雑誌

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Visual Diagnosis

Startles, Stiffness, and *SLC6A5*: Do You Know the Condition?

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This seven-month-old girl presented with excessive startle and episodic tightening of body since birth. She was born to nonconsanguineous parents with a normal perinatal period. There was no history suggestive of seizures, dyskinesia, or neuroregression. Her development was appropriate for age. On examination, she had normal head circumference, exaggerated startle, persistent head retraction response (Fig 1), and brisk muscle-stretch reflexes. Systemic examination was unremarkable. Genetic testing discovered homozygous mutation of *SLC6A5* gene, p.L460P; both parents were carriers. She improved with oral clonazepam (0.03 mg/kg/day in three divided doses). At a recent one-year follow-up, her startle episodes have reduced in intensity and she has mild motor delay.

Hereditary hyperekplexia is characterized by generalized stiffness at birth, which may normalize during the first few years of life; excessive startle reflex to unexpected sudden stimuli; episodic stiffness related to the startle; and exaggerated head retraction response on tapping the nose-tip or mantle area.[1] This head retraction reflex with absence of habituation is also described in children with cerebral palsy secondary to severe perinatal asphyxia. A normal development and absence of adverse perinatal events differentiate the two conditions clinically. Children with hereditary hyperekplexia may develop mild intellectual disability later in life although the majority remains normal. *SLC6A5* mutations affecting presynaptic sodium and chloride-dependent glycine transporter-2 are a rare cause of hereditary hyperekplexia.[2] Clonazepam has been proposed as the most effective treatment to diminish stiffness and startle responses. Attacks of tonic neonatal cyanosis may be aborted by the "Vigevano maneuver" (forcible flexion of the head and legs over the trunk).[3]

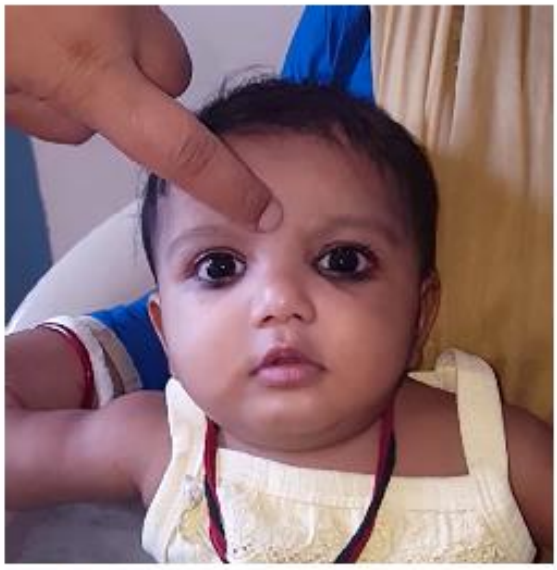

FIGURE 1.
Hereditary hyperekplexia in infancy is manifested by an. exaggerated startle response with no habituation on head tapping, exaggerated head and neck-retraction response on repeated tapping on the nose-tip, upper lip, and mantle area. The video related to this figure can be accessed at 10.1016/j.pediatrneurol.2017.06.005. (The color version of this figure is available in the online edition.)

Keywords: exaggerated startle reaction, hyperekplexia, stiff baby syndrome, *SLC6A5*
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