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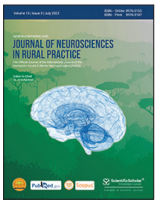

Letter to Editor

A child with global developmental delay and excessive startles: Never overlook the family history

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Dear Editor,

A 3-year-old boy, 2nd born to a non-consanguineously married couple, had a smooth perinatal transition presented with global delay and excessive startles. The child never attained neck holding, cannot sit with support or rollover, or hold things in hand. However, the child could respond to sound/light and vocalizes. He also had a history of intermittent twisting posturing in all four limbs. The child had microcephaly, normal hair, and short stubby fingers on examination. Visual fixation was present. The child had an excessive startle with loss of habituation. Fundus was normal. There was the variable tone and generalized dystonia. Family history gave us an important clue in the form of self-mutilation in a maternal cousin.

Investigations revealed normal neuroimaging and hyperuricemia (uric acid – 9 mg/dl). Diagnosis of Lesch-Nyhan syndrome (LNS) was established by genetic analysis, which revealed a pathogenic mutation in exon 7 of the hypoxanthine-guanine phosphoribosyltransferase (*HPRT1*) gene.

LNS is a rare X-linked disorder caused by an inborn deficiency of HPRT enzyme.[1] It can have varied neurological abnormalities such as intellectual disability, involuntary movements, and predominantly choreoathetosis. Children with this condition also show unusual, compulsive, and aggressive behavior, which can be self-injurious with the resultant partial or total destruction of oral and perioral tissues and fingers, which remain the hallmark of this deadly disease.[2] However, these classical features were conspicuously absent in the index child.

Allopurinol has a role in reducing serum uric acid levels, thereby preventing gout and nephrolithiasis and symptomatic treatment for neurological manifestations to be given. Gabapentin has a beneficial role in treating self-mutilating behavior.[3] Dopamine replacement therapy and deep brain stimulation in globus pallidus are in the pipeline but still not proven.[4,5]

A high index of suspicion is required to diagnose rare entities like LNS. Detailed history, including family history and methodological examination, assists in arriving at an exact diagnosis in resource-limited settings. Neurometabolic syndrome may not present with classic symptoms; rather, the presentation may be misleading, like in our index case, the child presented with excessive startles. A detailed 3-degree pedigree chart does help in establishing the clinical diagnosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

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