Muscular Dystrophy, Congenital, 1B

Alternative Names
MDC1B
Congenital Muscular Dystrophy Merosin Negative

WHO International Classification of Diseases
Diseases of the nervous system

OMIM Number
604801

Mode of Inheritance
Autosomal recessive

Gene Map Locus
1q42

Description
Congenital muscular dystrophy is a heterogeneous group of inherited autosomal recessive disorders with onset at birth or in early infancy, characterized by severe hypotonia, markedly delayed motor development, generalized muscle atrophy with weakness of limb and trunk muscles leading to contractures and joint deformities. Congenital muscular dystrophy 1B represents a heterogeneous group of conditions characterized by proximal girdle weakness, generalized muscle hypertrophy, and rigidity of the spine and contractures of the tendo Achilles.

Molecular Genetics
In the past few years, there has been remarkable progress in the understanding of the genetic and biochemical basis of the muscular dystrophies. The majority of these conditions are caused by mutations in genes that encode sarcolemmal or extracellular-matrix proteins. Genome-wide analysis in affected individuals with congenital muscular dystrophy 1B showed linkage to the region on 1q42.

Epidemiology in the Arab World

United Arab Emirates
Muntoni et al. (1998) described a form of congenital muscular dystrophy characterized by proximal muscle weakness, muscle hypertrophy, and early respiratory failure in a consanguineous family from the United Arab Emirates. The pattern of inheritance was clearly autosomal recessive. The muscle hypertrophy was generalized, and there was rigidity of the spine and contractures of the Achilles tendons. Severe diaphragmatic involvement was responsible for the early respiratory failure. Intellect and the results of brain imaging were normal. Serum creatine kinase levels were grossly elevated, and muscle biopsy samples showed dystrophic changes. Affected individuals were demonstrated to have a deficiency of laminin alpha-2 (LAMA2) in muscle, but this appeared to be a secondary phenomenon, since linkage to the LAMA2 locus on 6q22-q23 was excluded. Brockington et al. (2000) performed genome-wide linkage analysis on the family of Muntoni et al. (1998) and found that the two affected children (one male and one female) showed an identical homozygous region on 1q42, spanning 6 to 15 cM. Brockington et al. (2000) updated the clinical information of the two sisters from the same family who died because of respiratory complications at the ages of 4 and 7 years.

References

Muntoni F, Taylor J, Sewry CA, Naom I, Dubowitz V. An early onset muscular dystrophy with diaphragmatic involvement, early respiratory failure and secondary alpha2

Contributors
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