Lesch–Nyhan Syndrome: In a Yemani female child

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Abstract

Lesch–Nyhan syndrome usually affects young children in which there is a compulsive tendency of self-mutilation. There are learning difficulties along with and involuntary movements. In this case there was mental retardation along with osteomyelitis of the left big toe. It is a hereditary disorder and affecting mostly male children but this is a rare case affecting a female child.

Keywords: Lesch–Nyhan syndrome, osteomyelitis, fractures, behavioral disorder

Introduction

Lesch–Nyhan syndrome is said to be a disease showing neurological and behavioral disorders. Head banging and biting is a common manifestation in this disease (1). Chewing of lips and fingertips is also common, causing self-mutilation. Mechanism of production of these symptoms is not known till date (2). It is also known as Lesh-Nyhan disease, hypoxanthine-guanine phosphoribosyl transferase deficiency, HGPRT deficiency and HPRT deficiency (3).

In this disorder Purine metabolism is affected due to lack of an enzyme called hypoxanthine guanine phosphoribosyl transferase 1 (HPRT_1) (2) and simultaneously there is increased production of uric acid and this causes gouty arthritis (1). Biopsy of skin may reveal a decreased level of HGP enzyme (2). Nephro-calcinosis and renal failure has also been reported in patients suffering from this syndrome (4).

It is an inherited disorder linked with the X chromosome and affecting mostly males (2).

Hypoxanthine-guanine phosphoribosyl transferase (HGprt) essay strongly correlates to six biomarkers (AICAR mono- and tri-phosphate, nicotinamide, nicotinic acid, ATP and Succinyl-AMP) in red blood cells, is an effective for diagnosing Lesch–Nyhan Syndrome (5).

Prenatal gene mutation diagnosis of HPRT1 for causing Lesch–Nyhan Syndrome is accurate and helpful to prevent the birth of a baby suffering from Lesch–Nyhan Syndrome (6).

Accurate carrier detection by molecular diagnosis for genetic heterogeneity of HPRT1 gene which causes HGprt deficiency so that genetic counselling can be provided in time (7).

Case report

There was a 3 years Yemini female child with a history of pain and swelling of big toe since 15 days. There was a previous history of pain and swelling of

big toe which subsided with conservative treatment about 3 months back. In Jordan she had been diagnosed with Lesch–Nyhan syndrome.

On examination she was mildly mentally retarded and her facial features supported the diagnosis. Her incisors teeth had been removed to prevent mutilation of her own body parts by herself. Facial grimacing and repetitive movements of her arms and legs, similar to those seen in Huntington's chorea, were present.

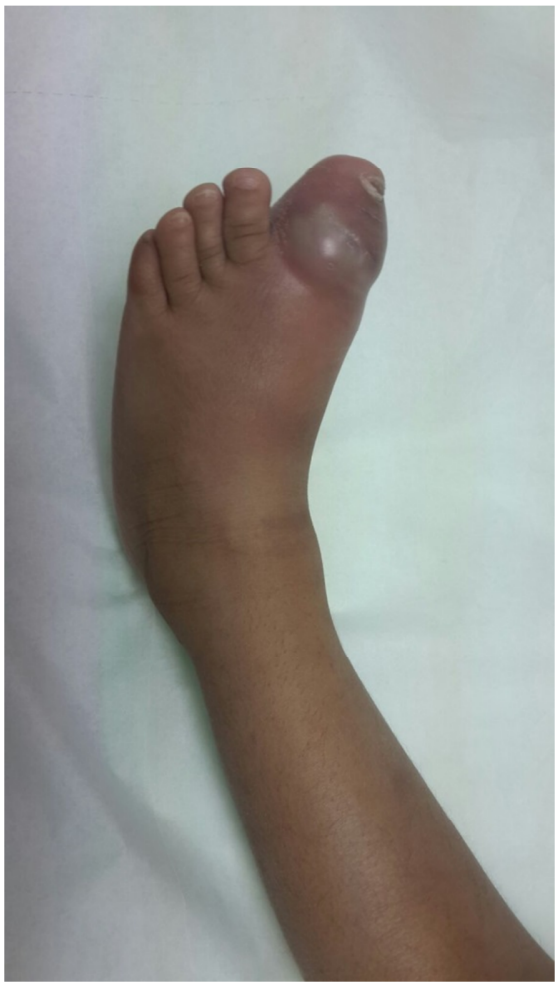

Fig. 1: Showing collection of pus and swelling and redness around big toe

WBC count and ESR was raised. She had hyper uric acid levels for which she had been given allopurinol and levels came to normal after treatment. X ray shows osteomyelitis of the proximal phalanx of big toe. MRSA methicillin resistant staph aureus growth was seen on culture on the sample taken during operation.

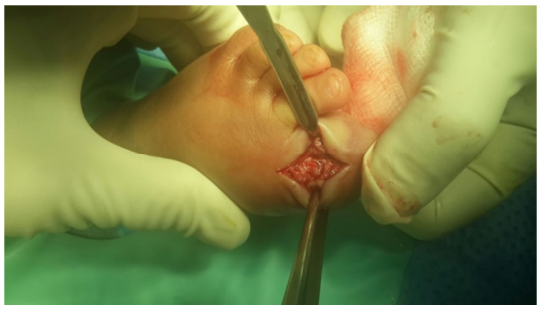

Fig. 2: Incision in the swelling showing bony changes of osteomyelitis and granulation tissue during surgery

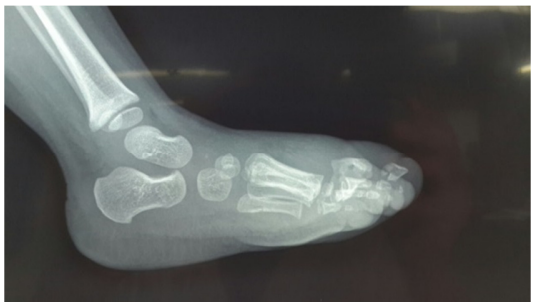

Fig. 3: X-ray image showing osteomyelitic changes in the proximal phalanx of big toe

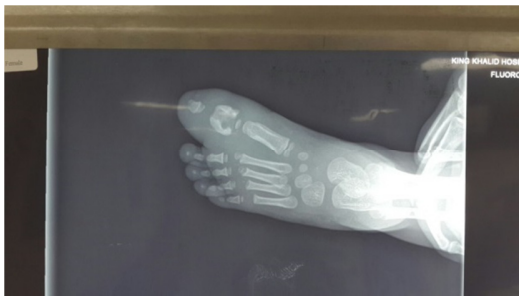

Fig. 4: X-ray image showing osteomyelitic changes in the proximal phalanx of big toe

Conclusion

Lesch–Nyhan syndrome was seen in a young female child showing mental retardation and osteomyelitis with raised WBC and ESR. Uric acid levels were raised which came down with treatment. The condition also improved by treatment with

allopurinol in an Indian child suffering from same disease (8). This case has been presented so that people can be aware of this rare disorder in a female child, which usually affects males, so that prenatal diagnosis of this syndrome should be made even in pregnancy with girl fetuses to prevent the birth of girls suffering from this syndrome. This is rare presentation affecting a female child.

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