A Case of Trichorhinophalangeal Syndrome

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Trichorhinophalangeal syndrome is a genetic disease and divided into three different types. Trichorhinophalangeal syndrome type I is characterized by alopecia, a bulbous pear-shaped nose and cone-shaped epiphyses in the hand. Diverse clinical manifestations can be observed such as short stature, mandibular abnormality, winged scapula, etc. It is inherited in an autosomal manner, and may cause grave joint abnormalities which should be corrected early in life.

We describe a 23-year-old woman with diverse clinical manifestations of trichorhinophalangeal syndrome type I, including prognathism and a winged scapula, two features which have not been previously described in the Korean literature. (Ann Dermatol 9:(4):298~301, 1997).

Key Words : Trichorhinophalangeal syndrome, Alopecia

The trichorhinophalangeal syndrome (TRPS) is a genetic disease, named by Giedion in 1966[1]. There are three different types of TRPS[2]. TRPS type I consists of sparse hair, a peculiar pear-shaped nose and skeletal abnormalities including clinobrachydactyly. TRPS type II, also called Langer-Giedion syndrome has the usual features plus multiple cartilagenous exostoses[3]. Another variant, TRPS type III is characterized by severe degrees of metacarpophalangeal shortness[4].

Only two cases of TRPS type I have been reported in Korea[5, 6]. We report herein another case with diverse clinical manifestations of TRPS type I.

CASE REPORT

A 23-year-old woman presented with scant hair since birth. She complained of fine sparse hair, short fingers with prominent joints and was bothered by her abnormal appearance. There was no history of inability to sweat. She complained of dysmenorrhea and constipation. Her mother and sister had similar features (Fig. 1). Both of them had sparse hair, angulation of the fingers. Her late mother had a history of hip joint abnormalities and her older sister had a bulbous nose and prognathism.

She was 157cm tall and had sparse thin hair on the scalp, axillae and pubic areas. Her nose had a bulbous tip and small alae, therefore contributing to a pear-shaped appearance. A well demarcated soft non-tender skin colored 1cm sized nodule on the lower border of the lip, malocclusion of the teeth and prognathism were observed (Fig. 2 a,b). The fingers were shortened, but had nearly a full range of motions except the 2nd finger, the tip of which

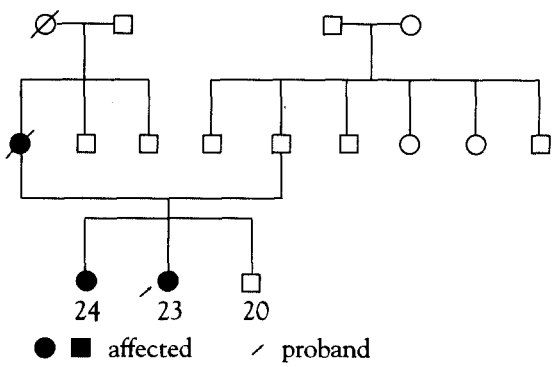

Fig. 1. Pedigree.

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This case was presented as a poster at the 48th Annual Fall Meeting of the Korean Dermatological Association on October 25, 1996.

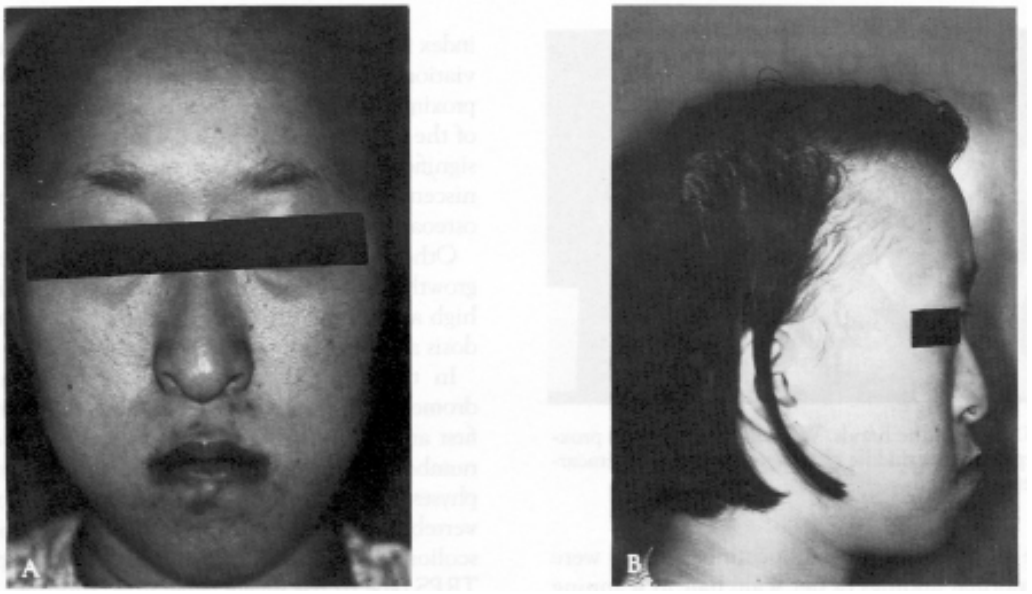

Fig. 2. A, B. Note sparsely distributed scalp hair, absent lateral part of the eyebrows, and bulbous nose.

