Upper extremity outcome measures for collagen VI-related myopathy and LAMA2-related muscular dystrophy.


Abstract
Congenital muscular dystrophy (CMD) comprises a rare group of genetic muscle diseases that present at birth or early during infancy. Two common subtypes of CMD are collagen VI-related muscular dystrophy (COL6-RD) and laminin alpha 2-related dystrophy (LAMA2-RD). Traditional outcome measures in CMD include gross motor and mobility assessments, yet significant motor declines underscore the need for valid upper extremity motor assessments as a clinical endpoint. This study validated a battery of upper extremity measures in these two CMD subtypes for future clinical trials. For this cross-sectional study, 42 participants were assessed over the same 2-5 day period at the National Institutes of Health Clinical Center. All upper extremity measures were correlated with the Motor Function Measure 32 (MFM32). The battery of upper extremity assessments included the Jebsen Taylor Hand Function Test, Quality of Upper Extremity Skills Test (QUEST), hand held dynamometry, goniometry, and MyoSet Tools. Spearman Rho was used for correlations to the MFM32. Pearson was performed to correlate the Jebsen, QUEST, hand-held dynamometry, goniometry and the MyoSet Tools. Correlations were considered significant at the 0.01 level (2-tailed). Significant correlations were found between both the MFM32 and MFM Dimension 3 only (Distal Motor function) and the Jebsen, QUEST, MyoGrip and MyoPinch, elbow flexion/extension ROM and myometry. Additional correlations between the assessments are reported. The Jebsen, the Grasp and Dissociated Movements domains of the QUEST, the MyoGrip and the MyoPinch tools, as well as elbow ROM and myometry were determined to be valid and feasible in this population, provided variation in test items, and assessed a range of difficulty in CMD. To move forward, it will be of utmost importance to determine whether these upper extremity measures are reproducible and sensitive to change over time.

Copyright © 2016 Elsevier B.V. All rights reserved.
KEYWORDS: Collagen VI-related muscular dystrophy; Congenital muscular dystrophy; Laminin alpha 2-related dystrophy; QUEST, Jebsen, Motor Function Measure; Upper extremity measures

PMID: 28087121 DOI: 10.1016/j.nmd.2016.11.017

[PubMed - as supplied by publisher]