

Muscular dystrophies are a group of heritable muscle diseases that weaken the muscles of the body. They include disorders such as Duchenne muscular dystrophy, facioscapulohumeral muscular dystrophy, and limb girdle muscular dystrophy. The main symptoms involve progressive muscular weakness, muscle wasting, frequent falls, difficulty walking, muscle cramps and stiffness, limited range of motion, difficulty breathing, problems with swallowing, developmental delay or regression, and orthopedic problems, including scoliosis and contractures. Although muscular dystrophies predominantly affect skeletal muscle, they may affect other types of muscle (cardiac and smooth) or other body systems. They are caused by genetic defects in muscle proteins, and the death of muscle cells. The diagnosis is made by detecting elevations of elevated muscle enzymes in the blood (creatine phosphokinase, i.e. CPK, or aldolase), abnormalities typical of a muscle disease on EMG/nerve conduction testing, magnetic resonance imaging (MRI) of muscle, and findings on muscle biopsy that include evidence of damage to muscle fibers and a dystrophic pattern. Sometimes, but not always, a genetic abnormality can be found as well. Patterns of heritability include autosomal dominant, autosomal recessive, and X-linked, but sporadic cases occur and can have onset at any age. Recognition of the specific genetic defect allows doctors to counsel patients in regards to both the rate and pattern of disease progression and the likelihood of passing the disease on to their offspring. There is no cure for muscular dystrophies and treatment is often limited to supportive care.