CASE REPORT

An additional X chromosome

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A 35-year-old man presented with symmetrical inflammatory polyarthritis of the small and large joints and associated early-morning stiffness. He was assessed as having features in keeping with Klinefelter’s syndrome. It is evident from the literature that there is a relationship between Klinefelter’s syndrome and developing rheumatic conditions.

Chromosomal studies were in keeping with Klinefelter’s syndrome. A skin biopsy showed mild hyperkeratosis, and a dual-energy X-ray absorptiometry (DEXA) scan revealed osteopenia.

Discussion

We reassessed the patient as having Klinefelter’s syndrome with SLE complicated by lupus panniculitis, although we could not prove it on skin biopsy owing to an inadequate specimen. His arthritis was treated with methotrexate and we also administered chloroquine, vitamin D and calcium. In view of the osteopenia we referred the patient to an endocrinologist for testosterone replacement, to which he responded well.

Klinefelter’s syndrome was named after Henry F Klinefelter, who practised rheumatology at Johns Hopkins University School of Medicine, Baltimore, Maryland, USA. He was a medical student in the 1940s, when he contributed to the description of the syndrome.1 Most patients with Klinefelter’s do not seek medical attention and are therefore not diagnosed.1 It is evident from the literature that there is a relationship between Klinefelter’s syndrome and developing rheumatic conditions. Rovensky et al.2 highlighted the strong association between Klinefelter’s syndrome and rheumatoid arthritis, juvenile idiopathic arthritis, psoriatic arthritis, dermatomyositis/polymyositis, SLE, systemic sclerosis, ankylosing spondylitis, primary biliary cirrhosis and mixed connective tissue disease.

References