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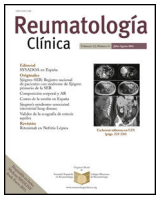

Case Report

Trichorhinophalangeal syndrome☆

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ABSTRACT

Trichorhinophalangeal syndrome I (TPRSI) has an autosomal dominant inheritance; the proportion of de novo cases is unknown.[1] It is characterised by unique facial features, bulbous nose, flat and elongated nasolabial furrow, thin hair and slow growth. Skeletal abnormalities that include short phalanges and metacarpals -brachydactyly-, cone-shaped epiphyses, hip dysplasia and short stature.[1–3]

Síndrome tricorinofalángico

Palabras clave:
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RESUMEN

El síndrome tricorinofalángico I (TPRSI) tiene una herencia autosómica dominante, la proporción de casos "de novo" es desconocida.[1] Se caracteriza por rasgos faciales únicos, nariz de extremo bulboso, surco nasolabial plano y alargado, cabello escaso y de crecimiento lento. Anomalías esqueléticas que incluyen falanges y metacarpianos cortos -braquidactilia-, epífisis en forma de cono, displasia de cadera y estatura baja.[1–3] Presentamos una familia con 7 miembros afectos de TRPSI.

Introduction

Trichorhinophalangeal syndromes (TRPS) are rare congenital syndromes that are caused by a chromosome alteration. These include TPRS I—or Giedion—(caused by a heterocygotic pathogenic variant in TRPSI) and TPRS III—or Langer-Giedion — a phenotypic variant of the previous type—, and TPRS II —or Sugio-Kajii— (caused by a deletion in genes adjacent to TPRSI, RAD21 and EXTI). They are characterised by skeletal anomalies, delayed growth, distinctive facial features, ectodermic alterations—thin hair that is depigmented and hardly grows, dystrophic nails and small breasts—and in TRPS II multiple osteochondromas, as well as varying degrees of intellectual disability. TRPS I or Giedion (MIM 190350) is a malformative syndrome characterised by facial and skeletal alterations. It is inherited with a pattern of autosomal dominance with high penetrance and variable expressivity, associated with alterations in chromosome 8q24.1.It is characterised by unique facial characteristics, a nose with a bulbous tip, a flat and prolonged nasolabial groove, little hair and slow growth. The skeletal anomalies include short phalanges and metacarpals – brachydactily –, cone-shaped epiphyses, hip dysplasia and short height.[1–3]

Clinical case

We present the cases of a family with 7 affected members in 4 generations. The cases were diagnosed based on a female patient with the facial and skeletal characteristics of the syndrome (Figs. 1 and 2). Only 3 members of the family with similar physical characteristics are being monitored by rheumatology, and their history does not mention urethral, endocrine or renal alteration or

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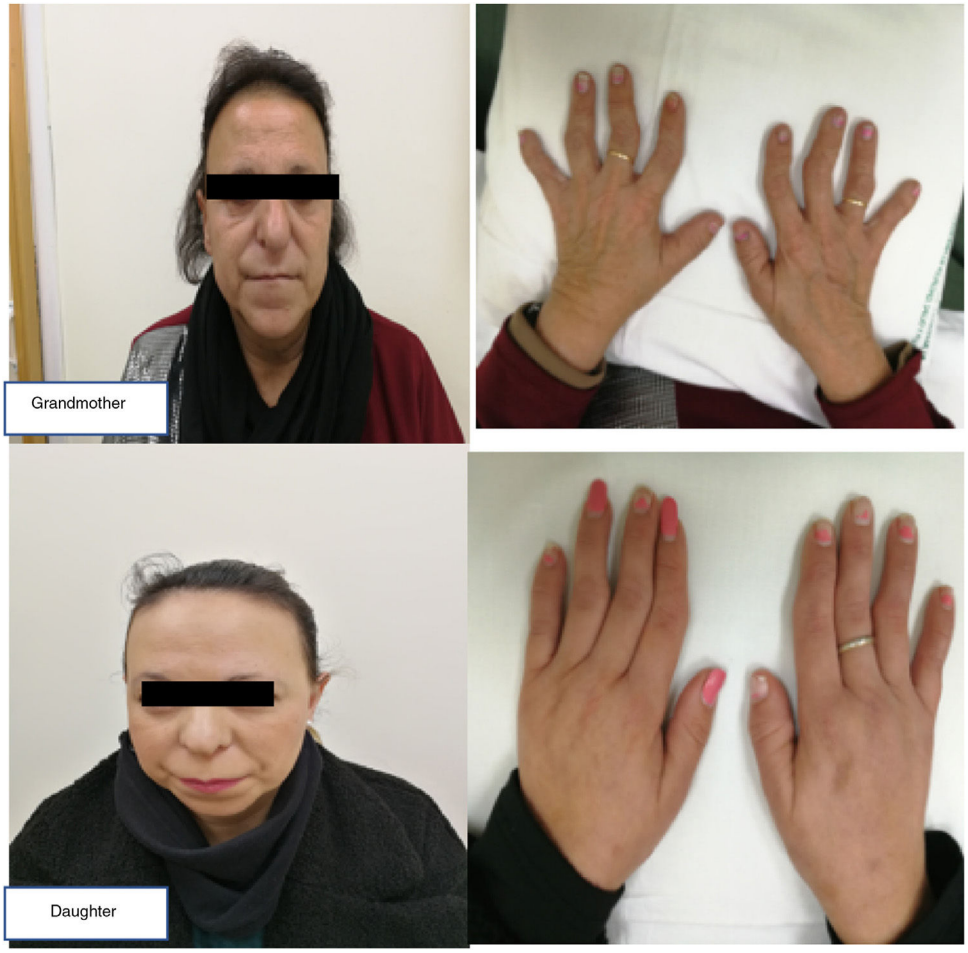

Figure 1. Sparse hair and bulbous nose.

