Prevalence, pathological mechanisms, and genetic basis of limb-girdle muscular dystrophies: A review

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Limb-girdle muscular dystrophies (LGMDs) are a highly heterogeneous group of neuromuscular disorders that are associated with weakness and wasting of muscles in legs and arms. Signs and symptoms may begin at any age and usually worsen by time. LGMDs are autosomal disorders with different types and their prevalence is not the same in different areas. New technologies such as next-generation sequencing can accelerate their diagnosis. Several important pathological mechanisms that are involved in the pathology of the LGMD include abnormalities in dystrophin?glycoprotein complex, the sarcomere, glycosylation of dystroglycan, vesicle and molecular trafficking, signal transduction pathways, and nuclear functions. Here, we provide a comprehensive review that integrates LGMD clinical manifestations, prevalence, and some pathological mechanisms involved in LGMDs.

autosomal disorders

dystrophin

limb-girdle muscular dystrophy

pathophysiology

weakness

dystrophin

glycoprotein

clinical feature

disease classification

disease course

genetic analysis

glycosylation

human

limb girdle muscular dystrophy

pathogenesis

prevalence

priority journal

Review

sarcomere

signal transduction

cachexia

genetics

high throughput sequencing

limb girdle muscular dystrophy

metabolism

neuromuscular disease

pathology

skeletal muscle

Cachexia

High-Throughput Nucleotide Sequencing

Humans

Muscle, Skeletal

Muscular Dystrophies, Limb-Girdle

Neuromuscular Diseases

Sarcomeres