Muscular dystrophy

Muscular dystrophy (MD) is a group of muscle diseases that results in increasing weakening and breakdown of skeletal muscles over time.

The disorders differ in which muscles are primarily affected, the degree of weakness, how fast they worsen, and when symptoms begin.

Many people will eventually become unable to walk.

Some types are also associated with problems in other organs.

There are nine main categories of muscular dystrophy that contain more than thirty specific types.

The most common type is Duchenne muscular dystrophy (DMD) which typically affects males beginning around the age of four.

Other types include Becker muscular dystrophy, facioscapulohumeral muscular dystrophy, and myotonic dystrophy. They are due to mutations in genes that are involved in making muscle proteins.

This can occur due to either inheriting the defect from one's parents or the mutation occurring during early development.

Disorders may be X-linked recessive, autosomal recessive, or autosomal dominant.

Diagnosis often involves blood tests and genetic testing.

In two syndromic autism families, Ghasemi et al. discovered variants in two muscular dystrophy genes, DMD and CHKB. Given that DMD and CHKB are recognized for their participation in the non-cognitive manifestations of muscular dystrophy, it indicates that some genes transcend the boundary of apparently unrelated clinical categories, thereby establishing a novel connection between ASD and muscular dystrophy. The findings also shed light on the complex inheritance patterns observed in Iranian consanguineous simplex families and emphasize the connection between autism spectrum disorder and muscular dystrophy. This underscores a likely genetic convergence between neurodevelopmental and neuromuscular disorders [1)].

1)
Ghasemi MR, Sadeghi H, Hashemi-Gorji F, Mirfakhraie R, Gupta V, Ben-Mahmoud A, Bagheri S, Razjouyan K, Salehpour S, Tonekaboni SH, Dianatpour M, Omrani D, Jang MH, Layman LC, Miryounesi M, Kim HG. Exome sequencing reveals neurodevelopmental genes in simplex consanguineous Iranian families with syndromic autism. BMC Med Genomics. 2024 Aug 5;17(1):196. doi: 10.1186/s12920-024-01969-6. PMID: 39103847.