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TRICHO-RHINO-PHALANGEAL SYNDROME ASSOCIATED WITH PERTHES-DISEASE-LIKE BONE CHANGE AND SPONDYLOLISTHESIS

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Summary A case of tricho-rhino-phalangeal syndrome associated with Perthes-disease-like bone change of the unilateral hip joint is presented. This is the fourth such instance of this combination to be reported in the world literature and is sporadic as other previous cases. In addition, spondylolisthesis of the fifth lumbar spine was also observed.

INTRODUCTION

The term "tricho-rhino-phalangeal (TRP) syndrome" was first coined by Giedion in 1966. The characteristics of this syndrome are hypotrichosis of the scalp hair, a peculiar pear-shaped nose, a long philtrum, and cone-shaped epiphyses of the middle phalanges of the hands, bilaterally, resulting in deformity of the fingers. The author *et al.* reported three Japanese kindreds with the syndrome indicating autosomal dominant inheritance and presented a review of the world literature in a previous issue of this Journal (1976).

The purpose of this paper is to report briefly an additional sporadic case of TRP syndrome accompanied by unilateral Perthes-disease-like bone change of the hip joint and spondylolisthesis of the 5th lumbar spine.

CASE REPORT

Case. T. A., 12 year-old boy.

Chief complaint. Limp of the left leg and low back pain.

Present illness. For about 6 months he was complained of back pain without noticeable cause. Recently, his parents noticed limping of his left leg. The scalp hair had been markedly hypotrichotic since infancy.

Past and family histories. He was born on June 16, 1960, weighing 2,800 g at his birth. He was the product of a normal pregnancy and delivery with no history of illness or exposure to teratogenic agents. His father was 33 year-old and his

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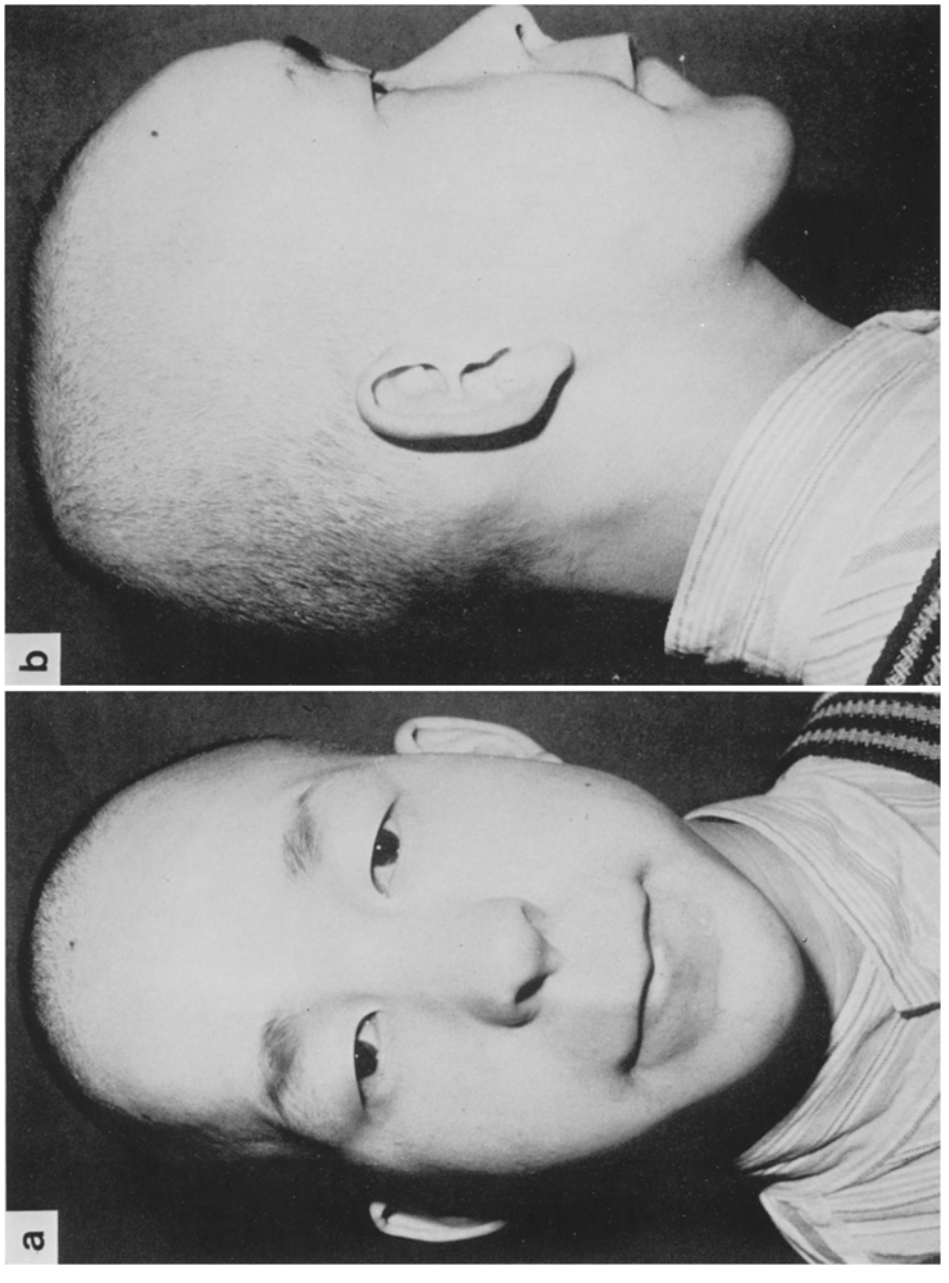

