

# Case Report

## An Unusual Startling

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### 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 startle response to external stimuli; this can be disabling, 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 startle 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.<sup>[1]</sup> It is mostly genetic (startle disease).<sup>[2]</sup> However acquired cases have been documented.<sup>[3]</sup> Its pathogenesis is linked with deficiency of glycine, an inhibitory central neurotransmitter.<sup>[4]</sup> 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.<sup>[5]</sup> 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.

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### 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 startle. Other systemic examinations were not remarkable. The cerebrospinal spinal fluid analysis,

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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'.<sup>[6]</sup> 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."<sup>[7]</sup> However in 1966 Suhren *et al.*, evaluated members of a Dutch pedigree with excessive startle reflexes and called the disorder hyperekplexia.<sup>[8]</sup> 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.*<sup>[9]</sup> 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.<sup>[10]</sup> Clonazepam is the drug of choice in

treating hyperekplexia, while levetiracetem has shown promises in managing this disorder. However, other sedative-hypnotics like carbamazepine, phenytoin, diazepam, valproate, and phenobarbital have been used.<sup>[11]</sup> 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.

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