

Childhood onset autosomal dominant limb-girdle muscular dystrophy with cardiac conduction block

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摘要

Abstract

We report childhood-onset autosomal-dominant limb-girdle muscular dystrophy (LGMD) in a Chinese family with complete atrioventricular conduction block in the adult members. Six patients, including 5 men and 1 woman with an age of onset from 3 to 7 years, were affected. The grandfather had exercise intolerance since childhood and complete heart block with pace-maker placement at age 52. Three siblings had proximal muscle weakness and/or wasting since age 5 and heart block in their 40s. Two grandsons at the ages 7 and 3 showed exercise intolerance, and proximal muscle weakness and wasting. Sinus bradycardia was present in the elder grandson. Muscle enzymes were elevated in 3, particularly in childhood. Muscle biopsies from the proband showed myopathic changes with fatty degeneration, whorled fibers, and rimmed vacuoles. In adult patients, muscle magnetic resonance imaging scans disclosed atrophic changes and fatty degeneration in the gluteal, quadriceps, adductors, hamstrings, gastrocnemius, and soleus muscles, while in child probands the early atrophic changes were seen in the gluteal and hamstrings muscles. We conclude that this distinct family is characterized by childhood-onset autosomal-dominant LGMD with heart block and that prevention of sudden death in these patients is important.