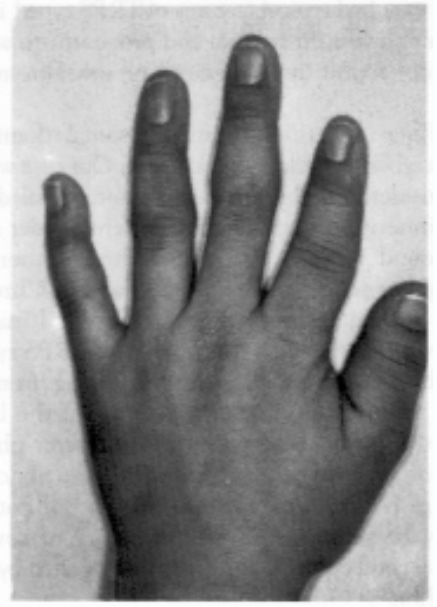

Fig. 3. Short fingers and swelling and angulation at the proximal interphalangeal joints.

could not reach the palm (Fig. 3). Short big toes (Fig. 4), pes planus and a winged scapula in the left were also observed. A gynecological examination revealed no abnormal findings.

A histopathological examination of her scalp skin only showed decreased hair follicles, and any pathological findings in the nodule on the chin were not observed. Laboratory findings including a serum testosterone level and thyroid function tests were normal. A radiological examination revealed brachymetacarpia of the bilateral 4th and 5th fingers, cone-shaped epiphyses of the middle phalanges of the hand (Fig. 5) and proximal phalanges of the foot and mild lumber scoliosis was also seen. A barium enema examination which was carried out to find the cause of her constipation showed no abnormal findings in the colon. Chromosomal analysis revealed normal karyotyping (46,XX) with no evi-

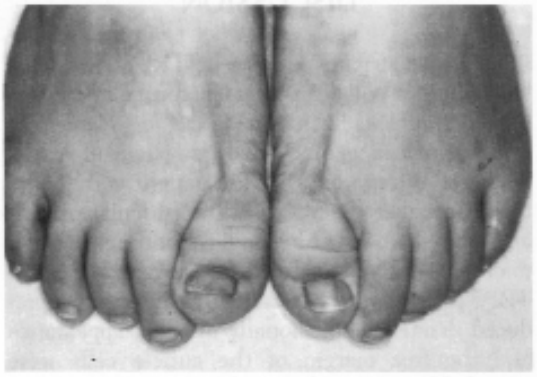

Fig. 4. Short great toes.

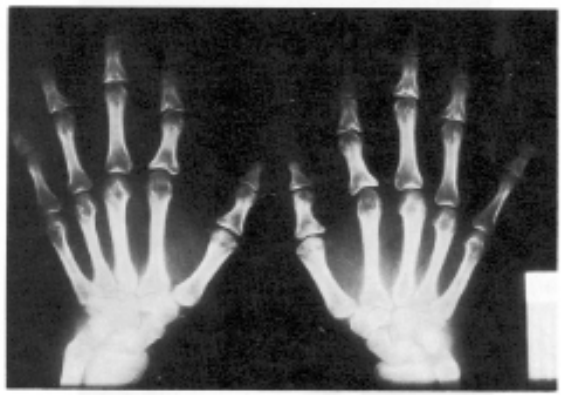

Fig. 5. X-ray of the hands. Widened cone-shaped proximal epiphyses of middle phalanges and brachymetacarpia were observed.

dence of deletions or translocations. There were no abnormal findings of her scalp hair in scanning electron microscope (SEM).

DISCUSSION

The most striking features of TRPS type I are sparse hair, a bulbous pear-shaped nose and cone-shaped epiphyses in the hands and feet. The scalp hairs are of fine texture, slowly growing and brittle. Perns, et al[7] reported that the patient with TRPS type I revealed alterations of the cuticular pattern and hair shaft structure under scanning electron microscopy. They also mentioned that the hair of TRPS patients was characterized by its extremely reduced diameter, occasionally dimpled appearance and the free margin of the cuticle cells were jagged. However, other authors[8] reported only a reduced diameter of hair. There were no abnormal findings in scanning EM of the hair in our case.

In addition to sparse hair, the characteristic facial features are a bulbous nose, a long extended philtrum, a thin upper lip, and medially thick and laterally thin or absent eyebrows. In TRPS type I, the nose has a bulbous tip, tented alae and a typical pear-like shape.

Peripheral dysostoses include cone-shaped epiphyses of the middle phalange of the hands, brachydactyly, angulation of fingers and a Perthes-like change of hip joint. Cone-shaped epiphyses alone are not characteristic of TRPS and may be found in normal individuals[9]. The angular deformities of the phalanges are most common in the index finger. The characteristic deformity is ulnar deviation of the index and middle fingers at the proximal interphalangeal joints, and radial deviation of the ring and little finger[10]. The main orthopedic significance of this syndrome is hip changes reminiscent of Legg-Calvé-Perthes disease. Premature osteoarthritis can occur in this disorder.

