

# Trichorhinophalangeal syndrome

## Abstract

We describe a trichorhinophalangeal syndrome in a 9 year-old female, showing thinning hair, rarefaction of the lateral eyebrow, pear-shaped nose, long and flat philtrum, thin upper lip, receding chin, short stature and brachydactyly. In spite of being rare, trichorhinophalangeal syndrome may be more frequent than postulate in the literature, since less expressive phenotype cases may be more difficult to diagnose. Therefore, knowledge of this condition becomes important to allow a proper genetic counseling and follow-up.

**Keywords:** brachydactyly, alopecia, hair, scalp, syndrome

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## Case report

FEMALE patient, 9 years old, with thin, non-growing hair. It presents diffuse rarefaction of the hair, with a pattern of non-cicatricial alopecia, fine and short hair, rarefaction of the distal third of the eyebrow, bulbous nose and enlargement of the nasolabial filter, fine upper lip, micrognathism, short stature, brachydactyly, curved feet and shortening of some metacarpals (Figure 1) (Figure 2). Intellectual development was normal and absence of similar cases in the family.



**Figure 1** Non scarring hair loss, fine and short hair, rarefaction of the distal third of the eyebrow, bulbous nose and enlargement of the nasolabial filter, fine upper lip, micrognathism.



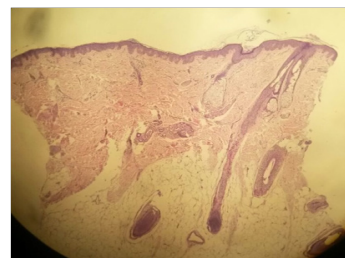
**Figure 2** Brachydactyly, curved feet and shortening of some metacarpals and metatarsals.

The laboratory tests requested were hemogram, electrolytes, albumin, glycemia, thyrotrophic hormone, parathyroid hormone, vitamin D and zinc; all normal. Radiographs showed bone changes of the phalanges, with cone-shaped epiphyses (Figure 3) and advanced bone age in 2 years. Trichogram with 44% anagen, 53% telogen, 1% catagen and 2% dystrophic anagen.

Hystopathology with rarefied pilo-sebaceous follicles, most of the bulbs anchored in the subcutaneous tissue, besides some smaller diameter follicles in the dermis (Figure 4).



**Figure 3** Radiographs showed bone changes of the phalanges, with cone-shaped epiphyses.



**Figure 4** Hystopathology with rarefied pilo-sebaceous follicles, most of the bulbs anchored in the subcutaneous tissue, besides some smaller diameter follicles in the dermis.

## Discussion

Giedion, in 1966, was the first to describe the trichorhinophalangeal syndrome (TRPS) and to propose the denomination that persists to the present. Three variants are classified, classified as TRPS type I, characterized clinically by the association of fine, sparse and slow-growing hairs, pear-shaped nose, elongated filter, thin upper lip and bone alterations, particularly cone-shaped epiphyses in the fingers

**Table I** Phenotypic alterations associated with TRPS

Trichorhinophalangeal syndrome type I	Trichorhinophalangeal syndrome type II (Langer-Giedion)	Trichorhinophalangeal syndrome type III (Sugio-Kajii)
Sparse and fragile scalp hair	Sparse scalp hair	Same characteristics of type I
Pilli torti	Prominent pear-shaped nose	Accentuation of short stature
Ticorrexo nodosa	Pear-shaped phalangeal epiphyses (x-rays)	Increased sharp shortening of the phalanges, metacarpals and metatarsals
Prominent pear-shaped nose	Multiple exostoses	
Broad and raised nasal bridge	Prominent ears	
Pear-shaped phalangeal epiphyses (x-rays)	Microcephaly	
Curved fingers and toes	Short stature	
Brachydactyly	Hyperextensible joints	
Brittle and thin nails	Redundant skin in childhood	
Short stature	Increased number of nevi	
Winged scapulae	Mental retardation	
Deep voice		
Degenerative diseases of the hips in youth		

Diagnosis should be suspected through the presence of specific clinical features of the syndrome, and may be confirmed by clinical evaluation, detailed history and radiological study. Analysis of molecular genetics may demonstrate mutations of the TRPS1 gene, located on the long arm of chromosome 8 (8q24.1).<sup>1,2</sup> Our case is classified as type I, considering that there is no accentuation of shortening of all phalanges, metacarpals and metatarsals, nor of short stature.

Of the possible clinical alterations observed in the TRPS-I, alopecia may be the most frequent complaint.<sup>2</sup> Findings of the trichogram are not homogeneous, with some studies demonstrating pattern of androgenetic alopecia combined with diffuse alopecia and others with no definite pattern.<sup>2</sup> Some studies also mention advanced bone age in radiological investigation.

Histopathology shows a reduction in the number of follicles, with the majority of bulbs anchored in the subcutaneous tissue, in addition to smaller diameter anagen and absence of inflammatory infiltrate.<sup>1</sup>

Differential diagnosis of TRPS-I is made with the other variants of the syndrome itself, with androgenetic alopecia of early onset (in the milder phenotypes) and with others that include alopecia and structural abnormalities of the nose or osteoarticular abnormalities.

## Conclusion

The treatment is symptomatic and directed to the symptoms of each individual affected.<sup>1</sup> Although considered to be rare (with a prevalence of 0.2-1per 100,000),<sup>6</sup> it is postulated that TRPS is more frequent than has been described, since less expressive phenotypes

(distinctive feature of the syndrome).<sup>1-5</sup> TRPS type II, which is associated with microcephaly and mental retardation, cartilaginous exostoses and others more severe osteoarticular manifestations; and TRPS type III, a type I proximal variant, which is phenotypically differentiated by the accentuation of shortening of the phalanges, metacarpals and metatarsals, and of short stature.<sup>2</sup> Other phenotypic alterations may be associated and are described in Table 1.

may go unnoticed. Therefore, knowledge of the syndrome is important in order to initiate adequate genetic counseling and follow-up.<sup>2</sup>

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## Conflicts of interest

The author declares no conflict of interest.

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