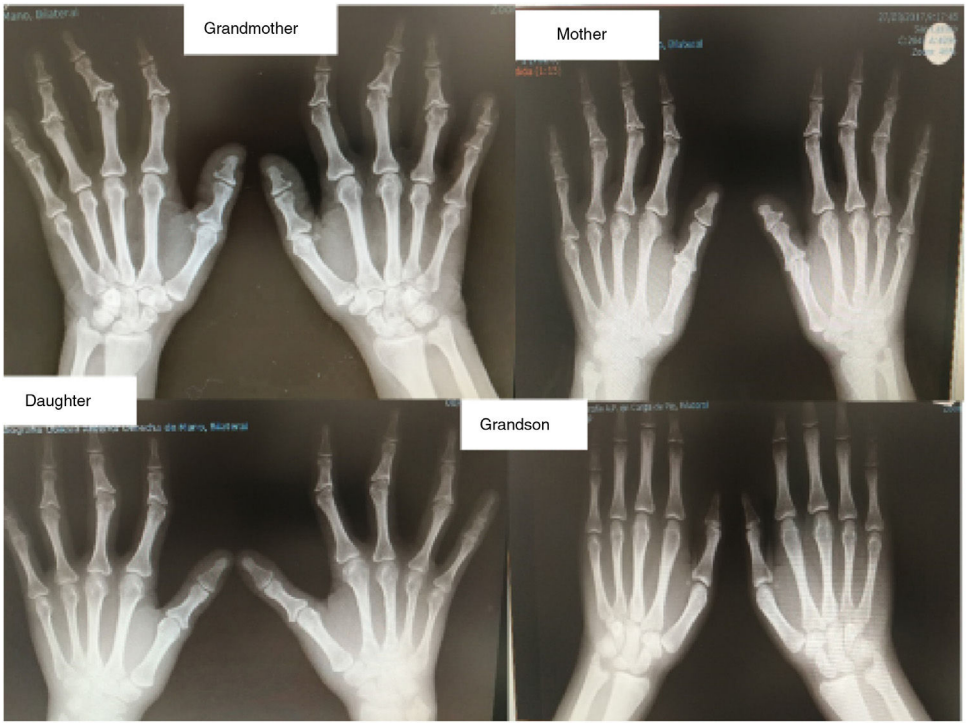

Figure 2. Cone-shaped epiphysis.

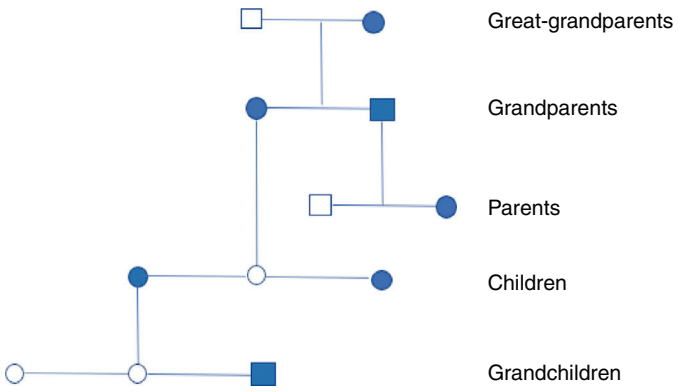

Figure 3. Family tree: circles are women, squares are men, figures shaded in blue are affected.

disease; they all have sparse hair as dermatological involvement. The youngest member has slight scoliosis and *pectus excavatum* as a differentiating trait from the others. Our patient was previously diagnosed with rheumatoid arthritis and psoriatic arthritis. She consulted due to deformity and discomfort in the interphalangeal joints of both hands. The heterozygous variant c.2894 G > A (p.Arg965His) was detected in the TPRSI gene. This is known to be a pathogenic variant in the said syndrome. There is a 50% risk of transmission to descendents[4] (Fig. 3).

Discussion

The name TRPS covers 3 rare genetic diseases that are characterised by craniofacial and skeletal anomalies. The TPRSI (OMIM 604386) gene in chromosome 8q23.1-q24.1 is associated with the development and differentiation of bones, kidneys and hair follicles.[1,2,5] Diagnosis of this rare syndrome is based on the physical and radiological characteristics of the patients, and it is confirmed by the detection of a pathogenic genetic variant. Detection in an adult, as in this case, makes it possible to avoid errors in treatment, which in TRPS is exclusively supportive, with analgesia and orthopaedic measures when necessary.[1–3]

Conclusions

TRPS are rare genetic diseases, and awareness of them makes it possible to avoid diagnostic errors and subsequent possibly harmful treatments.

Conflict of interests

The authors have no conflict of interests to declare.

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