Molecular Genetics and Metabolism Reports 1 (2014) 85–97

Contents lists available at ScienceDirect

Molecular Genetics and Metabolism Reports

journal homepage: http://www.journals.elsevier.com/molecular-genetics-and-metabolism-reports/

# Risk factors for osteoporosis, falls and fractures in hereditary myopathies and sporadic inclusion body myositis — A cross sectional survey ☆

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ARTICLE INFO

*Article history:*
Received 20 December 2013
Accepted 20 December 2013
Available online 11 February 2014

*Keywords:*
Fracture
Calcium
25-Hydroxyvitamin D
Osteoporosis
Limb girdle muscular dystrophy
Myopathy

ABSTRACT

*Background*: The risk of osteoporosis is known in myopathies requiring long-term steroid treatment and Pompe disease, but not in other hereditary myopathies or sporadic inclusion body myositis (sIBM).

*Methods*: Risk factors of osteoporosis, laboratory parameters of bone metabolism, frequency of falls and fractures, walking ability, and pain were surveyed using questionnaires in 89 patients with sIBM and genetically confirmed myopathies facioscapulohumeral muscular dystrophy (FSHD), myotonic dystrophy types 1 and 2 (DM1, DM2), limb girdle muscular dystrophies (LGMD2A, LGMD2B, LGMD2I), *MATR3* myopathy, and oculopharyngeal muscular dystrophy (OPMD). Additionally laboratory parameters of bone metabolism were determined.

*Results*: The mean age at examination per disease group ranged from 32 years in LGMD2A to 70 years in sIBM. Myopathies with a higher degree of walking impairment had a higher risk of falls (sIBM, LGMD2A, LGMD2B). At the time of examination 3.4% had a history of osteoporosis. The 25-OH D3 level was decreased in 20% of patients (and in 55% of patients with LGMDs), 57% of them were ambulatory. The 25-OH D3 level was significantly lower in patients with myopathies than in other neurological disorders ($p < 0.001$). 2.7 falls per year per person occurred. Fractures were reported in 6.8% of patients within the last year. They involved frequently the tibia bone. The pain score didn't correlate with either the walking disability (WGMS) score or the 25-OH D3 level.



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2214-4269/$ – see front matter 
http://dx.doi.org/10.1016/j.ymgmr.2013.12.005

*Conclusion*: The risk for osteoporosis and reduced 25-OH D3 level seems to be increased in wheelchair-bound patients with myopathy but also in patients with DM1 and autosomal-recessive myopathies.


## 1. Introduction

Osteoporosis has been described in the past decade in several chronic neurological conditions, most of them associated with severe walking impairment [1–11]. A reduced bone density increases the risk of fractures and is associated with pain; hence this might contribute to an increased comorbidity in patients who also have neuromuscular disorders. Established risk factors for low bone mineral density are increased age, low body mass index (BMI), post-menopause in women, current smoking, chronic corticosteroid use, history of prior fractures and falls [12,13]. In men, hypogonadism and excessive alcohol intake further contribute to an increased risk for osteoporosis [12]. The diagnosis of osteoporosis is confirmed by bone densitometry using dual-energy X-ray absorptiometry (DXA) technology [14,15].

The frequency, complications and management of reduced bone mineral density have been extensively described in boys with Duchenne muscular dystrophy and both juvenile and adult patients with dermatomyositis and polymyositis [1–6]. In these conditions long-term steroid treatment regimens and wheelchair dependence were main predisposing factors. Recently, bone mineral density was assessed in children and adults with Pompe disease, an inherited metabolic myopathy. Most of them were receiving enzyme replacement therapy. In these studies osteoporosis was more frequent in patients who were wheelchair-bound, but was also observed in ambulant patients. There was also a correlation between proximal muscle strength and total body bone mineral density [9–11]. However, there is no data about either the frequency or the risk of decreased bone mineral density in other hereditary myopathies and sporadic inclusion body myositis (sIBM).

The objectives of this study were to systematically analyze risk factors for decreased bone mineral density, the frequency of falls and fractures, and to correlate them with pain, walking ability, and markers of bone metabolism in different hereditary myopathies and sIBM.

## 2. Patients and methods

### 2.1. Patients

The study was performed at the Neuromuscular Clinic at the Department of Neurology at Martin-Luther-University Halle-Wittenberg, Germany. The Ethics Committee of Martin-Luther-University Halle-Wittenberg approved the study protocol. Written informed consent was obtained from all patients. Adult patients with a genetically confirmed myopathy or histopathologically-defined sporadic inclusion body myositis [16] were enrolled in the one-time survey between January 2011 and March 2013. Patients with genetically confirmed Pompe disease were excluded from the analysis; their data will be presented elsewhere.

### 2.2. Questionnaires

A questionnaire was designed to capture the onset of the myopathy (paresis as the initial symptom), the frequency of falls during the last twelve months, the history of fractures and osteoporosis, and factors influencing the occurrence of osteoporosis (gender, age, body mass index (BMI), cigarette smoking, use of steroids and other medication, onset of menopause) (Supplemental Fig. 1).

The short form of the Brief Pain Inventory (BPI) is a self-administered 9-item questionnaire and was used to assess the presence and severity of current pain (pain within the previous 24 h) and its interference with daily activities. Four specific questions asked patients to rate the worst, least, and average pain experienced in the previous 24 h, and also to rate current pain. Patients were asked to rate pain on a scale from 0 (no pain) to 10 (pain as bad as you can imagine). The average of these 4 questions