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Modeling and gene therapy of dysferlinopathy

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Abstract

Dysferlinopathies is a group of autosomal-recessive inherited neuromuscular diseases, which are characterized by defect in mRNA expression or in functionioning of dysferlin protein, appearing in about 1/200 000 births. Dysferlin is encoded by DYSF gene (Dystrophy-associated fer-1- like). It's disruption can cause various types of primary dysferlinopathies, which include Miyoshi myopathy (MM), Limb-girdle Muscular Dystrophy type 2B (LGMD2B) and distal myopathy with anterior tibial onset. Also, dysferlin deficiency can be associated with other diseases, such as caveolin- And calpainopathies. Here we discuss dysferlin protein structure and function, it's clinical phenotypes, known animal models and developing treatment strategies for dysferlinopathies. © Human stem cells institute, 2013.

Keywords

Distal myopathy with anterior tibial onset, Dysferlin, Dysferlinopathy, Limb girdle muscular dystrophy, Miyoshi myopathy