<https://doi.org/10.47660/CBR.2020.16963>

PACHYDERMOPERIOSTOSIS: A RARE AND STIGMATIZING DISEASE

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BACKGROUND

Hypertrophic osteoarthropathy (OAH), also known as pachydermoperiostosis, is a rare genetic syndrome with probable autosomal dominant inheritance. The first cases were reported by Friedreich in 1868. In 1935, Toutaine et al. individualized pachydermoperiostosis as the primary form of hypertrophic osteoarthropathy. It represents approximately 3 to 5% of cases of hypertrophic osteoarthropathy, with varied clinical presentations, with skin thickening, clubbing and symmetrical periostosis. Its etiology is unknown. Classically, it affects men, starts in adolescence and progresses slowly, with self-limitation of its course after a few years. We report the case of a patient with clinical changes and family history for pachydermoperiostosis.

CASE REPORT

Male patient, 41 years old, white, starts with arthralgia and symmetrical arthritis, mainly of small joints of the hands and feet at 17 years old, considering his main clinical complaint, for limiting his daily activities. He has a family history of a brother diagnosed with pachydermoperiostosis. Clinical examination revealed skin thickening of the hands and feet, skin thickening on the face with oily skin, hypertrophy of the hands and feet, with increased soft tissue and joint volume and digital clubbing. The radiological study of hands and feet showed symmetrical and bilateral changes, with increased soft tissue, clubbing, cortical thickening and periosteal reaction in metacarpophalanges and metatarsophalanges. As well as cortical thickening with periosteal reaction at the end of long bones, irregularities in the tuberosities and bone remodeling – findings compatible with hypertrophic osteoarthropathy.

CONCLUSION

The diagnosis of primary OAH is made based on clinical and radiological findings, consisting of periosteal proliferation with progressive, symmetrical and bilateral bone neoformation in long bones, most evident in tendon and ligament insertions. The observation of clubbing and radiographic periostosis strongly suggest its diagnosis. There is no specific treatment for the primary form of OAH. For the control of joint symptoms, nonhormonal anti-inflammatory drugs, systemic corticosteroids and colchicine appear to be useful. Knowledge of pachydermoperiostosis is important, as it is a stigmatizing syndrome and with a decrease in patients' quality of life. The diagnosis, in its varied presentations, and regular follow-up with a rheumatologist, are necessary.