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Case Study

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DUCHENE MUSCULAR DYSTROPHY DURING ITS COURSE DIMINISHES HOPES OF A FAMILY: A FAMILY CASE REPORT

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INTRODUCTION

Duchene muscular dystrophy, an X linked recessive disorder; is the most common and severe form of muscular dystrophy affecting males.[1] Patients usually become wheelchair dependent in early teens and die in their late teens to early twenties.[2] It is clinically characterized by proximal muscle weakness and calf hypertrophy because of failure to manufacture dystrophin. The purpose of this paper is to describe cases of 3 male siblings with DMD.[3]

CASE REPORT

A 7 year old male child was brought to a civil dispensary OPD with the symptoms of upper respiratory tract infection. On clinical examination a limp because of proximal muscle weakness was observed. The six member family had three boys diagnosed with DMD and a normal 5 year old female child. The eldest boy at his age of 6 year had first symptoms in the form of weakness and waddling gait. In the period of two years parents consulted several health institutions for his persistently increasing symptoms but the diagnosis of DMD was made finally at a tertiary care hospital. The second boy who was initially asymptomatic developed same symptoms of weakness of both lower limb and waddling gait by the time he reached 6 year age. No time was wasted by parents for the diagnosis of their second son and they directly consulted same tertiary care hospital. After the diagnosis of DMD of second child the parents stopped the treatment for both the children. However, on questioning parents said, *"Humara bahut kharcha ho gaya, Doctor ne bola is bimari ka koi illaz nahin hai. Teesre ladke ko bhi ho sakta hai, isliye humne aage koi illaz nahin karwaya"* (we have spent a lot. Doctor said that there is no cure for this disease. This can happen to third boy as well, that's why we didn't take any treatment further). The third son, youngest among three, at the age of 7 year also developed the symptoms. The parents had no consultation and treatment for him, as the parents accepted the truth that the youngest son also had DMD.

The doctor from civil dispensary during his field visit to their home examined all the three boys. The eldest boy (aged 11 years) was non ambulatory and fully dependent on his family for mobility. The middle one (aged 9 years) had difficulty in walking and climbing stairs. He had toes walking with frequent falls and inability to get up from floor. Whereas the youngest son (aged 7 years) had difficulty in walking, frequent falls and Gower sign positive.

Picture 1. Eldest son (11 years old), not able to stand and walks his own.

Picture 2. Second son (9 years old), attempts to stand but not able to stand his own but when assisted to stand walks with waddling gait.

Picture 3. Youngest son (7 years old), stands with positive Gover's sign and walks with waddling gait.

"Kuch saal me ye teeno chal bh nahin paenge, Doctors ne ye bhi kaha ye ki ye 15 ya 20 saal se jyada nahin jee paaenge". As described by the doctors to the parents about the outcome of the disease the parents had accepted the truth that all of their sons will not be able to walk one day and will die at late teenage. The parents followed hedonic adaptation thinking that we maintain a relatively stable level of happiness despite the positive or negative changes that take place in our lives.

DISCUSSION

DMD is the most common muscular dystrophy caused by mutation in the dystrophin gene on Xp21 chromosome because of which body is unable to synthesis dystrophin which is required for muscle contraction.[4] Affected boys clinically present with difficulty in walking and climbing stairs, toe walking, frequent falls and inability to get up from floor and eventually leading to death.[5] The diagnosis of DMD is made on the basis of clinical examination, high Creatinine kinase (CK) levels, muscle biopsy, electromyography and genetic analysis. There are 5 stages of DMD starting varying from pre symptomatic, early ambulatory, late ambulatory, early non ambulatory and late non ambulatory.[3] In the present case all the three siblings had the onset of symptoms at the age of 6-7 yrs and in the present situation the eldest one is in 4th stage of early non ambulatory, middle one in 3rd of late ambulatory and the youngest one in 2nd stage of early ambulatory.

There is no cure of DMD but management involves corticosteroid therapy and physiotherapy which prolongs the ambulation and quality of life can be improved. Children needs to be as active as possible, avoid spending too much time in sitting in front of television, computer of using electronic games. Counselling of the family members or the caregivers plays an important role. Prenatal counselling and genetic testing are being used as a hope in DMD to improve the quality of life.[6,7]

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