Quality of Life (QOL) Outcomes in Patients with Duchenne Muscular Dystrophy

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Introduction

Duchenne muscular dystrophy (DMD) is a fatal, X-linked recessive disorder associated with a lack of dystrophin in skeletal, cardiac and smooth muscles. Clinically, patients develop progressive motor and cardiopulmonary dysfunction with loss of ambulation in the teen years and death in the twenties. Long-term glucocorticoid treatment slows disease progression; however, there are no curative treatments. Health-related quality of life (HRQOL) represents patients' perceptions of the impact of an illness and its treatment on their own functioning and well-being. The PedsQL™ 4.0 Generic Core Scales (GC QOL) with subcategories of Physical, Emotional, Social and School QOL, and PedsQL™ 3.0 Neuromuscular module (NM QOL) have been used as Patient Reported Outcomes in the Comprehensive Neuromuscular Center (CNC) to monitor HRQOL in patients with DMD over time.

Hypothesis

We hypothesized that the GC QOL and the NM QOL are biased towards physical function with scores decreasing with disease progression.

Methods

We performed an IRB-approved retrospective review of the electronic health records of DMD patients at the CNC with visits occurring between January 2012 and April 2017. Data pertaining to HRQOL, motor function (Functional Mobility Score/FMS), cardiac function (Left Ventricular Ejection Fraction/LVEF) and pulmonary function (Forced Vital Capacity Percent Predicted/FVC%) were collected. We examined the effects of FMS, LVEF, and FVC on QOL scores over time.

Results

The GC QOL and the NM QOL decreased with declining motor function for the ambulatory patients ($p<0.0001$), but not for the non-ambulatory patients ($p>0.12$). NM QOL and Emotional QOL decreased with decreasing FVC% ($p<0.0001$). Cardiac function had no effect on any QOL measures. Social QOL and School QOL are not affected by worsening physical, pulmonary or cardiac function in patients with DMD. Apart from emotional QOL ($p=0.2$), patient reported QOL scores were significantly higher than parent reported QOL scores ($p<0.0001$).

Conclusions

Declining motor function adversely affects GC QOL and NM QOL in ambulatory patients with DMD. Declining pulmonary function adversely affects NM QOL and Emotional QOL. In general, patient reported QOL scores were higher than parent reported scores.

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