Case Report

Self-injurious Behavior in a Young Child with Lesch-Nyhan Syndrome

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ABSTRACT

Lesch-Nyhan syndrome (LNS) is a rare inherited disorder caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyl transferase-1. Few reports on behavioral aspects especially self-injurious behavior in LNS patients are available. We report a case of LNS in an 8-year-old male child, who presented with characteristic self-injurious behavior.

Key words: *Behavior, Lesch-Nyhan syndrome, self-injurious*

INTRODUCTION

Lesch-Nyhan syndrome (LNS), is a rare inherited disorder caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyl transferase-1 (HGPRT-1), produced by mutations in the HPRT gene located on the X-chromosome. LNS affects about one in 380,000 live births. The disease process mainly affects the male child, and females are asymptomatic carriers. HGPRT deficiency leads to the characteristic triad of features:

i. Hyperuricemia,
ii. Spectrum of neurological dysfunctions, and
iii. Cognitive and behavioral disturbances.[1]

Hyperuricemia may lead obstructive uropathy and gouty arthritis.[2] Neurological manifestations include dystonia, choreoathetosis, opisthotonos, and sometimes ballismus.[3] Cognitive and behavioral disturbances present as mental retardation, aggressive, and self-injurious behavior (SIB). Increased plasma uric acid levels damage the basal ganglia leading to cognitive disturbances, neurologic dysfunction, and SIB.[4]

SIB is the most salient feature of this disorder and is apparent in 85% of affected males.[5] It usually emerges by the age of 3 years. The self-injury begins with biting of the lips, cheeks, and tongue; as the disease progresses, affected individuals frequently develop finger biting and head banging and poking fingers into the eyes.[6] The self-injury can increase during times of stress. The severity of self-injury did not change over years. Age of onset was a predictor of outcome. The earlier the age of onset the worse the self-injury eventually became.[7] This SIB continues, resulting in partial or total destruction of the perioral tissues, especially the

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lower lip and to a lesser extent, the upper lip. Partial or complete amputation of the fingers, nose, and tongue are also common.

Few reports on behavioral aspects especially self-injurious behavior in LNS patients are available.[7,8] We report a case of LNS in an 8-year-old male child, who presented with characteristic SIB.

CASE REPORT

Mr A, an 8-year-old male child with uneventful birth history without a family history of neurological and psychiatric illness presented with complaints of SIB, irritability, stubbornness, emotional lability from last 5 years. From the age of 3 years, he gradually started biting of the lips, cheeks, and tongue. Over the next 6 months, such type of biting behavior of the patient gradually increased. He started banging his head over the wall and floor, repeatedly punching, and scratching his face. This type of self-injurious behavior has led to swelling and inflammation of the lips, cheeks, and tongue. After 1 year, the patient started repeatedly poking fingers into his eyes and picking the upper and lower eyelids around outer canthus of both eyes. This has led to swelling of the periorbital soft tissues of both eyes. The family members of the patient have to restrain him, to avoid his SIB. The patient was unable to do his daily activities. Developmental history revealed delayed gross motor and language milestones. Physical examination revealed periorbital edema, facial edema, abrasion of cheeks, ulceration of cheeks, swelling and inflammation of both lips and tongue [Figure 1]. Along with this contusion was present over the scalp in the frontal area. Laboratory investigations including hemogram, thyroid function tests, liver function tests, computed tomography scan brain were within normal limits except serum uric acid which was increased to 4.78 mg/dl (normal limits 3.50-7.20 mg/dl). A pediatric and skin opinion was sought for his above problems. For his skin lesions broad spectrum antibiotic (syrup amoxicillin trihydrate/potassium clavulanate; 125 mg/5 ml) 3 times daily along with regular dressings were started. The child was prescribed oral allopurinol (4 mg/kg) to suppress uric acid production. He was started on tablet risperidone 1 mg/day in divided doses along with tablet clonazepam 1 mg/day. To prevent further SIB, parents were taught various techniques of physical restraints, i.e., dental guard and hand gloves. With this treatment, his SIB and irritability came under control after 4 months and currently the patient is being followed up.

Figure 1: Facial injury of the child

DISCUSSION

SIB in LNS can be differentiated from SIB associated with autism or developmental disabilities by its sudden and more severe onset. Allopurinol will lower uric acid levels to normal but does not affect the behavioral aspects of the disease. Treatment options for management of SIB are physical restraints, behavioral, and pharmacological treatment (benzodiazepines) but the success rate is limited.[7] Physical restraints have been the sole reliable resource for preventing SIB. Cloth body restraints, cloth mittens, and plastic arm splints have all been useful in reducing the frequency of injury. Drastic measures, such as removal of the teeth or provision of tooth guards, are often taken to prevent further tissue damage. However, in many cases even with physical restraints, self-injury continues. Our patient's self-injurious behavior controlled with both pharmacotherapy and physical restraints. The aggressive behavior usually wanes in patients older than 10-12 years of age. Patients with LNS usually die in their late second or third decade. The cause of death is renal failure or infections that are a result of decrease in lymphocyte and immunoglobulin G levels.[9]

Diagnosis of LNS is usually missed in children with self-injurious behavior due its relative rarity. Hence, we should be careful while dealing with self-injurious behavior in children and a multidisciplinary approach is required for their management.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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