CASE REPORT

Duchenne Muscular Dystrophy : Case Reports of Siblings

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Introduction :

Muscular dystrophy weakens the muscles that help the body move. People with muscular dystrophy have incorrect or missing information in their genes, which prevents them from making the proteins they need for healthy muscles. Because muscular dystrophy is genetic, people are born with the problem - it's not contagious and can gradually lose the ability to do the things most people take for granted, like walking or sitting up. Someone with muscular dystrophy might start having muscle problems as a baby or their symptoms might start later. Some people even develop muscular dystrophy as adults1. Duchenne Muscular Dystrophy is named after the French neurologist Guillaume Benjamin Amand Duchenne who, in the 1861 described and detailed the case of a boy who had this condition[1]. In 1868 he gave an account of 13 other affected children. Duchenne was the first who did a biopsy to obtain tissue from a living patient for microscopic examination. Duchenne muscular dystrophy (DMD), the most common type of the disease, is caused by a problem with the gene that makes a protein called *dystrophin*. This protein helps muscle cells keep their shape and strength. Without it, muscles break down and a person gradually becomes weaker. DMD affects boys. Symptoms usually start between ages 2 and 6. By age 10 or 12, kids with DMD often need to use a wheelchair. The heart may also be affected, and people with DMD need to be followed closely by a lung and heart specialist. They can also develop scoliosis (curvature of the spine) and tightness in their joints. Over time, even the muscles that control breathing get weaker, and a person might need a ventilator to breathe.

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Case Report-1 :

A 15 year old boy named Atiqur Rahman, from Muradnagar, Comilla got admitted in Holy Family Red Crescent Medical Hospital in January 2013. According to the informant mother he was quite well 7 years back. Then

On examination, the boy was alert and cooperative. His built was average with 40 kg of weight.

In motor System, Gower's sign was positive in gait, power was diminished in lower limb but normal in upper limb, power was 1/6, he developed weakness in walking and going upstairs and he used to fall during running, later he developed difficulty in rising from lying position for 5 years (Fig-1). He was born by an uneventful pregnancy and no resuscitation was required after birth. His younger sister had also suffered similar illness. muscle tone was mildly diminished but sensory nerves were intact.

Laboratory investigations showed S.CPK was 4454 U/L, WBC- 8,200 / cumm, Neutrophil-85.7%, Lymphocyte - 34.5%, RBC - 4.83 mmol/L, Platelet - 3,60,000 / cumm, RBS : 5.4 mmol/L. Serum electrolytes were Na+ - 130.0 mmol/L, K+- 4.0 mmol/L, Cl- - 91.0 mmol/L.

Figure-1: Difficulty in rising from lying position

Muscle biopsy revealed the muscle fibres were different in size and shape showing retrograde change. Some of the muscle fibres also showed interior migration of nuclei. These findings were suggestive of muscular dystrophy.

Case report- 2 :

Maria Akhter Mim of 13 years of age from Muradnagar, Comilla born of a consanguineous marriage through an uneventful pregnancy of normal body build and nutrition, fully immunized came with complaints of difficulty in walking, tendency to fall while running there was motor delay (Fig-2). On examination she was alert cooperative there was hypertrophy of calves with mild hypotonia with reduced power 2/6, and there was lumber lordosis and Gower's signs was present. Her elder brother also had similar problem for more than five years.

Figure-2 : Tendency to fall with motor delay

Muscle Biopsy : Macroscopic Description- Specimen received in formalin with proper label with patient's identification consists of an irregular grey brown piece of tissue measuring about 0.5 cm in maximum diameter. Microscopic description revealed skeletal muscle, in some areas the muscle fibres showed retrograde changes with individual fibres atrophy. Fatty infiltration within the muscle was seen. No malignancy was found. Muscle cuff left (biopsy) revealed features suggestive of myopathy.

Discussion :

Duchenne muscular dystrophy is caused by mutations in the dystrophin gene, which is located on the X chromosome. DMD has an incidence of 1 in 4,000 newborn males[2]. Mutations within the dystrophin gene can either be inherited or occur spontaneously during germline transmission which results in muscle degeneration and eventual death. The disorder is caused by a mutation in the dystrophin gene, located on the human X chromosome, which codes for the protein dystrophin, an important structural component within muscle tissue that provides structural stability to the dystroglycan complex (DGC) of the cell membrane. While both sexes can carry the mutation, females rarely exhibit signs of the disease. It becomes harder and harder for the boy to walk; his ability to walk usually completely disintegrates between the time the boy is 9 to 12 years of age. Most men affected with DMD become essentially "paralyzed from the neck down" by 21 years of age[3]. Muscle wasting begins in the legs a nd pelvis, then progresses to the muscles of the shoulders and neck, followed by loss of arm muscles and respiratory muscles. Calf muscle enlargement (pseudohypertrophy) is quite obvious.

Both the cases reported here as Duchenne muscular dystrophy (DMD) which is hard to find the as genetic trait among the siblings in same family and females rarely suffer from DMD rather than Limb Girdle Dystrophy.

There is no current cure for DMD, although some trials with exon-skipping treatment have halted decline and produced small clinical improvements in walking. Treatment is generally aimed at controlling the onset of symptoms to maximize the quality of life, and include Corticosteroids to increase energy and strength and defer severity of some symptoms[4]. Some trials have shown that beta 2 -agonists increase muscle strength but do not modify disease progression. Follow-up time for most beta 2 -agonists is only around 12 months and hence results cannot be extrapolated beyond that time frame. Mild, non-jarring physical activity such as swimming is encouraged. Inactivity (such as bed rest) can worsen the muscle disease. Physical therapy is helpful to maintain muscle strength, flexibility, and function. Orthopedic appliances (such as braces and wheelchairs) may improve mobility and the ability for self-care. Form-fitting removable leg braces that hold the ankle in place during sleep can defer the onset of contractures. Appropriate respiratory support as the disease progresses is also important.

Duchenne muscular dystrophy is a progressive disease which eventually affects all voluntary muscles and involves the heart and breathing muscles in later stages. The life expectancy is currently estimated to be around 25 years[5]. But this varies from patient to patient. Recent advancements in medicine are extending the lives of those afflicted. With high standards of medical care young men with Duchenne muscular dystrophy are often living well into their 30s. In rare cases, persons with DMD have been seen to survive into the forties or early fifties, with the use of proper positioning in wheelchairs and beds, ventilator support (via tracheostomy or mouthpiece), airway clearance, and heart medications, if required. Early planning of the required supports for later-life care has shown greater longevity in people living with DMD.

References :

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