Sarcomeric gene mutations in skeletal muscle diseases

The sarcomere is the fundamental unit of cardiac and skeletal muscle contraction. An important area that has been evolved over this period of time is a growing awareness of the etiology of skeletal and cardiac muscle diseases originating in the sarcomere. Many of the diseases of the sarcomere affect newborn children, i. e. are congenital myopathies. The skeletal muscle diseases vary in severity from paralysis at birth to relatively mild effects on life expectancy. Mutations in sarcomeric proteins are known to cause increasing number of different skeletal muscle diseases. The benefits of identifying the disease genes are correct diagnosis of the disease, accurate prognosis and targeted exploration of possible treatments for the diseases.

In this review, recent progress in the identification of sarcomeric gene defects in association with skeletal muscle disease will be focused. The genetics and the clinical manifestations of these disorders will be discussed.