CASE REPORT

Crooked fingers and sparse hair: an interesting case of trichorhinophalangeal syndrome type 1

Ramakrishna Narayanan,¹ Srinivasa Chennareddy²

¹Department of Radiodiagnosis, Kasturba Medical College, Manipal University, Udipi, Karnataka, India

²Department of Rheumatology, Nizam's Institute of Medical Sciences, Hyderabad, Telangana, India

Correspondence to
Ramakrishna Narayanan,
drkrish@gmail.com

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SUMMARY

Trichorhinophalangeal syndrome type 1 is a rare skeletal dysplasia of autosomal-dominant inheritance due to defects in the TRPS-1 gene. The syndrome is characterised by sparse slow-growing hair, a bulbous pear-shaped nose, cone-shaped epiphyses and deformities of the interphalangeal joints resembling those in rheumatoid arthritis. We present a case of trichorhinophalangeal syndrome in a 23-year-old man who presented with symmetrical painless progressive deformity of the fingers in both hands.

BACKGROUND

Trichorhinophalangeal syndrome (TRPS) is a rare and complex genetic disorder with clinical features that may mimic other skeletal dysplasias, Perthes disease or even inflammatory arthropathy such as rheumatoid arthritis. We present this case to demonstrate the importance of recognising its clinical features, which, in conjunction with the characteristic radiological abnormalities, will help arrive at the diagnosis. Early diagnosis allows adequate follow-up and timely therapeutic intervention.

CASE PRESENTATION

A 23-year-old male software professional presented with gradually progressive painless deformity of the fingers in both his hands, causing difficulty in typing on a keyboard.

Clinical examination of the patient showed symmetrical flexion deformities, and ulnar deviation of second and fourth digits at the proximal interphalangeal joint of both hands (figure 1). There was no limitation of motion. His fingernails appeared brittle. In view of the symmetric small joint involvement with progressive deformity, initially, a possibility of inflammatory arthritis such as rheumatoid arthritis, was considered.

General physical examination revealed peculiar craniofacial abnormalities such as sparse and fine hair, broad forehead, rarefaction of lateral eyebrows and a bulbous, pear-shaped nose. Examination of the patient's feet showed short first, fourth and fifth toes (figure 2).

The patient was born to non-consanguineous parents and had an unremarkable personal and family history.

Based on these clinical findings the possibility of underlying skeletal dysplasia was considered and further radiographic evaluation by a skeletal survey was advised. Radiographs of hands, feet, dorsolumbar spine, pelvis and skull were performed.

INVESTIGATIONS

Radiograph of both hands (figure 3) showed cone-shaped epiphyses of the middle phalanges of second to fourth digits of both hands with deviation of the phalangeal axis. Bone density was well maintained with no juxta-articular osteopenia or articular erosions. Similar cone-shaped epiphyses were also seen in the proximal phalanx of the great toe of both feet (figure 4) with short first, fourth and fifth digits.

Radiographs of the spine, pelvis and skull were normal.

Based on these typical clinical and radiological features, the diagnosis of sporadic case of TRPS type 1 was made and orthopaedic consultation was sought.

DIFFERENTIAL DIAGNOSIS

Even though there was symmetrical involvement of small joints, rheumatoid arthritis was ruled out, as the deformities, though progressive, were painless with no restriction of mobility. There was also no radiological evidence of juxta-articular osteopenia or articular erosions.

Other skeletal dysplasias such as oral-facial-digital syndrome and chondroectodermal dysplasia were considered among the differentials as they can also present with alopecia, structural craniofacial abnormalities and cone-shaped epiphyses mimicking TRPS type 1. However, oral-facial-digital syndrome is associated with cleft lip, cleft palate, flat nasal bridge and hypertelorism, while chondroectodermal dysplasia is associated with postaxial polydactyly and congenital heart defects, which were not seen in our patient.

TREATMENT

As the deformities were progressive and interfering with the occupation of the patient, arthrodesis of the proximal interphalangeal joint of the index and middle fingers of both hands was suggested. However, the patient refused to undergo

Figure 1 Symmetrical flexion deformities, ulnar deviation of second and fourth digits of both hands at the proximal interphalangeal joint (white arrows).

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Figure 2 Frontal and lateral photographs showing sparse and fine hair, broad forehead, rarefaction of lateral eyebrows and a bulbous, pear-shaped nose.

surgical treatment. The patient was hence advised occupational therapy, mobility training and follow-up.

DISCUSSION

TRPS is a rare skeletal dysplasia of autosomal-dominant inheritance.^{1 2} It is caused by defects in the TRPS1 gene located on chromosome 8, which encodes a transcriptional repressor involved in hair development and chondrocyte modulation.¹ Three types of TRPS have been described: TRPS type I, TRPS type II and TRPS type III. In contrast to TRPS type I, patients with TRPS type II suffer from mental retardation and develop multiple exostoses.¹ TRPS type III is the rarest type, with patients having normal intelligence and severe shortening of all phalanges and metacarpals.¹

TRPS I was first described by Giedion.² Characteristic clinical features include trichological abnormalities such as sparse slow-growing hair.^{1 2} Sparseness is caused by thinning of individual

Figure 3 Anteroposterior radiograph of both hands showing cone-shaped epiphyses in the middle phalanx of second to fourth digits in both hands (white arrows) with deviation of the phalangeal axis. No evidence of erosions.

Figure 4 Anteroposterior radiograph of both feet showing symmetrical shortening of first, fourth and fifth digits (white arrows) with cone-shaped epiphyses of the proximal phalanx of the great toe (dashed arrows).

hair follicles and can affect the eyebrows, eyelashes, beard, axillary and pubic hair.¹ Facial abnormalities include bulbous pear-shaped nose, long and flat philtrum and high frontal hairline.¹ Other abnormalities involving nails, such as leuconychia and brittle nails have also been described.³

However, these clinical abnormalities often go unnoticed by the patient and most seek medical attention for the progressive deformities in the hands, as occurred in our case. In fact, a review of case reports shows that this is the most common presenting symptom.¹⁻³

Radiology plays a critical adjunctive role in the diagnosis. The most diagnostic and constant radiological abnormality involves the epiphyses of middle phalanges, which are cone-shaped and result in deformity at the proximal interphalangeal joints.^{1 2} Other tubular bones such as those of the feet can also be similarly affected, resulting in brachydactyly and clinodactyly with radial or ulnar deviation of digits.²

TRPS can also involve the hip joints, resulting in malformations such as coxa plana, coxa magna or coxa vara, and in children it may mimic Perthes disease.¹

Learning points

- ▶ Trichorhinophalangeal syndrome (TRPS) is a rare skeletal dysplasia with autosomal-dominant inheritance due to defects in the TRPS-1 gene.
- ▶ The syndrome has a characteristic triad of sparse slow-growing hair, bulbous pear-shaped nose and cone-shaped epiphyses.
- ▶ TRPS can mimic other skeletal dysplasias, Perthes disease or rheumatoid arthritis, but a combination of characteristic clinical and radiological features can help arrive at the correct diagnosis.
- ▶ Early diagnosis allows adequate follow-up and timely therapeutic intervention.

Competing interests None.

Patient consent Obtained.

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