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Association of Initial Maximal Motor Ability with Long-term Functional Outcome in Patients with COL6-related Dystrophies

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Abstract

Objective: To accurately categorize the phenotypes of individuals with collagen VI-related dystrophies (COL6-RDs) during the first years of life in order to predict long-term motor function and pulmonary function, provide phenotype-specific anticipatory care and improve clinical trial readiness.

Methods: This retrospective, multicenter, international study analyzed the relationship of long-term motor and pulmonary function with the initial maximal motor ability achieved in individuals with COL6-RD.

Results: We studied 119 patients with COL6-RD from Spain (n = 54) and the US (n = 65). The early maximal motor milestones of (1.) ability to rise from the floor unassisted and (2.) the ability to climb four steps without holding onto the railing demonstrated reliability in distinguishing between three COL6-RD phenotypic subgroups: (1.) Ullrich congenital muscular dystrophy, (2.) Intermediate COL6-RD and (3.) Bethlem myopathy. Long-term motor function and pulmonary function are strongly correlated with the maximal motor ability achieved during the first years of life. Maximal motor capacity can predict other disease relevant events such as the age at loss of ambulation and the need for the initiation of nocturnal noninvasive ventilation.

Conclusion: This work proposes a prospective phenotypic classification for COL6-RDs which will

enable an accurate prediction of a patient's COL6-RD phenotype during the first years of life. The ability to establish a patient's COL6-RD phenotypic classification early will enable a more accurate prognosis of future motor and pulmonary function thus improving anticipatory clinical care, and it will be instrumental in aiding the design of future clinical trials by allowing for early stratification of trial cohorts.

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