Clinical Image

Tricho-rhino-phalangeal syndrome - a case report

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Nicolas Balbinot Panteliades, Roberto Haendchen Bento, Paola Nassar de Aquino
Centro Universitário de Brusque – Brusque (SC), Brazil

Correspondence: Nicolas Balbinot Panteliades, Centro Universitário de Brusque – Brusque (SC), Brazil, Email nicolasb.panteliade@gmail.com

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Background

Tricho-rhino-phalangeal syndrome (TRPS) is a rare genetic disorder caused by mutations in the TRPS1 gene on chromosome 8. The syndrome has three types: distinctive hair, craniofacial and skeletal abnormalities. Type I is the most frequent. Multiple bony exostoses and mental disability characterize type II. Type III is a more severe type I form associated with short stature.

Case report

A 44-year-old woman with complaints of chronic arthralgia affecting hands, feet, hips and knees. No morning stiffness or other inflammatory signs. She reported deformities of fingers and toes since childhood, with chronic mechanical pain, which had been worsening over time. In the last few years, she became progressively impaired, with decreased mobility and loss of functional performance. Physical examination showed a long, narrow face with a bulbous nose (also called a pear-shaped nose); sparse and brittle hair; broad and thick eyebrows, especially at the median one-third; and a thin upper lip with a long flat philtrum. The musculoskeletal examination showed deformities of the fingers and toes, mainly at the proximal interphalangeal joints, in a symmetrical fashion, with ulnar deviation of the fingers. No signs of synovitis were noted. At the lower limbs, bilateral genu varus and flatfoot, with pain and limitation of movement of the hips. Laboratory tests were unimportant: erythrocyte sedimentation rate was 35 mm/h, C-reactive protein 4,6 mg/L (reference value <5 mg/L), ANA 1:80 speckled, rheumatoid factor negative. Blood count and creatinine were normal. Radiography of the hands showed shortening of metacarpals and phalanges and significant bone dysmorphism at the proximal interphalangeal joints, with the typical cone-shaped epiphyses. The ultrasound examination of the hands showed no signs of joint effusion or synovitis. The diagnosis was made based on extremely specific and distinctive physical and radiographic characteristics in this case. The patient received guidance and support about the diagnosis and was treated with symptomatic medications.

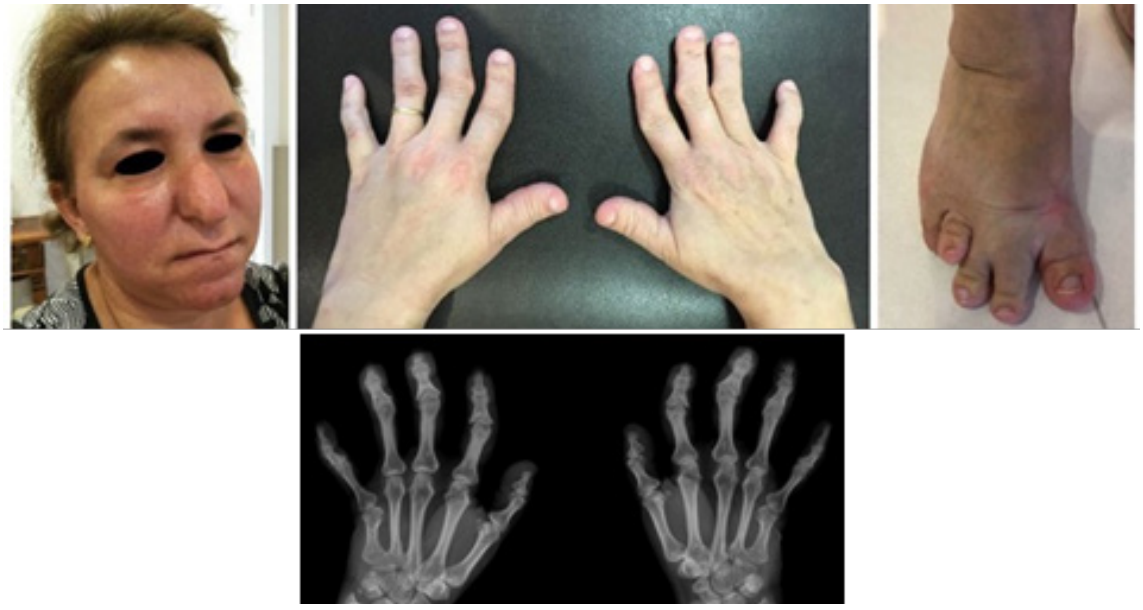

Figure 1 Pear-shaped nose and hair rarefaction. Joint deformities in the feet and hands. Hand x-ray showing shortening of metacarpals and phalanges, with cone-shaped epiphyses.

Conclusion

TRPS is a rare disorder with prominent musculoskeletal findings that can mimic other rheumatic diseases such as rheumatoid arthritis and juvenile idiopathic arthritis. This report aims to disseminate knowledge and awareness about the syndrome.

Acknowledgments

None.

Conflicts of interest

The authors declare no conflicts of interest.

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