

key points

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Muscular dystrophy refers to a range of muscle diseases

caused by defects in muscle proteins, leading to death of the muscle cells, with loss of muscle tissue, and weakness. The development of clinical symptoms is usually gradual, and the earliest features may be difficult to identify and determine. With established disease the presence of muscle weakness and wasting is clear.

In children, the presentation may be delayed walking, or

poor performance in sporting activity. In children and adults presenting symptoms may include: difficulty raising from a squat; difficulty raising from a chair; difficulty lifting the arms above the head; poor balance; drooping eyelids; and joint contractures.

In the presence of slowly progressive muscle weakness

and wasting, an elevated serum creatine kinase would be a strong pointer to a muscle disease. Retention of limb reflexes would favour a myopathy over a neuropathy. The major differential diagnosis is an inflammatory myopathy, such as polymyositis.

The muscular dystrophies have a genetic basis. There

may be important genetic issues to discuss with the family, including the possibility of prenatal diagnosis. In Duchenne muscular dystrophy the inheritance is X-linked, with typically only boys affected. Many limb girdle muscular dystrophies are autosomal recessive, affecting only one generation of a family and facioscapulohumeral dystrophy is autosomal dominant.

Myotonic dystrophy is one of the more common

muscular dystrophies. Clinical examination typically shows slowness in relaxing a tight grip, together with the muscle wasting or dystrophy in limbs and facial muscles.

There is no curative therapy for the muscular dystrophies.

There may be involvement of cardiac muscle, cardiac conduction disturbance and arrhythmias sometimes requiring pacemaker insertion. Involvement of respiratory muscles may require respiratory support. Swallowing and feeding difficulty may require nutritional support, and possibly gastrostomy. Orthoses and orthopaedic management may also be needed.