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TRICHO - RHINO - PHALANGEAL SYNDROME TYPE I*

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Tricho-rhino-phalangeal syndrome type I (TRPS I) is characterized by a bulbous nose, sparse hair and epiphyseal coning. Autosomal dominant and recessive transmission are suggested. The presence of cone-shaped epiphyses, the major complaint of patients due to swelling over the phalangeal joints, requires differential diagnosis among various syndromes. This paper, describing a ten-year-old girl with TRPS I, aims to bring the features of the syndrome to medical attention. *Key words: tricho-rhino-phalangeal syndrome, cone-shaped epiphysis.*

Tricho-rhino-phalangeal syndrome type I (TRPS I) is characterized by sparse, fragile scalp hair, bulbous nose and radiological abnormalities of the hands, mainly epiphyseal coning[1]. Unusual facies and hair may be detectable from birth, but usually children are first brought to medical attention because of swelling over the phalangeal joints. This report presents a 10-year-old girl who had the characteristic features of TRPS I.

Case Report

A 10-year-old girl was admitted to our clinic because of a one-year history of painless swelling of her fingers. Her history was otherwise unremarkable, except that her parents had first-degree consanguinity. There were no similar symptoms among relatives or siblings. Her height was 131 cm (10^{th}-25^{th} percentile). She had sparse and fine hair, bulbous nose, prominent ears, a long philtrum and micrognathia (Figs. 1a, b). A thin upper lip covering the lower, a high-arched palate and malpositioned teeth were noted. Widely positioned nipples were also detected (Fig. 2), along with broadening and slight flexion of the proximal interphalangeal joints and deviation of the phalangeal axis, with no limitation of motion (Fig. 3). She had fragile and brittle toenails, hypoplasia of the fourth and fifth toes and a short first toe as well as cutaneous syndactyly between the second and third toes (Fig. 4). her mental ability was not impaired.

X-ray examination of the hands revealed cone-shaped epiphyses in the proximal and middle phalanges of the second, third and fifth digits, in the proximal and distal phalanges of the first digit and in the middle phalanx of the fourth digit (Fig. 5).

