CASE REPORT

DUCHENNE MUSCULAR DYSTROPHY IN THREE MALE SIBLINGS: A RARE CASE REPORT

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ABSTRACT

This case report describes three male siblings, 7, 9, and 11 years old presented with with inability to stand and walk even with support. There was no family history of such a problem, and no consanguineous marriage of their parents.

Keywords: *Myopathy; Muscular dystrophy, Gower's sign*

INTRODUCTION:

Three male siblings 7,9, and 11 years old, presented to our orthopedic department with the inability to stand and walk even with support. According to history, weakness started gradually between 4 to 6 years of age in each boy, involving the lower limbs first and progressing in severity over time, and then involved the upper limbs later on. There was no family history of such a problem, and no consanguineous marriage of their parents. This uneducated family with poor socio-economic status was consulting religious leaders for the treatment of their children and was unaware of the diagnosis. The intelligence status of the boys was normal, and they did not give a history of any muscular pains.

Positive Gower's sign, along with calf muscle hypertrophy, was noted on physical examination. Requested serological analysis revealed CK (creatine kinase) levels elevated (9142 IU/L, 8951U/L, and 7836 IU/L) to many folds.

Electromyographic findings were abnormal in all three children, supporting the diagnosis of myopathy. Diagnosis of myopathy was established on the basis of the history, examination, elevated CK levels, and electromyography. Parents were counselled about the disease. Corticosteroid therapy, physiotherapy, and regular follow-up for respiratory problems use of intermittent positive pressure ventilation, were advised.

Fig. Three male siblings of a family affected with DMD (Permission granted by the father)

DISCUSSION:

DMD (Duchenne muscular dystrophy) was first described in the 1860s by Guillaume Benjamin Amand Duchenne, a French neurologist. It is an X-X-linked progressive myopathy with recessive transmission and affecting 1 in 3500 male births.[1,2]

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The failure of the dystrophin gene coding due to a defect in the X chromosome results in loss of dystrophin protein synthesis & function, which leads to destruction of muscle fibers and replacement by fibrofaty tissue.[3] DMD patients may have 200-300-fold high levels of serum creatine kinase (Normal 35–174 U/L).[4]

The condition remains silent until the child is around 3 years old when he comes across with difficulty in standing and stair climbing. He also has problems with running and falls frequently.

The patient usually becomes wheelchair bound around

10 years of age with loss of walking ability.

Then the disease progresses rapidly with the development of scoliosis and lung function deterioration. Generally, patient dies around 30 years of age with cardiopulmonary failure.4

In our study, older, middle, and younger siblings developed motor weakness symptoms between 4-6 years, leading to difficulty in standing from a sitting position, stair climbing, and walking. My study children are wheelchair bound with a greater risk of contracture formation. Physiotherapy is advised to prevent these contractures and scoliosis.

Stem cell and gene therapy are considered future hope for treatment of muscular dystrophy, but these are still under investigation.

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