

Spinal muscular atrophy: A comprehensive case report

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World Journal of Biology Pharmacy and Health Sciences, 2024, 18(01), 196–197

Publication history: Received on 28 February 2024; revised on 08 April 2024; accepted on 11 April 2024

Article DOI: <https://doi.org/10.30574/wjbphs.2024.18.1.0188>

Abstract

Muscle Weakness and Atrophy, Hypotonia, Progressive Degeneration, Motor Function Impairment are hallmarks of Spinal Muscular Atrophy, a neuromuscular genetically inherited disorder that causes muscle weakness and wasting usually diagnosed in infancy or early childhood and rarely identified in adults. The genetic components of the disease are inherited, meaning that the afflicted individual receives the gene from a parent with the same genetic makeup. It is inherited in an autosomal-recessive manner. The survival motor neuron gene 1 (SMN1) is the gene responsible for the most prevalent kind of SMA. A patient with Spinal Muscular Atrophy is described, associated their clinical with and genetic features.

Keywords: Spinal Muscular Atrophy (SMA); Supportive therapy; Survival motor neuron (SMN); Muscle Weakness; Atrophy; Progressive Degeneration

1. Introduction

Spinal Muscular Atrophy (SMA) is a group of disorders that causes muscle weakness and wasting [1]. SMA affects approximately, 1 in every 6,000 to 10,000 [2]. Spinal Muscular Atrophy is inherited by an autosomal recessive gene; a pattern typically inherits one mutated copy of the gene from each parent, making them carriers without showing symptoms. Spinal Muscular Atrophy is caused by an autosomal recessive survival motor neuron gene 1 (SMN1), is the gene responsible for the most prevalent kind of SMA. The survival motor neuron (SMN) protein, which is produced by the SMN1 gene on chromosome 5q, keeps motor neurons healthy and functioning normally. Insufficient amounts of the SMN protein are present in people with SMA, which results in the death of motor neurons in the spinal cord, skeletal muscle weakness, and wasting [3]. Based on the age at which muscle weakness first appears and the degree to which motor milestones are not met, the condition is commonly categorized as spinal muscular atrophy type SMA1 to SMA4. Early infancy is when SMA1 first manifests as generalized proximal muscular weakness and atrophy. SMA4 has a weaker adult onset [1]. Spinal Muscular Atrophy illness cannot be rectified with any treatment. While treating spinal muscular atrophy (SMA), infusion treatment (nusinersen, onasemnogene abeparvovec-xioi*) [4,5] uses needles or catheters to provide medicine. It functions to help manage symptoms and prevent progressive weakness and deterioration of your skeletal muscles. The mainstays of treatment are supportive therapy, which aims to minimize problems from muscular weakness and provide nourishment and respiratory assistance [1]. Low body weight, food issues, and swallowing difficulties are common in SMA patients [6]. It is advised to closely evaluate anthropometry and provide optimal dietary assistance [7].

2. Case description

This case involves a 32-year-old male patient who was diagnosed with Spinal Muscular Atrophy and admitted to the neurology department; The patient described a progressive loss of muscular strength, especially in the lower limbs, which made it difficult for them to walk and stay still for extended periods of time. He also showed obvious signs of

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muscular atrophy and had trouble with fine motor skills. The patient disclosed that, to the best of his knowledge, there is no family history of this illness. Genetic testing and electromyography (EMG) were used as diagnostic procedures, and the results showed distinctive findings that were compatible with SMA. A muscle sample that revealed evidence of atrophy of the muscular fibers and denervation confirmed the diagnosis. A multidisciplinary approach to the patient's care was given, which included physiotherapy to keep the patient's muscles strong and mobile. The patient and his family members were also provided with genetic counseling to talk about inheritance patterns and possible concerns for future generations. Follow-up sessions were planned on a regular basis to track the disease's course and modify care plans as necessary.

3. Conclusion

An inherited, long-term neuromuscular system disorder called Spinal Muscular Atrophy is marked by progressive muscle weakness and motor impairment due to the degeneration of motor neurons in the spinal cord. Due to the degenerative nature of Spinal Muscular Atrophy, patients and their families require continuing care and multidisciplinary treatment to get the best support possible.

Compliance with ethical standards

Statement of informed consent

Informed consent was obtained from the individual participant included in the study.

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