



## CASE STUDY

### SMA TYPE 2 PRESENTING AS FLOPPY INFANT

**\*Nasreen Ali, Sunil Kumar Agarwalla, Niranjana Mohanty and Debasis Patro**

Department of Pediatrics, M.K.C.G Medical College, Berhampur, Ganjam, Odisha-760004, India

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#### ABSTRACT

Spinal muscular atrophy is the second most common autosomal recessive disorder after cystic fibrosis (D'Amico *et al.*, 2011). Because of its high frequency, it should be considered first in a case of floppy infant. Here we are reporting a case of 18month female baby who was admitted with complaints of not able to walk for last 5 months.

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## INTRODUCTION

SMA is an autosomal recessive disease in which there is progressive damage of lower motor neuron, due to loss of anterior horn cells of spinal cord. Its incidence is 1 in 10,000. Of this, stage 2 is the largest group with 50% of all SMA cases. This occurs due to homozygous deletion of Survival motor neuron (SMN1) gene on 5q in 95% of cases (Brzustowicz *et al.*, 1990). The incidence of carrier is 1 in 50 (Ogino *et al.*, 2002). Because of this high incidence of carrier, almost all cases of floppy infant should consider SMA as differential diagnosis. There is also a recurrence rate of 25% in subsequent pregnancy (Prior *et al.*, 2010). The attentiveness and intelligence is good. The weakness is symmetrical and proximal mostly. It is greater in lower limbs compared to hands.

## CASE REPORT

A 18 month female baby admitted in paediatric department of MKCG with complains of inability to walk with support, for last 5 months. This was gradual in onset and progressive in nature. There is poor cry and regurgitation of food. The baby was born out of third degree consanguinous marriage by normal vaginal delivery. There was no history of birth asphyxia. The child had attained all milestones appropriate for age till 13 months of age. There is history of 2 abortions at 8

months of gestation and 1 sibling death at 18 months of age following respiratory distress. In the perinatal period there is history of reduced foetal movement. The baby is immunized as per National Immunization schedule. On examination, the child lie supine, still without any movement of limbs, if made to sit there was no neck control, the higher function and language is intact. There is fasciculation of tongue. The respiratory rate was 34/min regular and abdomino thoracic type. The bulk of the muscle is normal, there is gross hypotonia, superficial reflexes are preserved but the deep tendon reflexes are absent (areflexia). The sensory system is intact. Bowel bladder movement was preserved. Serum CPK came to be normal. In view of bad obstetric history and one sibling death at 18 months with similar kind of illness, a clinical diagnosis of SMA was made because of progressive LMN palsy leading to floppy baby with are flexia. The child didn't need any acute intervention hence the parents were counselled about the disease, course, recurrence and were advised about alternatives like adoption.

## DISCUSSION

ISMALC classification is based on age of onset. According to which:

- TYPE 1- Acute infantile (Werdnig Hoffman disease)-age of presentation 0-6 months.
- TYPE 2- Chronic infantile (intermediate type)-age of presentation 6-18 months.

\*Corresponding author: Nasreen Ali,  
Department of Pediatrics, M.K.C.G Medical College, Berhampur,  
Ganjam, Odisha-760004, India.

TYPE 3- Chronic juvenile (Krugelberg Walender disease)-age of presentation >18 months.

TYPE 4- Adult type-presents in 2<sup>nd</sup>-3<sup>rd</sup> decade.



Child lying frog like position



Child not able to sit and loss of neck control

Mortality is inversely proportional to the age of presentation. The differential diagnosis to SMA include congenital myotonic dystrophy, congenital myasthenic syndromes, metabolic myopathies, hypotonic CP, prader-willi syndrome, down syndrome, congenital hypothyroidism etc. These can be

differentiated by taking proper history and clinical examination. Investigations like SMN1 gene detection is confirmatory. Other investigations which can be done are EMG, NCS, CPK and muscle biopsy, but these are not confirmatory.

### Conclusion

The prognosis of SMA depends on the type of SMN gene affected. 95% have SMN1 gene affected (Wirth, 2000). In type 1 SMA death occurs by the age of 2 years and type 4 can lead a near normal life. The most common cause of death is respiratory infection. There is no treatment available now but with proper diagnosis we can suggest parents about adoption. Since the cost of genetic testing is high, the clinical diagnosis is helpful atleast to differentiate from other causes of floppy infant which do not have such high recurrence rate.

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