Case Report

An Unusual Startling

Ibrahim Aliyu, Zainab Ibrahim

ABSTRACT

Hyperekplexia is a rare movement disorder, which is mostly of genetic origin; though acquired cases are rarely reported. This disorder is characterized by excessive startling response to external stimuli; this can be disabenling, affecting quality-of-life. Furthermore, it can easily be mistaken for an epileptic disorder. Therefore, the case of a 10-year-old boy who presented with excessive startling shortly after been treated for cerebral malaria is reported; the patient responded to carbamazepine and was discharged home afterward.

Key words: Acquired, hyperekplexia, startle syndrome

INTRODUCTION

Hyperekplexia is a rare movement disorder characterized by an exaggerated response to trivial stimuli that are mostly acoustic and tactile.[1] It is mostly genetic (startle disease).[2] However acquired cases have been documented.[3] Its pathogenesis is linked with deficiency of glycine, an inhibitory central neurotransmitter.[4] Hereditary hyperekplexia is characterized by excessive startle response beginning soon after birth, with the associated generalized hypertonia and exacerbated body stiffness following the startle response.[5] However acquired cases may not have the typical characteristics so described, therefore, the case of a 10-year-old boy who developed an acquired hyperekplexia following an episode of cerebral malaria is reported.

CASE REPORT

This communication is that of a 10-year-old boy who suddenly developed involuntary jerking movements when exposed to loud sound, he had several episodes in a day; this was initially noticed when exposed to loud conversations at home; they had difficulty travelling to the hospital because the jerky movement was stimulated by sound of moving cars and car horn. He also had unstable gait. He could speak but avoided speaking because he felt it could induce the jerking; he could also hear and see and there was no history of loss of consciousness. He was discharged from hospital 2-week before onset of this illness and was treated for cerebral malaria—he had status epilepticus during that period but he completely recovered before discharge. His newborn period was not adversely eventful, and he had normal developmental milestones and scholastic achievement before onset of illness; and there was no behavioral or psychiatric complaint. There was no family history of similar illness.

He had no cranial nerve palsy or signs of meningeal irritation; normal tone globally; he walked with an ataxic gait [Video 1]. When he was stimulated [Videos 2a and b] he responded with excessive startling. Other systemic examinations were not remarkable. The cerebrospinal spinal fluid analysis,
electroencephalogram (EEG) and magnetic resonant imaging of the brain were not remarkable. Therefore, the diagnosis of acquired hyperekplexia was made. He was commenced on slow release carbamazepine and 3-week into treatment the excessive startling subsided [Video 3] with improved ambulation [Video 4]. He is currently being followed-up in the pediatric neurology clinic.

**DISCUSSION**

Hyperekplexia was first described by Kirstein and Silfverskiold, in 1958; when they described a family with ‘drop seizures’.[6] In 1962, Drs Kok and Bruyn further reported on this hereditary syndrome which was then unknown, and they simply described it as “hypertonia in infants.”[7] However in 1966 Suhren et al., evaluated members of a Dutch pedigree with excessive startle reflexes and called the disorder hyperekplexia.[8] Hyperekplexia has been classified into major and minor, and our patient fulfilled the criteria for hyperekplexia minor.

Hereditary hyperekplexia has been associated with mutation in several of the glycine receptor genes and has an established genetic-phenotypic correlation. However, this may not explain the mechanism in all cases because serotonin has also been implicated and Sechi et al.[9] in their report documented the beneficial effect of fluoxetine, further substantiating the serotonergic hypothesis.

Neuropsychiatric startle syndromes (culture-specific syndromes), reflex and startle epilepsy could easily be confused with hyperekplexia, but the absence of neuropsychiatric and intellectual handicaps in the index case, coupled with normal EEG and following critical review of the video clips made the diagnosis easy. Though this disorder is predominantly hereditary; acquired cases had been reported. Although cerebral malaria has been associated with neurologic complications but no mention of hyperekplexia has been reported before now; the exact mechanism is not completely understood, however cerebral hypoxia during periods of repeated seizures might be responsible.

“Vigevano” maneuver, which consist of forced flexion of the head and legs toward the trunk may relieve attacks, especially in the newborn period.[10] Clonazepam is the drug of choice in treating hyperekplexia, while levitiracetem has shown promises in managing this disorder. However, other sedative-hypnotics like carbamazepine, phentoyin, diazepam, valproate, and phenobarbital have been used.[11] Due to nonavailability of the first-line drugs we opted for carbamazepine and he responded with complete resolution of the startling.

**CONCLUSION**

Acquired Hyperekplexia is a rare movement disorder; even rarer is its association with cerebral malaria. The response of the index case to carbamazepine further buttresses its efficacy.

**REFERENCES**

1. Rajadhyaksha SB, Bahl VB. Hyperekplexia: A non-epileptic startle disorder. Indian Pediatr 2002;39:773-6.
2. Lingam S, Wilson J, Hart EW. Hereditary stiff-baby syndrome. Am J Dis Child 1981;135:909-11.
3. van de Warrenburg BP, Cordivari C, Brown F, Bhatia KP. Persisting hyperekplexia after idiopathic, self-limiting brainstem encephalopathy. Mov Disord 2007;22:1017-20.
4. Kingsmore SF, Giros B, Suh D, Bieniarz M, Caron MG, Seldin MF. Glycine receptor beta-subunit gene mutation in spastic mouse associated with LINE-1 element insertion. Nat Genet 1994;7:136-41.
5. Lindahl A. Startles jumps. Neurologic rarities. Pract Neurol 2005;5:292-7. Available from: http://www.pn.bmj.com. [Last accessed on 2014 Apr 18].
6. Kirstein L, Silfverskiold BP. A family with emotionally precipitated drop seizures. Acta Psychiatr Neurol Scand 1958;33:471-6.
7. Kok O, Bruyn GW. “An unidentified hereditary disease”. Lancet 1962;279:1359.
8. Suhren O, Bruyn GW, Tuynman JA. Hyperekplexia, a hereditary startle syndrome. J Neurol Sci 1966;3:577-605.
9. Sechi G, Sotgiu S, Valetti MP, Pitzolu MG, Peterlongo P, Larizza L, et al. Beneficial effect of fluoxetine in a case of sporadic hyperekplexia. Clin Neuropsycharmacol 2000;23:161-3.
10. Praveen V, Patole SK, Whitehall JS. Hyperekplexia in neonates. Postgrad Med J 2001;77:570-2.
11. Tijsen MA, Rees MI. Hyperekplexia. In: Pagon RA, Adam MP, Ardinger HH, Bird TD, Dolan CR, Fong CT, et al., editors. Gene Reviews. Seattle (WA): University of Washington, Seattle. Available from: http://www.ncbi.nlm.nih.gov/pubmed/20301437. [Last accessed on 2014 Mar 01; Last updated on 2012 Oct 04].