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Case Report

Spinal muscular atrophy

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Abstract

Spinal muscular atrophy (SMA) is a genetic motor neuron disease characterized by progressive degeneration of motor neurons. Herein, a 12-year-old female child, born to healthy nonconsanguineous parents, was brought with the chief complaints of wasting and weakness of both lower limbs for 3 years. There was no family history of neurological illness. On examination, higher mental functions and cranial nerve examinations were normal. Typical worm-like fasciculations were seen in tongue and both lower limbs. Upper extremities were less affected. The child was able to feed herself. Respiratory muscles were not affected. A diagnosis of SMA-Type 3 (Kugelberg–Welander disease) was made on the basis of clinical presentation and subsequently, the diagnosis was genetically confirmed by molecular analysis of SMN gene. Electromyography showed spontaneous fibrillation at rest. Nerve conduction study was normal. No medical treatment was able to delay the progression. Supportive therapy includes orthopedic care and mild physiotherapy.

Key words: Fasciculation, Kugelberg–Welander disease, spinal muscular atrophy

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INTRODUCTION

Spinal muscular atrophy (SMA) is an autosomal recessive degenerative disease of motor neurons. It usually starts in fetal life and continues to be progressive in infancy and childhood. SMA causes weakness and wasting of the voluntary muscles. Weakness is often more severe in the legs than in the arms.[1] More than 95% of the patients with SMA have a homozygous disruption in the *SMN1* gene on chromosome 5q, caused either by mutation or deletion. Mutation in the *SMN* gene results in a loss of function of the SMN protein.[2] The incidence of SMA is 10–15 in 100,000 live births, affecting all ethnic groups; it is the second most common neuromuscular disease, following Duchenne muscular dystrophy. The incidence of heterozygosity for autosomal recessive SMA is 1 in 50.[3,4]

CASE REPORT

This case was a 12-year-old female child born to healthy nonconsanguineous parents, presented with complaints of wasting and weakness of both lower limbs for 3 years. On examination, her higher mental functions were normal. Cranial nerves were intact. There were typical worm-like fasciculations seen in the tongue [Figure 1]. Fasciculations were present in both upper and lower limbs. A power of Grade 3–4 was present in the upper limbs. Grade 2–3 power was present in the lower limbs. There was no bulbar involvement. Respiratory muscles were not affected. Upper extremities were less affected. The child was able to feed herself. There was no family history of any neurological illness. Other two siblings were normal. Molecular genetic diagnosis for the analysis of SMN gene in blood sample was done, which confirmed the diagnosis. Serum creatine kinase levels were raised. Electromyography showed spontaneous fibrillation at rest. Nerve conduction study was normal. On the basis of clinical presentation and confirmation by lab investigations, a diagnosis of SMA-Type 3 (Kugelberg–Welander disease) was made by a pediatric neurologist.

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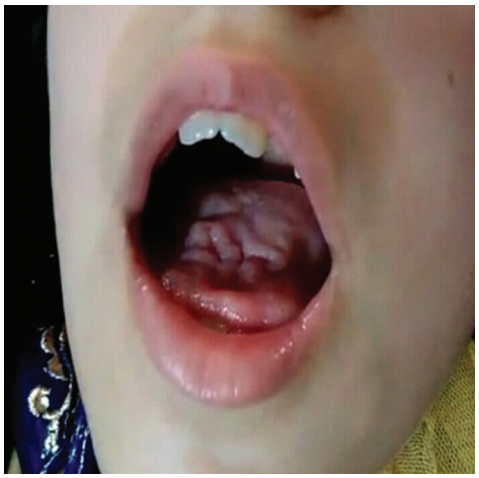

Figure 1: Worm-like tongue fasciculations

There is no medical treatment available to this condition. Supportive therapy includes orthopedic care and physiotherapy which was given to our patient.

DISCUSSION

SMA is the second most common neuromuscular disease.[3] SMA is classified into four types. Very severe SMA Type 0: manifests before birth and it is characterized by a reduction in fetal movements in the final months of pregnancy. SMA Type 1: severe infantile form (Werdnig–Hoffmann disease), manifests within the 1st few weeks or months of life when abnormally low muscle tone is observed in the infant (the floppy baby syndrome). SMA Type 2: late infantile and more slowly progressive form. SMA Type 3: more chronic or juvenile form (Kugelberg–Welander disease). Although the child may appear normal during infancy, there is a slow but progressive weakness of limbs. Bulbar dysfunction occurs late in the disease. As the disease progresses slowly, the overall course is mild. Many patients have normal life expectancy. SMA children do not differ from the general population in their behavior; their cognitive development can be slightly faster and certain aspects of their intelligence are above average.[5]

There is no medical treatment available to this condition. Respiratory system requires utmost attention in SMA, as once weakened it never recovers fully. Weakened pulmonary muscles in SMA Type I/II patients can make breathing more difficult and pose a risk of hypoxia, especially during sleep when the muscles are more relaxed. Impaired cough reflex poses a constant risk of respiratory infections and pneumonia.[6] Noninvasive ventilation (bilevel positive airway pressure) is frequently used, and tracheostomy may be sometimes performed in more severe cases.[6]

Genetic counseling should be offered to all families of patients with SMA. The role of prenatal diagnosis, particularly in pregnant carriers or those with juvenile or adult-onset forms, should also be addressed. Preimplantation genetic diagnosis can be used to detect SMA-affected fetus, especially when undergoing *in-vitro* fertilization. Prenatal testing toward SMA is possible through chorionic villus sampling, cell-free fetal DNA analysis, and other methods. Those at risk of being carriers of *SMN1* deletion, and thus at risk of having offspring affected by SMA, can undergo carrier analysis using blood or saliva sample.

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Conflicts of interest

There are no conflicts of interest.

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