Quality of Life in Adult Patients with Mitochondrial Myopathy

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Mitochondrial myopathies (MM) are caused by impairment of the respiratory chain [1]. They are some of the most common inherited neuromuscular disorders, with an estimated prevalence of >1/10,000 [2]. Uncomplicated MM (without major multisystem involvement) do usually not shorten the duration of life but rather involve the quality of life. A recent study has shown that mothers of children with mitochondrial disease had a significantly higher caregiver burden and poorer quality of life, particularly related to role limitations, vitality and mental health, compared with a group of mothers of children with intractable epilepsy and without mitochondrial disorder [3]. However, surprisingly, any studies addressing the issue of the quality of life in adult patients with MM are not available to date. The objective of this study was to evaluate if there was a relationship between quality of life and disease progression in MM.

We studied a group of 26 patients (18 females, 8 males; mean age 54.4 ± 11.9 years; 8 with mitochondrial DNA single deletion, 16 with multiple deletions) with pure MM [4] and without prominent comorbidities, by a scale of quality of life (SF-36) [5], a specific clinical score for mitochondrial disorders (Newcastle Mitochondrial Disease Adult Scale, NMDAS) [6] and a scale of muscle strength (Medical Research Council Scale, MRC) performed on previously reported muscular groups [7]. These evaluations were performed with the patients’ consent and permitted by the local Ethical Committee in a frame of wider protocols (2380/2007, 2473/2008). Data were expressed as means ± standard deviation. Analysis of data was carried out using MedCalc® version 7.3.0.1. The data were analyzed by Spearman’s coefficient of rank correlation (ρ). A p value < 0.05 was considered as significant.

The SF-36 quality of life score was inversely correlated with the NMDAS score, with a highly significant p value of 0.0012 (ρ = –0.649; 95% confidence interval for ρ = –0.828 to –0.349). Therefore, MM patients with higher disease progression scores had a worse quality of life (fig. 1a). Furthermore, the SF-36 directly correlated with the MRC scale (ρ = 0.003; ρ = 0.594, 95% confidence interval = 0.269–0.798); thus, MM patients with more preserved muscle strength had higher scores of quality of life (fig. 1b).

This study supports a role for the SF-36 scale in the evaluation of the quality of life in adult patients with MM (i.e. in clinical trials) and conversely reinforces the utility of the commonly used clinical scales MRC and NMDAS. We are aware of some possible limitations of our study, such as the apparently small number of patients. However, mitochondrial disorders are rare, and we included in the study only patients with uncomplicated MM. Large multicenter studies are strongly needed to better characterize the natural history of these diseases and to evaluate the quality of life in the affected subjects, in order to identify some countermeasures (i.e. pharmacological, physical or others) capable of benefiting patients with these chronic, still incurable disorders.
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Disclosure Statement
The authors declare no conflicts of interests.

References