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The Dutch patients' perspective on oculopharyngeal muscular dystrophy: A questionnaire study on fatigue, pain and impairments

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Abstract

Research on oculopharyngeal muscular dystrophy focuses mainly on genetic and pathophysiological aspects. Clinically, oculopharyngeal muscular dystrophy is often considered as a disease with a relatively mild initial disease course with no or only mild functional disabilities. However the occurrence of fatigue, pain and functional impairments other than dysphagia has never been studied systematically. The aim of this study is therefore to assess the prevalence of fatigue, pain, and functional limitations, and the social participation and psychological well-being of oculopharyngeal muscular dystrophy patients.

We performed a questionnaire study on fatigue, pain, functional impairments, social participation and psychological distress in 35 genetically confirmed oculopharyngeal muscular dystrophy patients with an average disease duration of 11.6 years.

We showed that 19 (54%) of the patients experienced severe fatigue and also 19 (54%) experienced pain. Limitations in daily life activities and social participation were detected in 33 (94%) of the patients. Many patients reported pelvic girdle weakness and limitations in ambulation. Fatigue severity was related to functional impairments, while pain and disease duration were not. Psychological distress was not different from healthy adults.

In conclusion, fatigue and pain are present among approximately half of the patients, and almost all patients are impaired in daily life activities, social participation and ambulation. These data should be taken into account in symptomatic management of oculopharyngeal muscular dystrophy.

Keywords: OPMD; Functional impairments; Pain; Fatigue

1. Introduction

Oculopharyngeal muscular dystrophy (OPMD) is a rare late-onset, usually autosomal dominantly inherited, muscle disorder. Most research on this myopathy focuses on genetic and pathophysiological aspects, while data on the patients' perspective are limited [1].

OPMD usually manifests in late adulthood, and many carriers of the OPMD mutation participate normally in society at the moment of diagnosis. The characteristic clinical pattern is the combination of bilateral ptosis, dysphagia and, later, limb girdle weakness [2]. Disease progression is usually slow and physical and functional impairments develop gradually. Consequently OPMD is considered to have a relatively mild course in the first years [3,4]. However, in 2001, Becher et al. described a Hispanic New Mexicans OPMD cohort and found that the phenotype was highly variable, ranging from physically active individuals with mild ptosis to patients of similar age with substantial dietary restrictions due to dysphagia and debilitating muscle weakness [5]. Also in our experience many OPMD patients become severely limited in daily-life activities and social participation in the course of their disease. Furthermore, OPMD patients in our cohort tend to underreport their limitations and some patients do not even visit a doctor. This is likely due to the slow progression, the fact that symptoms are considered part of normal ageing in their families, and the absence of a cure. This probably contributes to underestimation of the impact of the disease by physicians and researchers.

Over the recent years, a growing number of studies have investigated quality-of-life (QOL) aspects of various neuromuscular disorders (NMD). This shows that in facioscapulohumeral muscular dystrophy (FSHD), fatigue and pain occur frequently and impair daily life activities [6,7]. Furthermore, psychological distress is reported to impair daily

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life activities and social participation and to increase fatigue of patients suffering from various NMDs [6–9]. These studies did not include OPMD patients. Krause-Bachand reported the results of a qualitative pilot study (an unstructured interview format) of ten OPMD patients about their perspectives of living with ptosis, dysphagia, and a genetic disorder. All participants suffered both from physiological and psychosocial burdens related to living with OPMD. They formulated five comprehensive themes that described the essence of the experience: adjusting to change, managing misconception, seeking normality, facing the future, and informing children. They suggested that exploring QOL issues with larger groups of various ethnicity would provide additional data related to the experience of living with OPMD [10]. Further research on fatigue and pain in OPMD and on the psychological well-being of patients is limited to some anecdotal clinical observations. One case-report described an OPMD patient with proximal weakness and exercise-induced pain [11]. Another report described 22 Mexican OPMD patients, four of which had non-specific fatigue [12].

