SEPN1-related myopathies: clinical course in a large cohort of patients.


Abstract

OBJECTIVE: To assess the clinical course and genotype-phenotype correlations in patients with selenoprotein-related myopathy (SEPN1-RM) due to selenoprotein N1 gene (SEPN1) mutations for a retrospective cross-sectional study.

METHODS: Forty-one patients aged 1-60 years were included. Clinical data including scoliosis, respiratory function, and growth measurements were collected by case note review.

RESULTS: Mean age at onset was 2.7 years, ranging from birth to the second decade of life. All but 2 remained independently ambulant: one lost ambulation at age 5 years and another in his late 50s. The mean age of starting nocturnal noninvasive ventilation (NIV) was 13.9 years. One child required full-time NIV at the age of 1 year while in 2 cases NIV was started at 33 years. Two patients died from respiratory failure at the age of 10 and 22 years, respectively. The mean age at scoliosis onset was 10 years, in most cases preceded by rigidity of the spine. Fourteen patients had successful spinal surgery (mean age 13.9 years). Twenty-one were underweight; however, overt feeding difficulties were not a feature.

CONCLUSIONS: This study describes the largest population affected by SEPN1-RM reported so far. Our findings show that the spectrum of severity is wider than previously reported. Respiratory insufficiency generally develops by 14 years but may occur as early as in infancy or not until the fourth decade. Motor abilities remain essentially static over time even in patients with early presentation. Most adult patients remain ambulant and fully employed.

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