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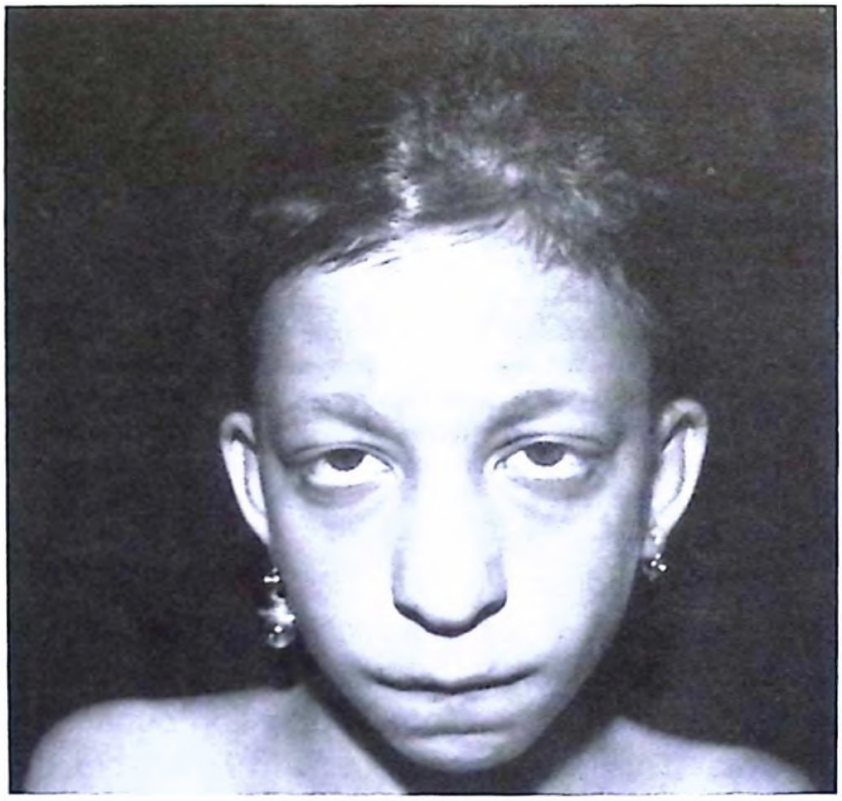
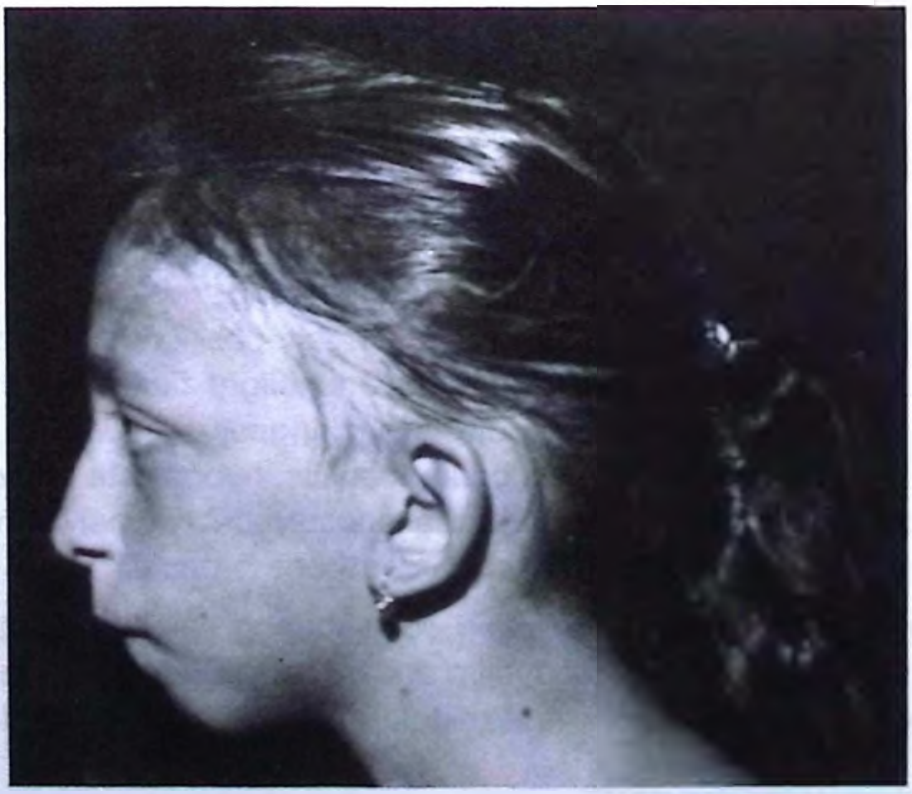


(a)



(b)

Figs. 1a, b: Sparse hair, bulbous nose, prominent and low-set ears, long philtrum, micrognathia and thin upper lip covering the lower were the prominent features.

