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Case Report

A -2- month old child with Spinal Muscular Atrophy Type 1 with Pneumonia: A case report

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Abstract:

Spinal muscular atrophy (SMA) is a autosomal recessive disease characterized by muscular weakness, hypotonia and atrophy. SMA is caused by mutation or deletion of the survivor motor neuron gene (SMN1), which is located in the telomere region of chromosome 5q13. Incidence of the disease is 1:6000-10000 newborn. Confirmatory diagnosis can be established by molecular genetic analysis. Here a 2- month- old female baby was admitted into the department of pediatrics of Sylhet Women's Medical College Hospital with because of less movement of whole body for 1 month and repeated cough with respiratory distress for same duration. Baby was dyspneic, chest in drawing and crepitation was present over both lung fields. Signs of lower motor neuron lesions were present. It was confirmed by Polymerase chain reaction (PCR) for SMN gene. Final diagnosis was Spinal muscular atrophy with pneumonia. Symptomatic, Supportive and antibiotic treatment were given. Genetic counseling, regular respiratory follow up was advised.

Key words: Spinal muscular atrophy, Polymerase chain reaction.

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Introduction

Spinal muscular atrophy (SMA) is a genetic disease with autosomal recessive inheritance. It is a degenerative disease of motor neurons that begins in fetal life and progressive in infancy and child hood .SMA is the important cause of infant mortality and is second in position in birth prevalence only to cystic fibrosis. Overall incidence of the disease is 1:6000-10000 newborn.[1] Frequency of carriers of SMA is 1 in 40 to 60 people.[2] It is caused by homozygous mutation or deletion of the survivor motor neuron gene (SMN1), which is located in the telomere region of chromosome 5q13.[3]

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The disease sometimes presents with decreased motor activity and muscle atrophy due to degeneration of anterior horn cell in the spinal cord and motor cells of lower cranial nerves nuclei.[4]

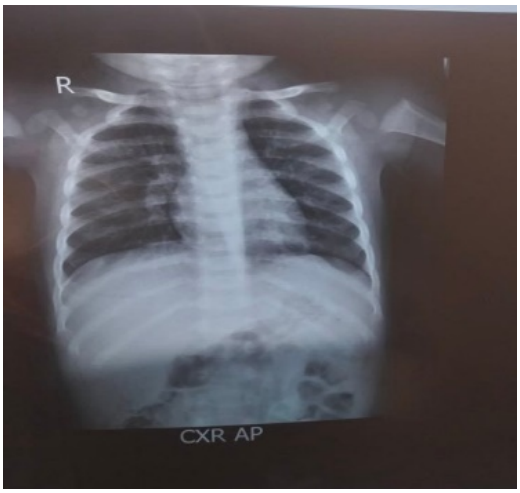
Occurrence of mutation in *SMN1* causes degeneration of anterior horn motor neuron and leads to hypotonia, muscular weakness and atrophy.[5] SMA is classified clinically into several types based on age of onset and degree of motor function achieved by the affected individual.[6] Severe infantile form is also known as Werdnig-Hofmann disease or SMA type 1. SMA type 1 usually present before the age of 6 month.[1] Diagnosis is based on evidence of denervation of muscle both in electrophysiology and genetic analysis.[6]There is no specific treatment of the disease. Lifelong medical follow up and palliative care both are important for every patient with SMA. Genetic counseling has important value to the families of these patients.[3,6] As the patients with SMA suffer commonly from recurrent respiratory infections which may lead to life threatening respiratory failure, so regular respiratory follow up is needed. Similarly our presented case, Type 1 Spinal Muscular Atrophy was also suffering from recurrent respiratory problem. So we reported the case.

Case Report: A 2 month old female child (**Picture-1**) was admitted at department of pediatrics of Sylhet Women's Medical College Hospital, Sylhet because of less movement of whole body for 1 month and repeated cough with respiratory distress for same duration. She had no history of delayed passage of meconium, prolonged jaundice, constipation and any heart disease. She was delivered at term at home without any perinatal complications. Baby was the first issue of her non consanguineous parents. No family history of similar type of illness. Baby was dyspneic, chest indrawing and crepitation was present over both lung fields. Her posture was severe hypotonic. Upper extremity power was grade 3 and lower extremity power was grade 2 symmetrically. Deep tendon reflexes were absent. Tongue fasciculation was absent. From history & clinical examinations, provisional diagnosis was Floppy Baby most probably due to Spinal Muscular Atrophy with Pneumonia. Molecular diagnosis showed (**table 1**) a deletion of the exon 7 and 8 of SMN-T (telomere region) of the DNA from her peripheral blood. Chest X-ray reveals pneumonitis (**Picture-2**). So final diagnosis of the baby was Spinal Muscular Atrophy Type 1 with Pneumonia.

Picture 1. A Baby with SMA

Table 1. **Genetic diagnosis**

Gene	No of copies
SMN 1 Exon 7	0 copy detected
SMN 1 Exon 8	0 copy detected
SMN 2 Exon 7	2 copies detected
SMN 2 Exon 8	2 copies detected



Picture 2: CXR

Discussion

Spinal muscular atrophy type 1 (SMA-1) is a severe form of neurodegenerative disorder. Hypotonia, muscle weakness, atrophy are the main symptoms. It begins during the fetal period and progresses through infancy and childhood. It is well known that infants become floppy in SMA type 1 due to weakness of the proximal limb muscles before progressing to respiratory failure secondary to paralysis of the intercostal muscles. In recent years, several drugs have been developed and reached phase I–III clinical trials in different region, a targeted treatment has been developed with the antisense oligonucleotides, which alters splicing of SMN2 pre-mRNA and thus increases production of functional SMN protein.[7] It is said, over few years nusinersen has shown some clinical efficacy in well-controlled clinical trials with prolonged survival beyond 2 years of age in different populations of SMA patients, including severe SMA1 patients.[8] Our patient presented with less movement of the limb and hypotonia. We suspected Spinal muscular atrophy (SMA) based on the presenting features of the baby. Diagnosis was confirmed by Polymerase chain reaction (PCR) for SMA. Early diagnosis of the disease avoid prolonged ventilation and allows appropriate counseling.[9] Lokesh et al. had reported a case of a three month old female child with SMA type 1 presented with recurrent respiratory infection in the same manner as like our study case .[10] Marie et al had reported on 80 infants with severe SMA-1, followed in France

between 2012 and 2016.[7] Along with generalized weakness patients may develop chronic respiratory failure and bulbar dysfunction in infants leads to death before 2 years of age without ventilator support.[11,12] There was recurrent respiratory tract infections observed in our study case. Several data have been published recently suggesting available simple tools to evaluate respiratory function among the infants with SMA type 1.[13]

Conclusion

SMA is an autosomal recessive disease affecting the part of nervous system that control the voluntary movement of muscle. SMA type 1 is the extreme form among the other type of SMA. This type of patient may develop recurrent respiratory infections that leads to chronic respiratory failure and ultimately early death. So genetic testing of the disease, respiratory follow up and counsel the parents are very important.

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Ethics approval: Informed consent was obtained from the patient parents before the case report was written

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