Nose-tapping Test in Hyperekplexia

A term, male baby was hospitalized on first day of life in view of excessive cry, frothing and stiffening followed by apnea during routine care after birth, and after receiving Hepatitis-B vaccination. Blood glucose, serum electrolytes, calcium, electroencephalogram and neurosonogram were normal. Nose-tapping test (***Fig.* 1** and ***Web Video* 1**) resulted in exaggerated startle and stiffening episodes without habituation. A diagnosis of hyperekplexia was considered, and treatment with clonazepam was started. On titrating the dose of clonazepam, the frequency of episodes decreased over the next 7 days, and baby was able to breastfeed from day 4. By discharge, baby was accepting breastfeeds well. Heterozygous mutations in *HSPG2* and *TUFM* genes located on chromosomes 1 and 16, respectively, were identified.

Hyperekplexia is a rare, hereditary neurological disorder characterized by episodes of tonic stiffening that is often confused with epilepsy in the neonatal period. In majority of cases, hyperekplexia is inherited as an autosomal dominant trait, but autosomal recessive or *X*-linked inheritance patterns are also noted. The genes associated with this condition are *GLRA1*, *SLC6A5*, *GLRB*, *GPHN*, and *ARHGEF9* (*X*-linked).

Clinical diagnosis of hyperekplexia is made by a nose-tapping test. Tapping the nose of a normal baby will elicit either a blink response or no response and habituation to the response will be present but in a baby with hyperekplexia, nose-tapping results in exaggerated startle and stiffening episodes without habituation. Close clinical differentials include jitteriness, myoclonic seizures, neonatal tetanus, startle epilepsy, and neonatal abstinence syndrome. The drug of choice for hyperekplexia is Clonazepam (0.05 to 0.1 mg/kg/day). Life-threatening events, including severe tonic spasms and apnea, are best treated by Vigevano maneuver (flexion of infant's head and neck toward the trunk). Long term outcome of these infants usually depends upon the severity of the problem.

FIG. 1 *Nose-tapping test (See video at website).*

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NOTICE

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