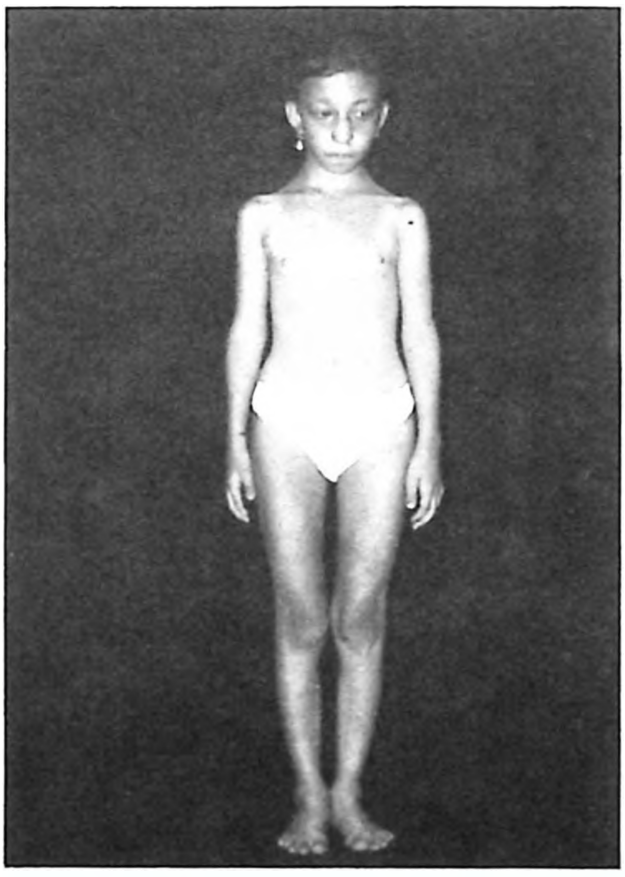


Fig. 2: Widely positioned nipples and coxa vara deformity.

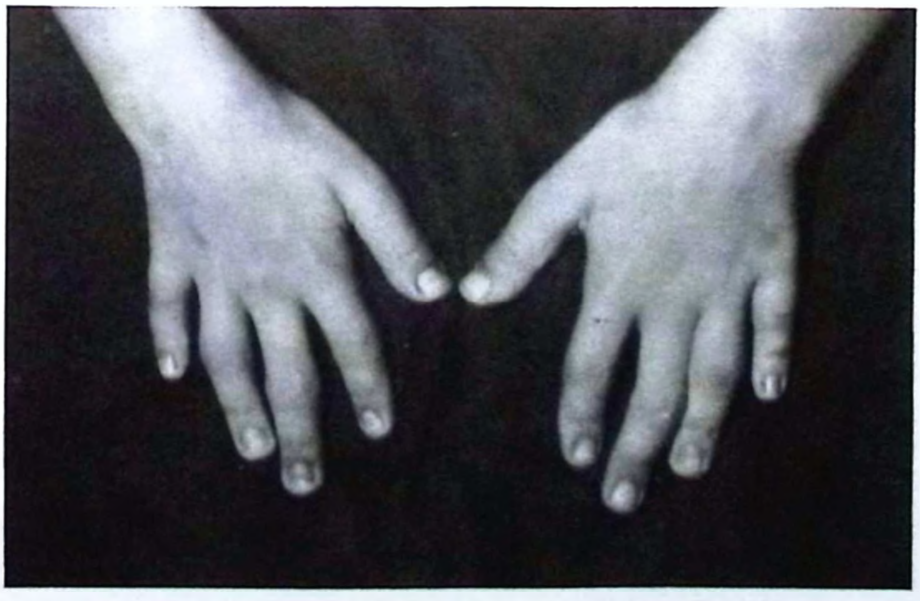


Fig. 3: Broadening and slight flexion of the proximal interphalangeal joints plus deviation of the phalangeal axis can be seen.

