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Case Report

A rare case of Lesch-Nyhan syndrome in adulthood

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ABSTRACT

Lesch-Nyhan syndrome (LNS) is a rare X-linked recessive disorder characterized by hyperuricemia, neurodevelopmental delay, self-mutilation, and gouty arthritis. We present a unique case of a 19-year-old male with a delayed diagnosis of LNS, highlighting the diagnostic challenges associated with adult presentations of this syndrome. The patient exhibited classic symptoms, including severe hyperuricemia, bilateral renal stones, gouty tophi on palms, polyarthralgia, self-injurious behavior, low IQ, delayed motor development, and ataxia. The diagnosis was confirmed through clinical evaluation and biochemical testing, revealing significantly elevated serum uric acid levels. Management included dietary modifications, aggressive hydration, and uricosuric medications, leading to symptomatic improvement. This case underscores the importance of considering LNS in patients with unexplained hyperuricemia and neurobehavioral symptoms. Early diagnosis and appropriate management can improve the quality of life and reduce complications.

Keywords: Lesch-Nyhan syndrome, Hyperuricemia, Gouty arthritis, Self-mutilation

INTRODUCTION

Lesch-Nyhan syndrome (LNS) is a rare hereditary disorder caused by a deficiency of hypoxanthine-guanine phosphoribosyltransferase (HPRT), an enzyme involved in purine metabolism. The condition was first described by Lesch and Nyhan in 1964. It is an X-linked recessive disorder, predominantly affecting males. The hallmark features of LNS include hyperuricemia, neurological abnormalities, self-mutilation, and behavioral disturbances. Several studies have reported that the prevalence of LNS is approximately 1 in 380,000 live births, though it may be underreported due to diagnostic challenges. Clinically reported studies have shown that patients with LNS typically present during early childhood with delayed developmental milestones, hypotonia, and failure to thrive.[1] Despite the classic pediatric presentation, there have been few studies documenting cases with delayed diagnosis in adulthood. These cases often involve patients who present with complications like gouty arthritis, renal stones, and neuropsychiatric symptoms. Such cases pose diagnostic challenges and require a multidisciplinary approach to management.[2]

CASE REPORT

A 19-year-old male presented to our clinic with complaints of generalized weakness, recurrent joint pain, and self-injurious behavior. The patient had a history of delayed motor development, poor academic performance, and behavioral disturbances, including self-mutilation of lips and fingers. Clinical examination revealed multiple gouty tophi on the palms and fingers, along with bilateral renal stones confirmed through imaging. Laboratory investigations showed elevated serum uric acid levels of 12.4 mg/dl. Renal function tests were within normal limits. Radiological investigations, including X-rays, confirmed the presence of bilateral renal stones and joint erosions characteristic of gout. Genetic testing revealed a mutation in the hypoxanthine-guanine phosphoribosyltransferase-1 (HPRT1) gene, confirming the diagnosis of LNS.[3] The patient was started on a low-purine diet, aggressive

hydration, and uricosuric medications, including allopurinol. Behavioral therapy was initiated to address self-injurious behavior. Over the course of six months, the patient showed significant improvement in joint symptoms and a reduction in self-mutilation episodes.

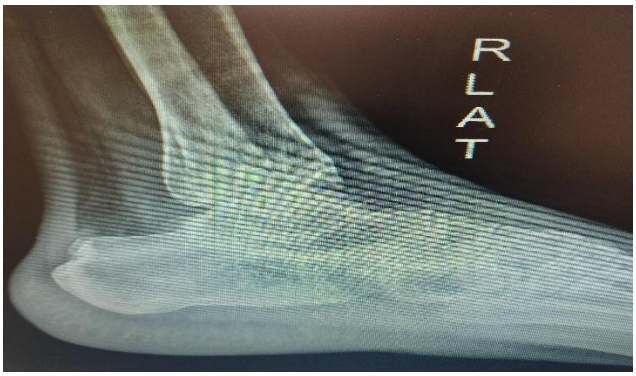

Figure 1:

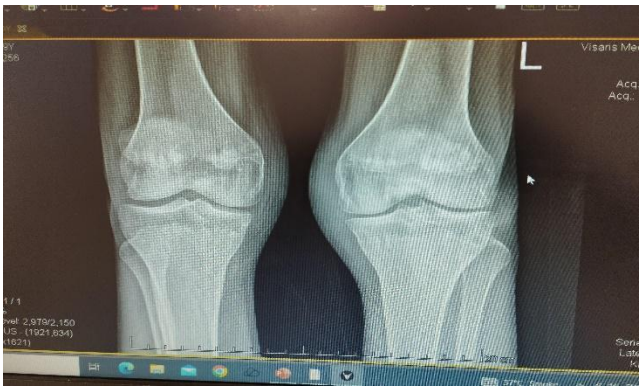

Figure 2:

DISCUSSION

LNS is a rare and debilitating disorder with significant morbidity. The delayed diagnosis in adulthood, as seen in this case, is uncommon and poses unique challenges. Hyperuricemia is a hallmark feature of LNS and can lead to gouty arthritis and nephrolithiasis if left untreated. Our patient exhibited classic signs of LNS, including severe hyperuricemia, self-mutilation, and neurological deficits.[4] The self-injurious behavior observed in LNS is a distinctive feature and is thought to result from a dysfunction in the dopaminergic pathways. Previous studies have suggested that dopamine dysregulation may play a role in the neuropsychiatric manifestations of LNS. Management of LNS involves a multidisciplinary approach, focusing on controlling hyperuricemia and addressing neurobehavioral issues.[5] In comparison to previously reported cases, our patient responded well to dietary modifications and uricosuric medications, demonstrating the importance of early intervention. Several studies have highlighted the role of allopurinol in reducing uric acid levels and preventing complications. However, behavioral management remains a challenge, requiring continuous psychological support and therapy.[6]

CONCLUSION

This case report highlights a rare presentation of LNS in adulthood, emphasizing the importance of considering LNS in patients with unexplained hyperuricemia and neurobehavioral symptoms. Early diagnosis and a multidisciplinary management approach are crucial in improving outcomes for these patients.

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