The principal aim of this study is therefore to assess the prevalence of fatigue and pain, the limitations in daily life activities and social participation, and the psychological well-being of OPMD patients using an open questionnaire with spontaneously reported complaints and a multidimensional assessment method [7,13]. Secondly, we determined whether limitations in daily life activities and social participation were related to fatigue severity, pain and disease duration. This study is performed in a Dutch cohort of OPMD patients, with relatively large mutation sizes. The average disease duration was 11.6 years.

2. Methods

The study was approved by the local ethical committee (CMO Regio Arnhem-Nijmegen) and written informed consent was obtained from all participating patients.

2.1. Participants

We used a cross sectional design. All genetically confirmed OPMD patients, known at the Dutch OPMD referral centre (Neuromuscular Centre Nijmegen), were invited to participate in this study. This is estimated to be approximately half of the Dutch OPMD population. Exclusion criteria were the presence of disabling systemic or psychiatric disease.

2.2. Assessment

The subjects received questions on general and specific OPMD symptoms and five validated questionnaires: Sickness Impact Profile (SIP-136), Checklist Individual Strength (CIS), The McGill's Pain Questionnaire (MPQ), Symptom Checklist 90 (SCL90) and Beck's Depression Inventory of Primary Care (BDI-PC).

The following demographical characteristics were collected: current age, age at onset of the symptoms, disease duration (from the onset of the symptoms), marital status and educational level. Furthermore, patients were asked to report all the complaints they attributed to OPMD ("spontaneously reported complaints").

2.2.1. Fatigue

To investigate the presence and severity of experienced fatigue, which is defined as an overwhelming sense of tiredness, lack of energy and feeling of exhaustion, we used the subscale fatigue severity of the CIS [14,15]. The subscale fatigue severity consists of 8 items (score range 8–56). A CIS-fatigue severity score equal to or higher than 35, being two standard deviations above the mean of healthy controls, indicates severe fatigue [16].

2.2.2. Pain

MPQ was used for the assessment of pain intensity, distribution of pain and use of analgesic drugs [17]. Patients indicate pain intensity on a visual analogue scale (VAS) for current pain, a minimal pain score and a maximal pain score; a VAS score of 0 mm indicates no pain, and 100 mm as the worst possible pain. The MPQ includes a whole body outline to indicate the distribution of the pain, and an inventory of the use of analgesic drugs.

2.2.3. Functional impairments in daily life

To evaluate the functional impairments we used the Dutch version of the SIP-136 [18]. This questionnaire consists of 136 items measuring functional impairments in 12 different domains of functioning: sleep and rest, emotional behavior, body care and movement, household management, mobility, social interaction, ambulation, alertness and intellectual functioning, communication, work limitations, recreation and pastimes, and eating. A total weighted SIP score is calculated and higher scores indicate more impairment.

2.2.4. Psychological well-being

To evaluate the psychological well-being of the patients we used the SCL-90 and BDI-PC [19,20]. The SCL-90 consists of 90-items. The total score ranges from 90 to 450. Low total scores reflect good psychological well-being, and high scores indicate more distress. The BDI-PC is a 7-item questionnaire assessing cognitive and affective symptoms of depression. Each item is scored on a 4-point scale (0–3). A total score equal to or higher than four indicates the presence of clinically relevant depressive symptoms. We used the BDI-PC, instead of the full BDI, to prevent an overlap in disease related somatic symptoms and the somatic symptoms of depression [20].

2.2.5. Statistical analysis

Data analysis was carried out using SPSS (version 16) for Windows. Descriptive statistics were used to describe the cohort and the scores on the different measures. Correlation between disease duration and age and fatigue severity (CIS-fatigue) and pain severity (VAS of current pain, maximal pain and minimal pain) was calculated by Pearson correlation coefficient. Multiple regression analysis was performed, method enter, with SIP total score as dependent variable and CIS-fatigue severity, VAS pain score and disease-duration as possible predictors. Statistical significance in all analyses was assumed at $p < 0.05$. Difference in prevalence and severity of fatigue and pain between the age groups (< or ≥60 years of age) and disease duration groups (< or ≥10 years disease duration)