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IMAGES IN PAEDIATRICS

Pachydermoperiostosis beyond juvenile idiopathic arthritis*

Paquidermoperiostosis más allá de la artritis idiopática juvenil

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We present the case of a male patient aged 17 years who had been followed from age 2 years for oligoarticular juvenile idiopathic arthritis (bilateral knee involvement, with negative antinuclear antibodies, rheumatoid factor and HLA-B27). He responded well to intra-articular corticosteroid injections and remained asymptomatic without treatment between the ages of 5 and 13 years. From age 13, he developed recurrent bilateral knee effusion (Fig. 1) that was refractory to corticosteroid injections, nonsteroidal anti-inflammatory drugs (NSAIDs), (ibuprofen and naproxen) and methotrexate, requiring repeated arthrocentesis.

At age 14, digital clubbing became evident (Fig. 2), followed by progressive skin thickening with deep frontal folds (cutis verticis gyrata) (Fig. 3). Magnetic resonance imaging of both knees showed large bilateral joint effusion with heterogeneous synovial thickening, and the synovial biopsy revealed nonspecific chronic synovitis. Genetic testing identified a heterozygous variant in the *SLCO2A1* gene,

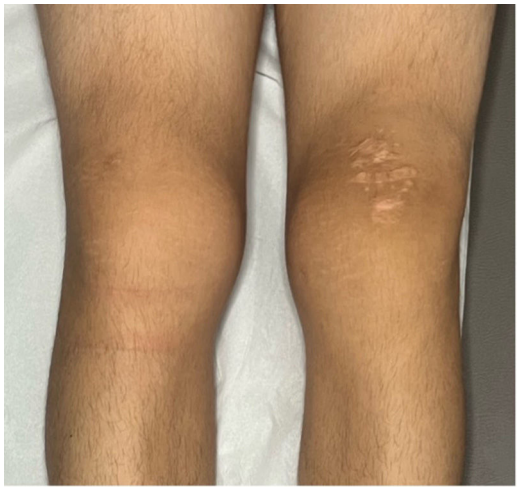

Figure 1 Bilateral knee swelling.

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Figure 2 Clubbing of fingers and toes.

Figure 3 Frontal pachydermia with deep frontal folds.

confirming the diagnosis of pachydermoperiostosis. Treatment with Etoricoxib was initiated, with a partial response. At age 17, he developed inflammatory arthritis of both ankles, refractory to NSAIDs; adalimumab was initiated, resulting in marked clinical improvement.

Pachydermoperiostosis is a rare genetic disorder characterized by digital clubbing, periostosis and pachydermia.[1,2] This case illustrates the co-occurrence of juvenile idiopathic arthritis and pachydermoperiostosis, two distinct entities rarely reported together, which may delay diagnosis and treatment.[3]

Informed consent

We obtained informed consent from the legal guardian of the patient for the publication of this case.

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Declaration of competing interest

The authors declare having no conflict of interests.

References

1. Lu Q, Xu Y, Zhang Z, Li S, Zhang Z. Primary hypertrophic osteoarthropathy: genetics, clinical features and management. Front Endocrinol (Lausanne). 2023;14:1235040, http://dx.doi.org/10.3389/fendo.2023.1235040.
2. Shakya P, Pokhrel KN, Mlunde LB, Tan S, Ota E, Niizeki H. Effectiveness of non-steroidal anti-inflammatory drugs among patients with primary hypertrophic osteoarthropathy: a systematic review. J Dermatol Sci. 2018;90(1):21–6, http://dx.doi.org/10.1016/j.jdermsci.2017.12.012.
3. Stoian M, Dumitrescu AM, Ciofu C, Gavrila B. From juvenile idiopathic arthritis to pachydermoperiostosis: a journey to an unexpected rare diagnosis. J Clin Med. 2025;15(3):956, http://dx.doi.org/10.3390/jcm15030956.