Other features are a short stature, delayed growth and development, a hypoplastic mandible, a high arched palate, a winged scapula[11], lumbar lordosis and congenital heart disease[12].

In the TRPS type II or Langer-Giedion syndrome, multiple exostoses develop between the first and fifth years of life and increase in size and number until maturity[3]. These occur in the metaphyses of long and short bones including the ribs and vertebra. Other features of TRPS type II include scoliosis, mental retardation and microcephaly. TRPS type III has severe generalized shortness of all phalanges, metacarpals and metatarsals in addition to the usual features of TRPS type I.

Our case had typical features of TRPS type I in addition to a winged scapula and prognathism which were not found in other cases reported from Korea.

TRPS is inherited in an autosomal dominant manner, but sporadic cases do occur. Our case was also considered to be affected by autosomal dominant inheritance because the patient's older sister was found to have TRPS and her mother was strongly suspected to be a TRPS patient. Chromosomal studies revealed that TRPS type I may be due to deletion of 8q 24.12[13], whereas TRPS type II may be caused by deletion extending from 8q 24.11 to 8q 24.13. It is considered that the larger deleted segment is responsible for severer phenotypes of TRPS type II[14]. There were no abnormal findings on chromosomal analysis in our patient. It may be possible that only the very narrow G-positive band 8q 24.12 is missing as suggested by other authors[15].

Functional hand problems in patients with TRPS type I seem to be rare and our patient did not complain of such problems. However cases with impaired hand functions have been reported[16] and degenerative changes of the hip in early childhood are also seen in this disorder. The procedures to correct or prevent the development of impaired hand and hip functions are necessary. In addition, the procedures for lengthening the affected pha-

langes with Ilizarov apparatus may be tried[5].

In summary, TRPS type I with diverse clinical manifestations has been described. We think a prompt and accurate assessment is important to give genetic and cosmetic counselling and prevent functional impairment of the affected joints.

REFERENCES

1. Giedion A. Das tricho-rhino-phalangeal syndrome. Helv Paediat Acta 21: 475-482, 1966. cited from reference 12.
2. Carrington PR, Chen H, Altick JA. Trichorhinophalangeal syndrome, type I. J Am Acad Dermatol 31:331-336, 1994.
3. Bauermeister S, Letts M. The orthopaedic manifestations of the Langer-Giedion syndrome. Orthorp Rev 21:31-35, 1992.
4. Niikawa N, Kamei T. The Sugio-Kajii syndrome, proposed tricho-rhino-phalangeal syndrome type III. Am J Med Genet 24:759-780, 1986.
5. Cho YH, Kim TH, Park KB, Song HR. A case of trichorhinophalangeal syndrome. Kor J Dermatol 32:920-925, 1994.
6. Kim HS, Sim WY, Haw CR. Trichorhinophalangeal syndrome, type I. Kor J Dermatol 35:344-349, 1997.
7. Perns EP, Peereboom-Wynia JDR, de Bruyn WC, van Joost TH, Stolz E. Clinical and scanning electron microscopic findings in a solitary case of trichorhinophalangeal syndrome type I. Acta Derm Venereol (Stockh) 64:249-253, 1984.
8. Van Neste D, Dumortier M. Tricho-rhino-phalangeal syndrome. Disturbed geometric relationships between hair matrix and dermal papilla in scalp hair bulbs. Dermatologica 165: 16-23, 1982
9. Hornsby VPL, Pratt AE. The trichorhinophalangeal syndrome. Clin Radiol 35:243-247, 1984.
10. Burgess RC. Trichorhinophalangeal syndrome. South Med J 84:1268-1270, 1991.
11. McCloud DJ, Solomon LM. The tricho-rhino-phalangeal syndrome. Br J Dermatol 96:403-407, 1977.
12. Weaver DD, Cohen MM, Smith DW. The tricho-rhino-phalangeal syndrome. J Med Genet 11:312-314, 1974.
13. Fryns JP, Van den Berghe H. 8q24.12 interstitial deletion in trichorhinophalangeal syndrome type I. Hum Genet 74:188-189, 1986.
14. Bühler EM, Malik NJ. The tricho-rhino-phalangeal syndrome(s): chromosome 8 long arm deletion: is there a shortest region of overlap between reported cases? TRP I and TRP II syndromes: are they separate entities? Am J Med Genet 19:113-119, 1984.
15. Bühler EM, Bühler UK, Beutler C, Fessler R. A final word on the tricho-rhino-phalangeal syndrome. Clin genet 31:273-275, 1987.
16. Noltorp S, Kristoffersson U, Mandahl N, Stigsson L, Svensson B, Werner C. Trichorhinophalageal syndrome type I: symptoms and signs, radiology and genetics. Ann Rheum Dis 45:31-36, 1986.