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CORRESPONDENCE

Uncommon Cause of Arthritis: Basic Skills Good Enough for Unusual Diagnosis

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To the Editor: X-linked agammaglobulinemia (XLA) can be associated with autoimmune manifestations like arthritis [1]. Herein we present a case referred to as juvenile idiopathic arthritis (JIA), however meticulous history and clinical evaluation suggested the underlying diagnosis and subsequent line of management. A 4-y-old boy presented with fever and arthritis of the bilateral hip, knee, and ankle joints for two months making him non-ambulatory. Further history elicited recurrent infections, including TB meningitis, ear discharges and gastroenteritis, in the past and a history of male sibling who succumbed to repeated infections at three years of age.

On examination failure to thrive and arthritis of bilateral hips, knees and ankles were noted. Investigations showed hemoglobin of 7.9 g/dl, total leucocyte count of 12,690/mm^3, differential count of 35% neutrophils and 53% lymphocytes, HIV screening was negative. Serum immunoglobulin levels revealed pan hypogammaglobulinemia with IgG: 1.25 g/L and IgM 0.20 g/L, IgA: 0.42 g/L. The peripheral blood flow-cytometry showed low B cell markers (CD 19; patient 0.59%, reference range:14–33%), whilst T cell subsets were normal. The above clinical and laboratory features were in keeping with X–linked agammaglobulinemia (XLA), and the same was reaffirmed by a pathogenic hemizygous mutation in exon 14 of the *BTK* gene *i.e.*, c,1181C>A on the X chromosome; the child was administered analgesics and a short course of steroids for inflammatory arthritis. For XLA, IVIG 400 mg/kg^2 intravenously every 4 weekly along with cotrimoxazole prophylaxis, was commenced [1]. At the last follow-up at 3 months the arthritis has resolved and there were no further infections.

XLA can present with autoimmune manifestations like arthritis and can masquerade as JIA. In various series, 7–45.9% of children with XLA are reported to have arthritis [2], which is usually similar to inflammatory arthritis on presentation but resolve with IVIG unlike other inflammatory arthritis which requires disease modifying antirheumatic drugs (DMARDs). The index case emphasises the importance of meticulous clinical evaluation to curb the hyposkilia in the current era [3].

Declarations

Conflict of Interest None.

References

1. Freeman AF, Holland SM. Antimicrobial prophylaxis for primary immunodeficiencies. Curr Opin Allergy Clin Immunol. 2009;9:525–30.
2. Qing-Qi R, Ya-Wen L, Huan C, et al. Retrospective study of 98 patients with X-linked agammaglobulinemia complicated with arthritis. Clin Rheumatol. 2022;41:1889–97.
3. Adam KAR. Hyposkillia: A sign of sagging medical profession-A pediatric perspective. Sudan J Paediatr. 2014;14:11–20.

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