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SCIENTIFIC LETTERS TO THE EDITOR

Hyperekplexia Masquerading as Epilepsy

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Sir,

Hyperekplexia is a rare neurogenetic disorder of glycine neurotransmission, characterized by an exaggerated startle response to tactile or acoustic stimuli and tonic spasms [1]. A non-habituating startle response to nose tapping is considered as clinical hallmark [2]. Glycine has an important role in the central nervous system as an inhibitory neurotransmitter. Here, we describe this disorder in two sibs, referred to us as cases of epilepsy. This report describes the first family with hyperekplexia from Bangladesh.

Two male sibs from Bangladesh presented to our hospital with complaint of startles and tonic spasms. Elder sib was aged 10 years and younger sib was aged 7 years at the time of presentation. They were born to third degree consanguineous Muslim couple with an uneventful antenatal and natal period. Both sibs also had history of neonatal onset exaggerated startle in response to auditory and tactile stimuli. Both sibs had history of nocturnal tonic spasms, which were brief, resulting in generalized hypertonia and aborted by flexing the head and trunk. Both sibs were treated with multiple antiepileptics but there was no response. There was no history of similar symptoms in other family members of three generation pedigree.

On clinical examination, both had normal anthropometry and intelligence, but had marked hyperactive behavior. Percussion over trigeminal afferent area (glabella and tip of nose) produced a startle response, which was non-habituating. Auditory startle response was easily elicited. Rest of the examination was normal. Investigations revealed normal electroencephalogram and magnetic resonance imaging of brain.

A diagnosis of hyperekplexia was made in view of early onset exaggerated startle response, episodic tonic spasms, non-habituating startle response, family history suggesting recessive inheritance and normal investigations. Both sibs were treated with clonazepam 0.1–0.2 mg/kg/day which controlled the tonic spasms completely and decreased the startles significantly.

A search for genetic mutations was performed in Shimane University Hospital, Japan but none of GLRA1, SLC6A5, GLRB, GPHN, and ARHGEF9 genes mutations were found in our patients. Absence of these mutations suggest possible role of novel mutation in the present cases.

Hyperekplexia should be distinguished from epilepsy as it is treatable. Clonazepam is the drug of choice, which dramatically diminishes exaggerated startle response. During attacks of tonic spasms, the limbs and head may be flexed towards the trunk in order to ameliorate the symptoms (Vigevano maneuver) [3].

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