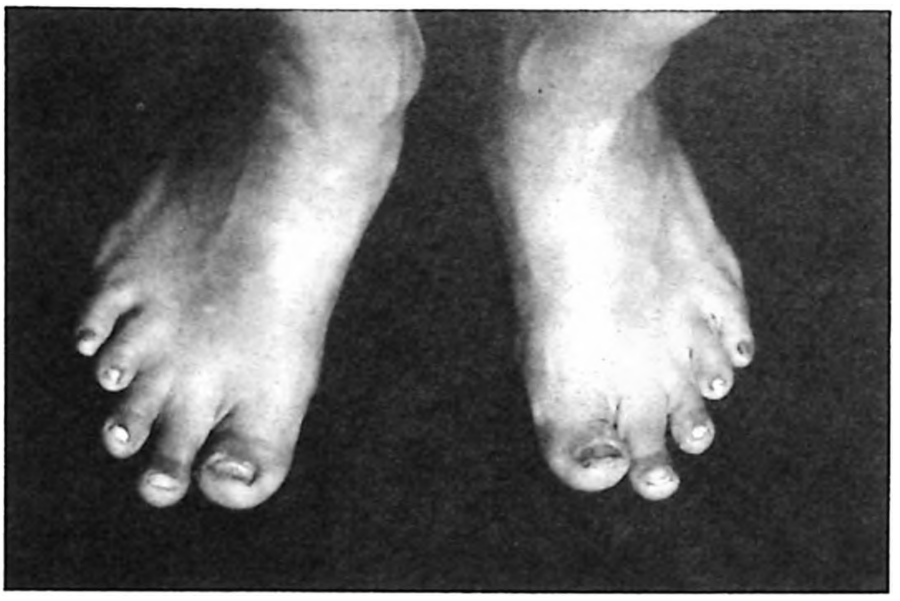


Fig. 4: Fragile and brittle toenails, hypoplasia of the first, fourth and fifth toes and cutanous syndactyly between the second and third toes.

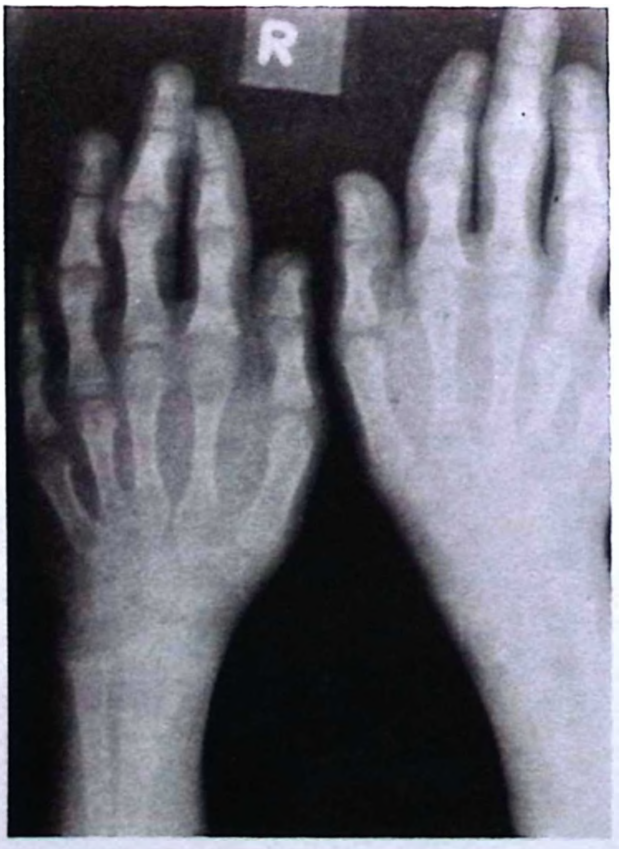


Fig. 5: Cone-shaped epiphysis in the proximal and middle phalanges of the second, third and fifth digits, in the proximal and distal phalanges of the first digit and in the middle phalanx of the fourth digit.

Discussion

The main features of TRPS I are facial dysmorphism; sparse, brittle and usually light-colored scalp hair; and short hands and feet with finger deformities. Facial dysmorphism includes a bulbous or pear-shaped nose, large and prominent ears, a long, broad philtrum, a thin upper lip and poorly developed eyebrows laterally. Skeletal changes include numerous, phalangeal, cone-shaped epiphyses of the hands, often associated with brachymetacarpism and brachymetatarsism. Clinically, cone-shaped epiphyses appear as swelling of the interphalangeal joints, mainly the proximal ones, and sometimes lead to deviation of the phalangeal axis[2]. There is little or no limitation of motion. Flattening of the capital femoral epiphyses, partial syndactyly, scoliosis, kyphosis, winged scapula, thoracic deformity, dental malocclusion, short stature and mental deficiency may sometimes accompany the main features. The nails are usually thin and brittle[1,2].

Our patient demonstrated the characteristic presentation of the syndrome. She did not have growth failure or mental deficiency. She was the only person with syndromic features in the family. Considering the parents' consanguinity, this patient represents the autosomal-recessive type of the syndrome. In TRPS I, autosomal-dominant transmission is usually suggested, but an autosomal-recessive pattern apparently occurs in affected children of normal parents, as in our case[3,4]. The chromosomal basis of the syndrome has been studied with prometaphase chromosomes; lately, small deletions in chromosome 8 (8q23, 8q24.12) have also been demonstrated and this region (8q24.12) provides the TRPS I gene[5-9].

