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Spinal muscular atrophy: A case report

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Abstract

Spinal muscular atrophy (SMA) is an autosomal recessive disorder which is a fatal neuromuscular disorder in infants. SMA type 1 is first noticed either during pregnancy or within the first few months of life. Symptoms include weakness and limited movement in the limbs and trunk, impaired breathing, and feeding difficulties. Here we describe a case of severe hypotonia and respiratory distress during infancy.

Keywords: Spinal muscular atrophy, hypotonia

1. Introduction

SMA is autosomal recessive disorder that leads to the degeneration, atrophy, and eventually to the death of the motor neurons in the spinal cord. It is a disease that is most frequently diagnosed in children, and is the most common fatal neuromuscular disorder in infants. There are three distinct types of spinal muscular atrophy: type I, type II, and type III, all of which are seen in children, and type IV which is diagnosed in adulthood. Type I SMA is the most severe and the most commonly diagnosed type of SMA [1].

The spinal cord, specifically the anterior horn cells, is affected by spinal muscular atrophy. These cells are responsible for the voluntary movements of large muscle groups such as the arms and legs. Proximal muscles are more severely affected than the hands, fingers, and toes. SMA only damages motor neurons, so the patient's sensory, mental, and other functions remain normal. Genetic testing is done to confirm a diagnosis, as well as other clinical assessments. A patient is typically diagnosed with SMA after an electromyography (EMG). This test measures electrical impulses as they travel from the brain to the voluntary muscles. SMA can also be diagnosed with a muscle biopsy, in which a small piece of muscle tissue is examined for degeneration [2].

Spinal muscular atrophy is an autosomal recessive disorder, in which both parents must be carriers in order for the disease to show. If both parents carry the gene, there is a 25% chance the child will be afflicted with the disease. All three forms of SMA result from a decrease in the Survival of Motor Neuron (SMN) protein. The SMN protein is coded by two genes is located on chromosome 5; SMN-1 and SMN-2. For an unknown reason, only SMN-1 abnormalities result in spinal muscular atrophy. Approximately 2% of all SMA cases result in new mutations to the SMN-1 gene, and are not inherited from the parents. It affects populations uniformly throughout the world.

Type I SMA is also known as Werdnig-Hoffmann disease. It is first noticed either during pregnancy or within the first few months of life. Symptoms include weakness and limited movement in the limbs and trunk, impaired breathing, and feeding difficulties. While pregnant, women may notice decreased fetal movement. Children who suffer from type I SMA never are able to gain gross motor skills, and the majority of these children die before their second birthday [3].

There is no cure for any form of SMA. There is hope that gene therapy may become possible to replace the mutated or missing gene, but this possibility is still years away.

Currently, the only treatment options involve the management of symptoms. Respiratory complications are the most critical issue for SMA patients, and result in the majority of deaths. As time progresses, a ventilator is typically required to aid breathing. The intercostal muscles between the rib cages are typically weakened, which causes lungs to not develop properly. As a result, diaphragm faces an increased demand and is responsible almost solely for breathing. This does not allow the rib cage to expand as necessary during inhalations. Hypoventilation is common during sleep.

It is often the first sign of respiratory illness seen in patients. Coughing is essential in SMA patients to clear the lungs and throat of moisture and prevent secondary infections, such as pneumonia. Chest physiotherapy (CPT) is used to trigger and assist patients in coughing.

2. Case description

A 5 month old child was transferred from a local hospital to male child was referred from local hospital to physiotherapy department of Pravara medical hospital with complains of severe hypotonia. He was born at 37 weeks of gestation by normal delivery with 2700 gm of birth weight. After birth he was admitted in NICU for investigations of intra uterine growth retardation but was discharged as no specific abnormality was found. There is no history of any neurological disorders / genetic conditions / early deaths in either parents' family.

At age of four months his mother consulted local practitioner for as he was not holding neck. On first day of consultation he looked lethargic with no social smile and no eye contact. Deep tendon reflexes were absent. Upper extremity power was grade 2 and lower extremity was grade 1. Tongue fasciculations were seen. His serum creatinine level was elevated. Tongue fasciculations were seen. Nerve conduction study showed decreased lower extremity nerve conduction velocity and reduced action potential. EMG studies show abnormal insertional activity and reduced recruitment pattern. Electrodiagnostic study anterior horn cell disease including SMA. Genetic studies show mutation of SMN-1 gene.

He receives physical therapy, and play therapy considering his limitations as well as chest physiotherapy to maintain his respiratory status. His respiration is now very effortful and progressively distressed. His sucking power is reduced. When not under the care of his mother, he is cared for by a private nurse. He is fully dependent for all daily living tasks. He now uses a tracheotomy for respiration, and receives nutrition through a gastric tube. During physiotherapy treatment sessions, the therapist works on maintaining the available range of motion by passive range of motion in her extremities and muscle facilitations. His current therapy goals are to maintain his current range of motion, to provide as many opportunities as possible for his age level considering his limitations, and to provide perceptual motor and visual motor activities with adaptations as required. Splints and other equipment are also provided as needed.

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