
Research progress of myosin heavy chain genes in human genetic diseases.

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Abstract

Myosins constitute a large superfamily proteins, which convert chemical energy, through ATP hydrolysis, to mechanical force for diverse cellular movements, such as cell migration and muscle contraction. The class II myosin forms the filaments in muscle and non-muscle cells as a hexameric protein complex, consisting of two myosin heavy chain (MyHC) subunits and two pairs of non-identical light chain subunits. There are several MyHC isoforms encoded by different genes of the MYH family in humans. At present, distinct mutations in different genes of the MYH family are associated with various human genetic diseases. Mutations in MYH2 are associated with skeletal myopathies, characterized by ophthalmoplegia. Mutations in MYH3 and MYH8 are associated with distal arthrogryposis syndromes. Mutations in MYH7 are associated with not only skeletal muscle diseases, such as Laing distal myopathy and myosin storage myopathy, but also hypertrophic cardiomyopathy. Mutations in MYH9 are associated with the so-called MYH9-related disease, characterized by giant platelets, thrombocytopenia and granulocyte inclusions. In this review, we briefly discuss the expression patterns of the MYH gene family and summarize the research progress in correlating the abnormalities of MYH gene family with various human genetic diseases.

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