The differential diagnosis of TRPS I should include TRPS II (Langer-Giedion syndrome) and TRPS III (Sugio-Kajii syndrome) as well as the disorders associated with cone-shaped epiphyses. In TRPS II, multiple cartilaginous exostoses of the long tubular bones, microcephalic mental retardation and short stature are the additional characteristic findings. TRPS III is characterized by the clinical features of TRPS I plus a severe form of generalized shortness of all phalanges, metacarpals and metatarsals and sometimes limitation of joint movements, pectus carinatum and thoracic scoliosis. Our patient had none of these features. Apart from normal variants, cone-shaped epiphyses can be seen in various disorders (Table I)[1-3,11]. Associated features of these syndromes are important in the differential diagnosis of TRPS I.

TRPS I is a syndrome with a normal life expectancy. Osseous changes may develop in early childhood and worsen until adolescent growth is complete. Progressive osteoarticular changes and degenerative hip disease may necessitate orthopedic care. Psychological guidance for dysmorphic features and genetic counseling may be considered.

Table I: Disorders Associated with Cone Epiphyses

Normal Variant	
Congenital Syndromes	
Achondroplasia	Dyggve-Melchior-Clausen
Acrodysostosis	Kaschin-Beck
Acromesomelic dysplasia	Orofaciodigital
Asphyxiating thoracic dysplasia	Osteoglophonic
Beckwith-Wiedemann	Otopalatodigital
Bilginturan	Peripheral dysostosis
Chondrodysplasia punctata	Pseudoachondroplastic dysplasia
Chondroectodermal dysplasia	Saldino-Mainzer
Christian	Seckel
Cleidocranial dysostosis	Trichorhinophalangeal I
Cockayne	trichorhinophalangeal II
Acquired	
Frostbite	
Infection (meningococcemia, viruses)	
Infarction (sickle cell)	
Trauma	

REFERENCES

1. Jones KL. Smith's Recognizable Patterns of Human Malformation (4th ed). Philadelphia: W.B. Saunders; 1988: 250-251.
2. Balcı S, Pırnar T, Özekmekçi F, Hanioğlu N, Say B. Koni şeklinde epifiz anomalisi gösteren bir vaka. Çocuk Sağlığı ve Hastalıkları Dergisi 1972; 5: 220-224.
3. Giedion A, Burdea M, Fruchter Z, Melani T, Trose V. Autosomal dominant transmission of the tricho-rhino-phalangeal syndrome: report of 4 unrelated families, review of 60 cases. Helv Paediat Acta 1973; 28: 249-259.
4. McKusick VA. Mendelian Inheritance in Man. A Catalog of Human Genes and Genetic disorders. Baltimore: John Hopkins University Press; 1994: 1479, 2242.
5. Yamamto Y, Oguro N, Miyao M, Yanagisawa M, Ohsawa T. Prometaphase chromosomes in the tricho-rhino-phalangeal syndrome type I. Am J Med Genet 1989; 32: 524-527.
6. Goldblatt J, Smart RD. Tricho-rhino-phalangeal syndrome without exostoses, with an interstitial deletion of 8q23. Clin Genet 1986; 29: 434-438.
7. Fryns JP, Van den Berghe H. 8q24.12 interstitial deletion in trichorhinophalangeal syndrome type I. Hum Genet 1986; 74: 188-189.
8. Buhler EM, Buhler UK, Beutler C, Fessler R. A final word on the tricho-rhino-phalangeal syndromes. Clin Genet 1987; 31: 273-275.
9. Wilkie AOM, Amberger JS, McKusick VA. A gene map of congenital malformations. J Med Genet 1994; 31: 507-517.
10. Niikawa N, Kamei T. The Sugio-Kajii syndrome: proposed tricho-rhino-phalangeal syndrome type III. Am J Med Genet 1986; 24: 759-760.
11. Poznanski AK. The Hand in Radiologic Diagnosis with Gamuts and Pattern Profiles, Vol I. Philadelphia: W.B. Saunders; 1984: 139-172.