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A case of Lesch-Nyhan syndrome in an adult presented with acute kidney injury

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Case Study : Lesch-Nyhan syndrome (LNS) is a rare X-linked disorder caused by a deficiency of the enzyme hypoxanthine-guanine phosphoribosyl transferase (HPRT) and underlying HPRT gene mutations. We report a case of LNS in a 26-year-old man, who presented with acute kidney injury and excessive hyperuricemia. He had a 7-year history of gout. He had tophectomy for gouty arthritis on left ankle 3 months ago and was taking aceclofenac (NSAID) from then on. He had a 3-year history of dystonia. Mutation analysis and enzyme assay revealed a mutation of exon 3 of the HPRT gene (c.295T>G (p.Phe99Val)) and total deficient HPRT confirming the diagnosis of LNS. His renal function and serum uric acid level improved after hydration and allopurinol treatment.

Keywords : Lesch-Nyhan syndrome, acute kidney injury