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EDITORIAL

Familial hyperekplexia: Lessons from the other face of the coin

Samir M Mounir*

*Correspondence: Samir M Mounir, Ass Professor of Pediatrics, Department of Pediatrics, Faculty of Medicine, Minia University, Minia, Egypt.

Email: sammmonir@gmail.com
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Abstract

In this issue, we represent a case which may posse special interest to many neonatologists. Apparently healthy female neonate was born by spontaneous vaginal delivery as a sixth offspring to a third-degree consanguineous Egyptian parent. No significant neonatal problems occurred necessitate NICU admission. She had come to our outpatient neurology clinic at age 15th day with episodic transient generalized stiffness, hypertonia and tonic spasms. These episodes were existed from the first days of life as a result of sudden acoustic or tactile stimulation. Both general and neurological examinations were normal. Routine laboratory workup, electroencephalography (EEG) and MRI brain were normal. Father told us about her two brothers who aged 8 and 6 years old, both have had the same story and diagnosed faultily as epileptics. On next visit, we examined her brothers and found them suffering from repeated fallings, injuries and myoclonic jerks only as a reflex to unexpected various sensory stimuli. Although normal EEG and brain imaging, anti-epileptic combinations drugs, not included oral clonazepam, were prescribed to both without improvement. Good to mention that both of them were short and cognitively impaired. The case was one of neonatal conditions mimic epilepsy (CME) called (hereditary hyperekplexia). Recognition of hyperekplexia in the neonatal period is critical to avoid erroneous diagnoses like epilepsy. In conclusion: Neonatologists should be aware of CME in neonatal period. hyperekplexia teaches us simply three unique lessons in neonatology: First, not all CME are benign as known, hyperekplexia may be fatal. Second, some CME like hyperekplexia may be inherited. Lastly, some antiepileptic medications as clonazepam may be used in treatment of non-epileptic conditions like hyperekplexia.

Keywords: hyperekplexia, familial, neonatal, epilepsy.

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Background and description

Epilepsy imitators are a variety of conditions characterized by occurrence of recurrent paroxysmal events like seizures and habitually misdiagnosed as epilepsies [1], Hyperekplexia (startle disease) is rare neurogenetic CME. It was described for the 1st time in 1958 by Kirstein and Silverskoild [2]. Although different names were given for this condition, a triad symptomatology is the base in clinical diagnosis; it is characterized by an exaggerated persistent startle reaction to unexpected sensory stimuli, generalized rigidity, and nocturnal myoclonus [3]. It can be provoked by glabellar tap which may be considered as a clinical hallmark [4] , (Video 1). Started since birth and attenuated by sleep and progress in age. It may be considered as a fatal disorder due to apnea spells, aspiration pneumonia, frequent injurious falls and a controversial link to sudden infant death syndrome [5] Hyperekplexia may be comorbid with abdominal hernia, hip dislocation and developmental delay.[6] it may be diagnosed in concert with epilepsy [7]. Hyperekplexia has a genetic basis which can explain familial tendency occurring in all forms of heritance yet autosomal dominant was the most commonly detected mode of inheritance. Different genetic mutations in a number of various genes encoding glycine receptor subunits that play an imperative role in the inhibitory glycinergic neurotransmission are involved.[4] Although MRIs and CT scans will be normal unless other conditions exist, EEG and electromyogram (EMG) studies may help in hyperekplexia diagnosis. In EEG: fast spikes initially during tonic spasms, followed by background slowness activity with eventual flattening corresponding to the phase of apnea, bradycardia and cyanosis. EMG shows a characteristic muscular response. Nerve conduction velocity is normal. Otherwise, genetic testing is the only definitive diagnosis. A differential diagnosis of hyperekplexia in neonatal period includes congenital stiff-man syndrome, myoclonic seizures, neonatal tetany, and phenothiazine toxicity. Although hyperekplexia isn't an epileptic phenomenon, Clonazepam, a gamma aminobutyric acid (GABA) receptor agonist, is the drug of choice for hypertonia and apneic episodes without influencing the stiffness degree. Forced flexion of the head and legs towards the trunk is known to be lifesaving when prolonged stiffness impedes respiration. [8]. A parental written consent obtained to record the video of this case

Conclusion Neonatologists should be aware of CME in neonatal period. hyperekplexia teachs us simply three unique lessons in neonatology: First, not all CME are benign as known,

hyperekplexia may be fatal. Second, some CME like hyperekplexia may be inherited. Lastly, some antiepileptic medications as clonazepam may be used in treatment of non-epileptic conditions like hyperekplexia.

Conflict of interest: The author declared no conflict of interest.

Author's details
Pediatric Department, Faculty of Medicine, Minia University, Egypt

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