The phenotype and long-term follow-up in 11 patients with juvenile selenoprotein N1-related myopathy.


Author information

Abstract
The selenoprotein N1-related myopathies comprise rigid spine muscular dystrophy, the "classical" form of multiminicore disease, a desmin-related myopathy with Mallory body like inclusions and a form of congenital fiber-type disproportion. To define the phenotype and long-term clinical course in juvenile Selenoprotein N1-related myopathies 11 juvenile patients from eight families with SEPN1 mutations were assessed over a mean period of 7.2 years. Clinical findings, histomorphological studies, respiratory investigations and genetic data were analyzed: age of manifestation varied within the first 2 years of life with muscle hypotonia, lag of head control and delayed motor development. Further gross motor development was normal in 9/11 patients. All patients were ambulant for at least 1000 m at a mean age of 13.7 years. Eight patients exhibited a rigid spine diagnosed at a mean age of 10 years. All patients had respiratory impairment with a vital capacity ranging from 18% to 65%. Four patients were intermittently nocturnally ventilated at a mean age of 11 years. Body mass index was below 20 (kg/m(-2)) in all patients. Muscle biopsies of eight individuals revealed multiminicores (n=2), congenital fiber-type disproportion (n=1), myopathic changes with single cores (n=2) and unspecific myopathic features (n=3). Mutations were distributed throughout the entire SEPN1 gene. Although the phenotype of juvenile selenoprotein N1-related myopathies is homogenous regarding the main symptoms we describe a variable degree of clinical severity. Major complications were early respiratory failure, impaired increase in weight and orthopedic problems. There seems to be no correlation between skeletal muscle weakness and respiratory failure.

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