Steroid therapy in an alpha-dystroglycanopathy due to GMPPB gene mutations: A case report.

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Abstract

Alpha-dystroglycanopathies are a group of progressive and untreatable neuromuscular disorders, due to aberrant alpha-dystroglycan glycosylation. We describe the effects of a short-term cycle of corticosteroid therapy in a 9-year-old boy, affected by an alpha-dystroglycanopathy due to GMPPB gene mutations. The patient was affected by a congenital progressive muscular dystrophy since the first month of life, associated with psychomotor delay, seizures, and congenital bilateral cataracts. Despite physical therapy he had a progressive motor impairment. At the age of 9 years, he was treated with 0.75mg/kg/day of prednisone for 3 months and showed improvements in muscle strength and function scores and creatine kinase reduction. When steroid therapy was discontinued he showed again clinical and biochemical deterioration. These data suggest that corticosteroid may be considered as a treatment for patients with alpha-dystroglycanopathies due to GMPPB mutations.

KEYWORDS: Alpha-dystroglycanopathy; Congenital muscular dystrophy; Corticosteroid therapy; GMPPB

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