DEFINITION

1. **Muscular dystrophy** has been defined as a myopathy associated with progressive muscle weakness, destruction and regeneration of muscle fibres and eventual replacement of muscle fibres with fibrous and fatty connective tissue, without involvement of the nervous system.

CLASSIFICATION

2. Classification of the muscular dystrophies is not merely academic but is the only safe guide to prognosis and genetic counselling.

3. The traditional clinico-anatomical classification has proved unsatisfactory, both clinically and genetically.

4. The more satisfactory clinico-genetic classification of pure muscular dystrophies derived from current knowledge and which is based upon the mode of inheritance, age of onset, rate of progression, distribution of the involved muscles and associated findings in muscle or other organs is as follows -

4.1. X-linked muscular dystrophies

4.1.1. Severe (Duchenne type)
4.1.2. Benign (Becker type)
4.1.3. Benign with early contractures (Emery-Dreifuss)
4.1.4. Scapuloperoneal (rare)

4.2. Autosomal recessive muscular dystrophies

4.2.1. Limb-girdle (scapulohumeral, pelvifemoral)
4.2.2. Childhood type
4.2.3. Congenital

4.3. Autosomal dominant muscular dystrophies

4.3.1 Facioscapulohumeral
4.3.2 Scapuloperoneal
4.3.3 Late-onset proximal
4.3.4 Distal
4.3.5 Ocular
4.3.6 Oculopharyngeal

5. Although this classification seems to be the most satisfactory at the present time, there are still some cases that are difficult to fit into any of the groups described.

CLINICAL MANIFESTATIONS

6. These depend upon which muscles are first involved by the disease process and upon the rate of progress of the disease. However, there are certain features common to all the dystrophies.

7. The essential feature is one of progressive weakness and wasting of the involved muscles, leading to slowness in walking, frequent falling, difficulty in climbing stairs or getting up from the floor when the pelvic girdle is involved. Eventually the patients develop an exaggerated lumbar lordosis and a waddling gait. Climbing up one's legs on rising from the floor (Gower's sign) is a characteristic feature of many of these conditions. When the shoulder girdle is involved, an unusually sloping appearance of the shoulders is produced along with winging of the scapulae and wasting of the pectoralis major muscle. Facial weakness causes an inability to whistle or close the eyes while involvement of the distal muscles leads to weakness of grip and foot drop. Other features are dysphagia and weakness of the external ocular muscles.

8. Contractures are common in all forms of muscular dystrophy in the late stages, but particularly in the severe Duchenne type.

AETIOLOGY

9. Males are more frequently affected than females, direct transmission by unaffected females is common and the types of inheritance differ in different families. Most muscular dystrophies begin in childhood although some are present at birth (congenital) or may present in adult life (distal). Some may present both in childhood and in adult life (facioscapulohumeral).

10. Although all forms of muscular dystrophy are genetically determined, the exact nature of the process causing the muscles to waste is unknown. No deficiency of a single enzyme or a specific abnormality of a muscle protein has been discovered. The most convincing evidence now available suggests that the primary abnormality may lie within the plasma membrane of the muscle cell, allowing uncontrolled entry of extra-cellular calcium ions, which in turn activate neutral proteases. These proteases then digest the contents of the muscle fibre.

11. The muscular dystrophies are progressive conditions unaffected by external factors or remedies. The rate of progress depends upon the type of muscular dystrophy.

CONCLUSION

12. **Muscular dystrophies** are progressive, genetically determined disorders of muscles which are unaffected by external factors. The clinical features and rate of progress depend upon the type of muscular dystrophy, but there may be some degree of overlap.

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