A Study on Duchenne Muscular Dystrophy (DMD)

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Abstract
Duchenne muscular dystrophy or Pseudo Hypertrophic Muscular Dystrophy is heterogenous group of inherited disorder C graddal degeneration of muscle fibres. It is Inheritance sex linked disease and boys are sufferer while girls are carrier. Patient dies in 2nd /3rd decade of life due to pulmoray infection and cardionyopahy.

Keywords: X linked disease, mutation, Grower’s Sing, Baker Muscular dystrophy, serum creatine kinase, EMG, muscle Biopsy.

Introduction
Duchenne Muscular Dystrophy (DMD) is X-linked inheritance disease, boys are sufferer while girls are carrier. Perintal history is normal. Early development of child is normal or delayed when child starts walking in 2nd years gait is usually slight abnormal hypertrophy of calf cusele may develop at age of 3rd years but patient may not notice till age of 4 to 5 years. Toddler has difficulty in climbing stairs. He takes of support of railing and wall. Distribution of atropic/hypertrophic are characteristic muscles. Calf muscle, glutie, pectoral, braclio radialis and tongue are large. All muscles get atrophy and child becomes bed ridden. Kyposcoliosis is other complication of disease. Diagnosis is suspect especially if there is family history of myopathy.

Incidence
Most common muscular dystrophy is duchenne muscular dystrophy (DMD) is X- linked recessive disorder. It occurs in 2 to 3 boys per 10,000 boys. It is due to mutation in hyertrophin gene. The Gene is expressed in all muscles and brain. This causes loss of muscle cells and progressive proximal to distal muscle weakness. Despite advance in medical management it continues to be fatal in 2nd and 3rd decade of life.

Backer-Muscluar Dystrophy (BMD) results from less extensive citations in the same gene and produces a partially funcational dystrophin protein. Clinically disease is less severe with a later onset and less profound weakness.
Diagnosis

1. Serum Creatine Phospho Kinase (CPK):
   Are elevated in most boy patients as well as in carrier females which is 200/300 times more than normal value even before clinical, manifestations becomes obvious and help in genetic counselling.

2. Electromyogram (EMG):
   Shows- decreased amplitude and duration of motor unit potential.

3. Electro Cardiogram (ECG):
   Shows Arrythmia.

4. Histopathology of muscle fibres:
   Shows diffuse changes of degeneration, variation in size and central nuclei of muscle fibres. Muscle atrophy secondary to neurological disease shows bundles of muscle fibres in the degeneration interspersed with bundles of normal muscle fibres.

Prognosis

By the age of 12 Years Ambulation caeses and by the age of 2nd / 3rd decade ptdies due to recurrent chest infection and heart failure due to associated cardiomyopathy.

Factor-Prolonging Muscles weakness

Prolonged immobilization like plaster caste after hastens weakness of muscles.

Management

☐ There is no definite and effective treatment.
☐ Prolong Immobilization should be avoided.
☐ Physiotherapy/Exercise to be done.
☐ Kyphoscoliosis to be prevented by using tight brdces.
☐ Respiratory complication showed be prevented by vigorous antibiotic therapy.
☐ Cardioryopathy and congestive cardiae failure are managed by conventional approaches.
☐ Emotional support to be given to patient and family.
☐ Progrosis should be explained.
☐ Corticosteroid is main drug of D.M.D.
☐ Gene therapy

Differential Diagnosis

1. Myelopathies
   In myelopathies, sensory changes and distribution of weakness is variable. Deep tendon reflexes are generally brisk. Abdominal are often absent. EMG shows neurogenic pattern, peripheral N. Conduction delayed in late stage. Muscle-Biopsy shows neurogenic type of atrophy. CSF protene may be elevated, muscle enzymes like, CPK, Aldolase and SGOT are within normal limit.

2. Polyneuritis
   Sensory changes are usually present. There may be paralgesia or hyperalgesia. Distribution of weakness is distal and symmetrical except in case of Infective Polyneuritis (in which proximal weakness may be there):
   ● Tendon reflexes are absent.
   ● Abdominal reflexes are present.
   ● Sphincters are usually not involved.
   ● EMG and muscle Biopsy are of Neurogenic type.
   ● CSF proteins is elevated in gullain Barre syndrome.
   ● Muscle enzymes are normal.

3. Backer Type
   It is X linked inheritance, age of onset between 5 and 25 years. Becker muscular dystrophy result from less extensive mutations in same gen and produces a partially functional dystrophin protein. Clinical disease less sever with later onset and less profound weakness.

4. Fascio scapulo-Humoral
   Both sexes in several gene rations are affected. Age of onset around puberty.

5. Limba girdle (Erbtype)
   Autosomal recessive inheritance. Age of onset is around 10 years.

6. Congenital myopathies:
   Rare inherited dis-order.
**Case History**

Patient about 12 years male resident of Distt. – Basti came to Govt. Medical College Basti in ortho OPD. Patient came in our OPD walking with support and his gate was waddling gait. Patient was examined and his calf muscles were enlarged in size B/L. Local temperature was normal, skin/muscle was non tender and soft to firm. His spine was normal. On standing from ground he used his both hands on both knees to take support. Patient having one sister who is 6 years old. She is normal in each aspect. Patient had normal delivery at hospital without any problem/complication. Patient having no history of fever. His serum creatine kinase is 7111. EMG showed decreased amplitude. Histopathology could not be done as consent was not given by his parents. Patient has one sister 6 years old without any problem her serum creatine kinase was 3212. Her gait was normal.

**Analysis**

Patient was 12 years boy and diagnosed as DMD/Muscular Hypertrophy. He was unable to stand self but can with support and had waddling gait. His both calf muscle was abnormally enlarged without other finding. Spine was normal. SCPK was raised to 300 times more than normal. Patient had one sister 6 years old without any problem though her SCPK was also raised in same proportion as brother (patient). EMG also showed abnormal changes. Histopathology of calf muscle also important diagnostic parameter in this case. But parents refused to due to injury/trama during biopsy procedure. Progress of disease was explained to the parents of patient as at the age of 20-30 years. Life can be fatal due to chest infection and heart failure due to cardiomyopathy. DMD boys are sufferer and girls are carries.

This disease is X-linked disease and boys are sufferers while girls are carrier. Diagnosis and investigation are mandatory. Physical exercise and corticosteroids are main therapy of DMD patients.

**Conclusion**

Ducinne Muscular Dystrophy is X-linked inheritance disease, boys are sufferers while girls are carrier, Though this disease appear at the age of 20-30 years. Patient dies due to chest infection and heart failure due to cardiomyopathy. Psysiotherapy corticosteroid and reassurance are main treatment.

**References**

3. Ad Hoc Committee on Mutation Nomenclature (1996). Update on


