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Update on the Genetics of Congenital Myopathies.

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Abstract

The congenital myopathies form a large clinically and genetically heterogeneous group of disorders. Currently mutations in at least 27 different genes have been reported to cause a congenital myopathy, but the number is expected to increase due to the accelerated use of next-generation sequencing methods. There is substantial overlap between the causative genes and the clinical and histopathologic features of the congenital myopathies. The mode of inheritance can be autosomal recessive, autosomal dominant or X-linked. Both dominant and recessive mutations in the same gene can cause a similar disease phenotype, and the same clinical phenotype can also be caused by mutations in different genes. Clear genotype-phenotype correlations are few and far between.

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