HYPEREKPLEXIA AND EXCESSIVE STARTLE RESPONSE IN AN INFANT: A CASE REPORT

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ABSTRACT

We present an infant girl with hyperekplexia, hypertonia, hyperreflexia and a characteristic exaggerated response to nose tap. This disorder is important to recognize because of the increased risk of apnea and sudden infant death. This infant responded to clonazepam.

Keywords: Hyperekplexia, Hypertonia, Startle.

INTRODUCTION

Hyperekplexia is a rare familial disorder associated with whole body myoclonus presenting as a hyperactive startle reflex which occurs during the neonatal or early infancy periods. When handled, a minority of infants become stiff with severe hypertonia leading to apnea and bradycardia. These movements must be distinguished from startle epilepsy. Prognosis is variable, and seizures do not accompany the benign form of this disorder. Clonazepam or valproic acid may be useful.

CASE REPORT

A 45 day old infant girl suffering from seizure like episodes was admitted in the pediatric ward of Imam Reza Hospital of Mashhad University of Medical Sciences. The infant was born by normal vaginal delivery and her weight was 3300 grams. On the first day she was admitted to the nursery and treated with phenobarbital and phenytoin because of seizures. But there was no response and after a few days her mother took her home. Her gestation, labor and delivery were unremarkable and Apgar scores were 7 at 1 minute and 1 at 5 minutes. On examination, the infant was alert, and attention and movements were appropriate for her age. Head circumference was normal and present weight was 3700 grams. Gag and sucking of this child seemed to be hyperactive. Touching the child’s face produced an immediate head recoil with extension of the limbs. Tapping the nose appeared to be the most effective method of eliciting the head recoil. Tone was symmetrically increased and ankle clonus was present. Deep tendon reflexes were increased. Feeding was difficult because touching the breast to her mouth elicited the head recoil response. Tremor of the limbs was elicited by touch, a loud sound, or by shining light in the infant’s eyes. The asymmetric tonic neck reflex was normal. Brain CT scan and electroencephalogram, formerly performed, were normal. After diagnosing hyperekplexia and startle response clonazepam was begun which immediately improved these movements and feeding. The baby was discharged after a week. Social and cognitive function were normal at 4 and 10 months of life but gross motor development was mildly delayed and remained slightly hypertonic. At the age of 16 months she was able to walk.

DISCUSSION

Hyperstartle syndrome (or Hyperekplexia) is a neurologic disorder characterized by hypertonicity, tremor, and exaggerated response to tactile, auditory and visual stimulation. Two clinical groups of this syndrome have been proposed. Major hyperekplexia is the term proposed to describe patients with the following features: hypertonicity in infancy, excessive startle response, startle induced falls without loss of conscious-
Hyperekplexia in an Infant

Hyperekplexia may present in the newborn period with hypertonia, hyperreflexia, and a characteristic exaggerated response to nose tap. This disorder is important to recognize because of the increased risk of apnea and sudden infant death. Most infants respond to clonazepam and close follow up is recommended.

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REFERENCES
