

Adult and Adolescent Onset Muscular Dystrophies - Table 1

	Genetic Inheritance	Genetic/Protein Defect	Age of Onset	Clinical Features
Emery-Dreifuss Muscular Dystrophy (EDMD)	X-linked (most common) Some forms autosomal dominant or autosomal recessive	Loss of emerin protein from nuclear membrane, also loss of lamin A and lamin C	About 10 years old	Contractures (Achilles, elbow), Cardiac abnormalities
Facioscapulohumeral Muscular Dystrophy (FSHD)	Autosomal dominant	Reactivation of DUX4 gene, leading to aberrant production of DUX4 protein	5-10 years old (early onset) 15-30 years old (classic)	Scapular winging, kyphoscoliosis, sensorineural hearing loss, cardiac conduction abnormalities, retinal telangiectasias
Limb-Girdle Muscular Dystrophy (LGMD)	Varies, may be autosomal dominant or autosomal recessive	Varies; missing or defective proteins in the sarcolemma	8-15 years old (typically) but may span from 2-40 years old	Cardiomyopathy, respiratory involvement
Myotonic Dystrophy Type I (DM1)	Autosomal dominant	Abnormal trinucleotide repeat (CTG) in DMPK gene	20-40 years old	Myotonia, intellectual disabilities, cataracts, cardiac conduction abnormalities, dysphagia, metabolic comorbidities, pulmonary issues (nocturnal hypoventilation)
Myotonic Dystrophy Type II (DM2)	Autosomal dominant	Abnormal DNA expansion (CCTG) in ZNF9 gene	20-70 years old	Myotonia, weakness, cataracts, cardiac conduction abnormalities Intellectual disability and respiratory compromise less common