent 274th ENMC 2024 Workshop. [33] We propose that knowledge about the natural history of bone strength needs to be enhanced by natural history studies in specific disease cohorts. This will enable optimizing management, including best timing for diagnosis and monitoring, and the most proper pharmacological and non-pharmacological approaches to suggest in this population. With optimization of bone strength monitoring, diagnosis and treatment, we anticipated that the prevalence of fragility fractures in patients with NMDs will fall, with the potential for significant improvement in quality of life.

Frequency assessing bone health at follow-up visits. Figure showing how frequent HCP participants assess vertebral fractures and perform DXA-scans at follow-up visits. Total n = 52. More than one answer possible.

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Declaration of competing interest

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Data availability

The data supporting the findings of this study (full set of responses of both surveys) are available within its supplementary material from the corresponding author upon reasonable request.

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