
MOTOR OUTCOME MEASURES IN PATIENTS WITH FKRP MUTATIONS, LONGITUDINAL FOLLOW-UP

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Abstract

Objective: To test the hypothesis that we will be able to detect change in motor outcome measures over time in a cohort with mutations in FKRP.

Methods: Individuals with documented FKRP mutations were evaluated annually with a battery of established motor outcome measures including limited quantitative myometry and timed function measures. Results were analyzed using random coefficient regression to determine annual change in each measure. Due to the non-linear progression through the lifespan of the study participants, pediatric (<19 years) and adult (>19 years) cohorts were analyzed separately. Effect of genotype was evaluated in each cohort.

Results: Sixty-nine participants (30 pediatric, 44 adult) with at least two evaluations were included. There was a small but statistically significant decline in timed motor function measures in both pediatric and adult cohorts. Genotype significantly affected rate of decline in the pediatric but not the adult cohort. Some pediatric patients who are homozygous for the c.826C>A mutation showed improving motor performance in adolescence. Performance on the 10-meter walk/run was highly correlated with other timed function tests.

Conclusions: There is a slow annual decline in motor function in adults with FKRP mutations that can be detected with standard motor outcome measures, while the results in the pediatric population were more variable and affected by genotype. Overall, these analyses provide a framework for development of future clinical trials. The DG natural history study may be found on clinicaltrials.gov (NCT00313677).

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