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A Boy with Duchenne Muscular Dystrophy Who Never Walked

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ABSTRACT

Background

Duchenne Muscular Dytrophy (DMD) is an X-linked genetic disease characterized bydelay in walking, progressive muscle weakness, and sometimes mental retardation. This condition is caused by deletion, duplication or point mutations in the dystrophin gene, and it is the most common form of muscular dystrophy . The gene is located at Xp21.2-p21.1. DMD gene consist of 79 codons and may result in clinical symptoms depending on where the deletion occurs but with a wide range of variations . We describe a 5-year-old boy with DMD who has never walked.

Case Report

This boy, the first child in the family, presented with delay in walking. There was no family history of muscle disease. He was delivered at term and was deemed hypotonic during follow-up. A diagnosis of DMD was made at the age of two, and two years later daily oral prednisolone treatment was initiated. He is followed by symptomatic treatments with physiotherapy. At the last follow-up he was 5 years and 6 months old with no effort to ambulate independently. In due course, he developed multiple joint contractures mainly in lower extremities.

Conclusions

No matter how large the mutation, almost every DMD boy will walk, albeit late. Our case had exons 3-60 hemizygously deleted and although he was 5 years and 6 months old, he had not taken a few steps. Although physical therapy and aerobic exercises had been applied from the time of diagnosis, joint contractures were not refrainable.

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KEYWORDS: Muscular Dystrophy, DMD, BMD, Neuromuscular Diseases

INTRODUCTION

Duchenne Muscular Dystrophy (DMD) is an X-linked genetic disease characterized by delay in walking, progressive muscle weakness, and sometimes mental retardation. This condition is caused by deletion, duplication or point mutations in the dystrophin gene, and it is the most common form of muscular dystrophy [1]. The gene is located at Xp21.2-p21.1. DMD gene consist of 79 codons and may result in clinical symptoms depending on where the deletion occurs but with a wide range of variations [2]. We describe a 5-year-old boy with DMD who has never walked.

CASE REPORT

This boy, the first child in the family, presented with delay in walking. There was no family history of muscle disease. He was delivered at term and was deemed hypotonic during follow-up. He was able to hold his neck at 9 months. When he was 16 months old, he could only sit without support. He never spoke in sentences. Genetic testing of the patient was performed and a hemizygous mutation were detected between exons 3 to 60. A diagnosis of DMD was made at the age of two, and two years later daily oral prednisolone treatment was initiated. He is followed by symptomatic treatments with physiotherapy. At the last follow-up he was 5 years and 6 months old with no effort to ambulate independently. In due course, he developed multiple joint contractures mainly in lower extremities.

DISCUSSİON

The DMD gene is the largest known gene in the human genome, containing 79 exons and at least 2.3 million base pairs. It is located on the X chromosome and encodes the dystrophin protein [2]. Mutations in the DMD gene can lead to a variety of different muscle disorders including severe and milder forms along with X-linked cardiomyopathy. Females can be carriers and sometimes symptomatic [3]. DMD is the classical form of which characterized by progressive muscle weakness and degeneration. By definition DMD boys become off feet by age 13. Losing walking before age 10 is called ‘severe’ DMD. Becker muscular dystrophy (BMD) is a milder

form with a later onset and slower progression, where boys continue to walk after age 16 [4].

CONCLUSIONS

No matter how large the mutation, almost every DMD boy will walk, albeit late. We previously reported a DMD boy who walked only for 9 months between ages 5 and 6, then stopped [5]. Our case had exons 3-60 hemizygously deleted and although he was 5 years and 6 months old, he had not taken a few steps. Although physical therapy and aerobic exercises had been applied from the time of diagnosis, joint contractures were not refrainable. This limitation and his age suggest that the patient will probably never be able to walk in the future. We think that this case may add new insights to our knowledge of the clinical spectrum of DMD.

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