Longitudinal changes in clinical outcome measures in COL6-related dystrophies and LAMA2-related dystrophies.


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OBJECTIVE: To identify the rate of change of clinical outcome measures in children with 2 types of congenital muscular dystrophy (CMD), COL6-related dystrophies (COL6-RDs) and LAMA2-related dystrophies (LAMA2-RDs).

METHODS: Over the course of 4 years, 47 individuals (23 with COL6-RD and 24 with LAMA2-RD) 4 to 22 years of age were evaluated. Assessments included the Motor Function Measure 32 (MFM32), myometry (knee flexors and extensors, elbow flexors and extensors), goniometry (knee and elbow extension), pulmonary function tests, and quality-of-life measures. Separate linear mixed-effects models were fitted for each outcome measurement, with subject-specific random intercepts.

RESULTS: Total MFM32 scores for COL6-RDs and LAMA2-RDs decreased at a rate of 4.01 and 2.60 points, respectively, each year ($p < 0.01$). All muscle groups except elbow flexors for individuals with COL6-RDs decreased in strength between 1.70% ($p < 0.05$) and 2.55% ($p < 0.01$). Range-of-motion measurements decreased by 3.21° ($p < 0.05$) at the left elbow each year in individuals with LAMA2-RDs and 2.35° ($p < 0.01$) in right knee extension each year in individuals with COL6-RDs. Pulmonary function demonstrated a yearly decline in sitting forced vital capacity percent predicted of 3.03% ($p < 0.01$) in individuals with COL6-RDs. There was no significant change in quality-of-life measures analyzed.

CONCLUSION: Results of this study describe the rate of change of motor function as measured by the MFM32, muscle strength, range of motion, and pulmonary function in individuals with COL6-
RDs and LAMA2-RDs.


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