

**A RARE CASE REPORT: NEONATAL HYPERKPLEXIA WITH FAILURE TO THRIVE WITH UMBILICAL HERNIA****J. K. Roshith<sup>1</sup> and K. N. V. Prasad<sup>2\*</sup>**<sup>1</sup>MBBS, Post Graduate (MD Pediatrics).<sup>2</sup>Professor & HOD of Pediatrics. MBBS, MD Paediatrics.

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**ABSTRACT**

**Background:-** Hyperekplexia is an inherited autosomal dominant and a rare non-epileptic disorder which is characterized by an exaggerated and persistent startle reaction to the unexpected tactile and acoustic stimuli. The disorder is occasionally associated with generalized muscular rigidity and the clinical confirmatory test is by positive nose tapping test. Hyperekplexia is easily misdiagnosed as disorders such as spastic cerebral palsy and epilepsy. Clonazepam is of benefit, but it does not always stop the prolonged spasms. Forced flexion of baby's head toward the trunk is the most effective treatment. We report a 8 months female infant who presented with excessive jerky movements of the and complaints of vomiting since 20 days of age. Mother complaining of her child having jerky movements were cured after the use of clonazepam completely.

**KEYWORDS:** hyperekplexia, spasms, clonazepam.**INTRODUCTION**

Hyperekplexia, or familial startle disease or congenital stiff-man syndrome was first described by Kirstein and Silfverskiold in 1958.<sup>[1]</sup> It is an uncommon hereditary neurological disorder characterized by brief episodes of intense generalized tonic spasms in response to sudden unexpected loud noises or tactile stimulation such as a tap on the tip of nose or shoulder. Affected neonates suffer from prolonged periods of rigidity and are at risk for sudden death from apnea or aspiration.<sup>[2,3]</sup> One form, with neonatal onset, can be very severe and characterized by repeated myoclonic jerks and increased muscle tone with life threatening apnea due to contraction of respiratory muscles. It can cause life threatening death or can die of cardiopulmonary arrest if a tonic episode occurs in sleep and this can be terminated by life-saving maneuver by forcible flexion of the head and neck toward the trunk.

**Abstract –** A 8 months old female infant came to out patient department with swelling around umbilicus since 7 days of life, complaints of vomiting, abnormal jerky movements of both upper and lower limbs since 20 days of life. No history of excessive cry or refusal of feeds. Infant was admitted for similar complaints in hospital at 3 months of age but treatment details unknown to mother. No medications were advised on discharge. Mother gives history of increase of swelling over umbilicus while coughing, straining for passing motion and reduces by its own. Mother gives a history of

uneventful antenatal, natal and postnatal. She also gives history of normal developmental age for age and fully immunized for age. On physical examination Infant was conscious, looking pale over eyes, vitals were stable. Anthropometry measures like weight, length and head circumference were less than 5<sup>th</sup> and 3<sup>rd</sup> percentile as per WHO charts. Central Nervous system – Rigidity was seen in all muscle groups including trunk and abdominal wall.

Deep tendon reflexes were normal. Infant held herself in a flexed posture, with clenched fists and had an anxious look as in below mentioned picture. Exaggerated non-habituating startle response along with increased period of hyper tonicity were demonstrated on tapping of the nose. She also had stereotypic movements in the form of rocking herself to and fro while lying supine, for long periods of time especially when disturbed by excessive hand ling, loud or sudden sound and on separation from parents. No neurological deficit.

**Abdomen examination–** Infant was having swelling of 10cm length and 10 cm breadth present over umbilicus. Swelling was soft, reducible, non tender, impulsive on coughing, increases on coughing and straining.

Child was treated with Clonazepam initial dose (0.1mg/kg/day) and maintenance dose of (0.3mg/kg/day) to be continued. Mother's complaining of her infant having jerky movements were completely cured.

Nutritional rehabilitation advises were given to mother and pediatric surgery opinion for umbilical hernia was given and follow up.

The blood cell count, glucose, calcium, serum electrolyte levels, renal function test and blood cultures were normal. Her mother had found that severe generalized stiffness could be stopped promptly by flexing the infant's head toward her trunk. This maneuver was always life saving and effective during apnoeic episodes. Episodic attacks decreased remarkably after starting clonazepam. Her feeding pattern improved, irritability decreased and spontaneous attacks disappeared. No other seizure-like episodes and generalized stiffening were noted in the subsequently.



## DISCUSSION

Startle disease or hyperekplexia is an unusual autosomal dominant neurological disorder that is often confused with epilepsy in the neonatal period. The disorder is categorized into major and minor forms.<sup>[5]</sup> The major form is seen in the newborn period when the patient experiences momentary generalized muscular stiffness, hypertonia and an exaggerated startle response. These signs are typically heightened by the slightest stimulus including nose tapping, and disappear with sleep. The muscle stiffness usually returns to normal during the first years of life.<sup>[6]</sup> In the minor form the startle response is exaggerated, being more than normal, without any additional symptoms. Excessive startle occurs as a manifestation of tension, fatigue and sleep deprivation in normal people. Familial history was not significant in our case.

Diagnosis of hyperekplexia is made by a nose tap test. Tapping the tip of the nose of an unaffected baby will elicit no response, but in hyperekplexia there is an obvious startle response which is repeated each time the nose is tapped. Tactile stimulation, especially nose tapping test, was more provoking than other stimuli (auditory, visual) in our case. The underlying basis for the hyperekplexia is a mutation or change in the gene involved in the function of neurotransmitter glycine.

Glycine is an inhibitory transmitter because it damps down the action of nerve cells, particularly in the spinal

cord and brain stem. When glycine receptors are impaired, the nerve cells are too easily excited. C.S.F gamma-amino-butyric acid (GABA) levels were reported to be low in some affected patients and because clonazepam acts through the GABA Type-A receptors, a genetic defect in GABA neurotransmitter receptor is thought to be the cause of this disorder.<sup>[4]</sup>

Hyperekplexia must be differentiated from other disorders including neonatal tetanus, neonatal drug withdrawal syndrome, Tay-Sach's disease, apneic spells, tonic seizures, spastic quadriplegia and startle-provoked epileptic seizures (startle epilepsy) that are often due to occult congenital lesion. These seizures are often therapy resistant and the prognosis is generally poor.<sup>[5,7,8]</sup>

Clonazepam appears to be the drug of choice (0.05 to 0.1 mg/kg/day) for these patients. No definite guideline exists regarding the duration of therapy but the drug can be tapered slowly once the symptoms subside.<sup>[9]</sup> Flexion of infant's head and neck toward the trunk has been found to be the best way of treatment in life-threatening events (severe tonic spasms and apnea).<sup>[9]</sup> Development is eventually normal in some children.<sup>[5,10]</sup>

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