Fig. 1. External appearance of the head and face showing marked hypotrichosis of the scalp hair, a peculiar pear-shaped nose, a long philtrum and anteverted ears. a. front view, b. lateral view.

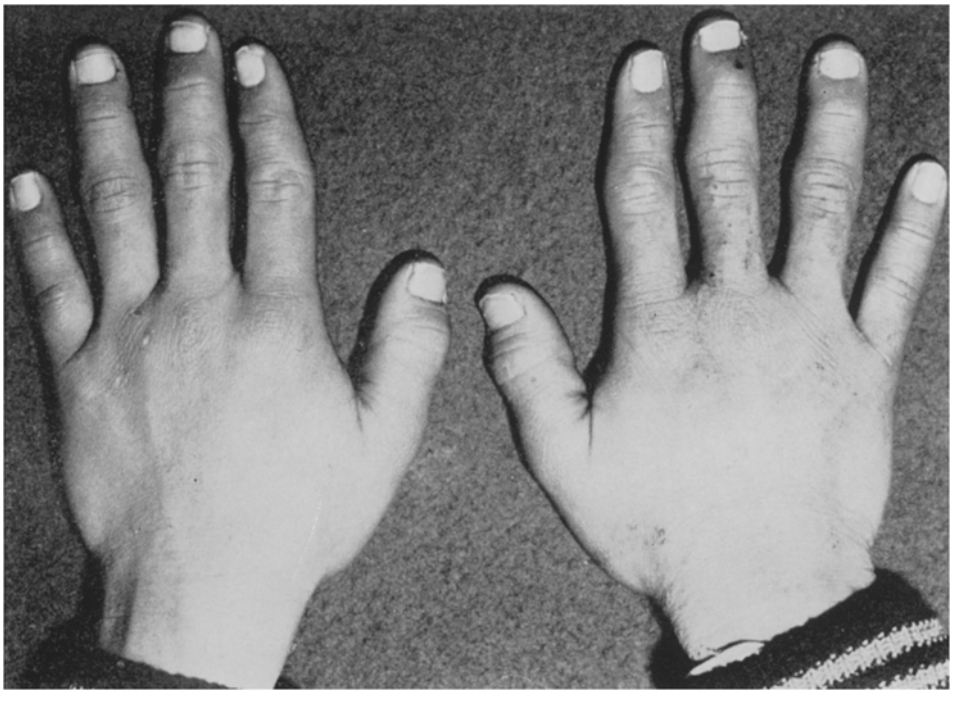

Fig. 2. External appearance of the hands showing marked shortening of the middle phalanges of the index through little fingers. The bases of the middle phalanges of the fingers are broad.

mother, 27 at the time of his birth. The mother had no history of previous abortion. Both parents, his elder brother are alive and well. There was no family history of consanguinity or malformations.

Physical examination (Nov. 9, 1973). Physical examination revealed a well nourished boy of normal stature (145.5 cm). The scalp hair was markedly sparse and of fine texture. The lateral halves of both eyebrows were also sparse. The nose was markedly prominent and bulbous with a long philtrum. The ears were moderately anteverted (Figs. 1a and 1b). Marked shortening of the middle phalanges of the index through little fingers of both hands was apparent. The bases of the middle phalanges of these fingers were broad (Fig. 2). Mobility of the fingers was normal. Internal rotation and abduction of the left hip joint were limited. Tenderness was not observed anywhere bilaterally over the hip joints. The left major trochanter was located higher than the right. The left spino-malleolar distance was 77.5 cm, while the right was 79.0 cm. Trendelenburg's sign was positive on the left. Mobility of the lumbar spine was checked as normal without causing any pain. Straight-leg-raising test was negative in both sides.

Roentgenographic findings

At his first consultation: Each hand showed cone-shaped configurations at

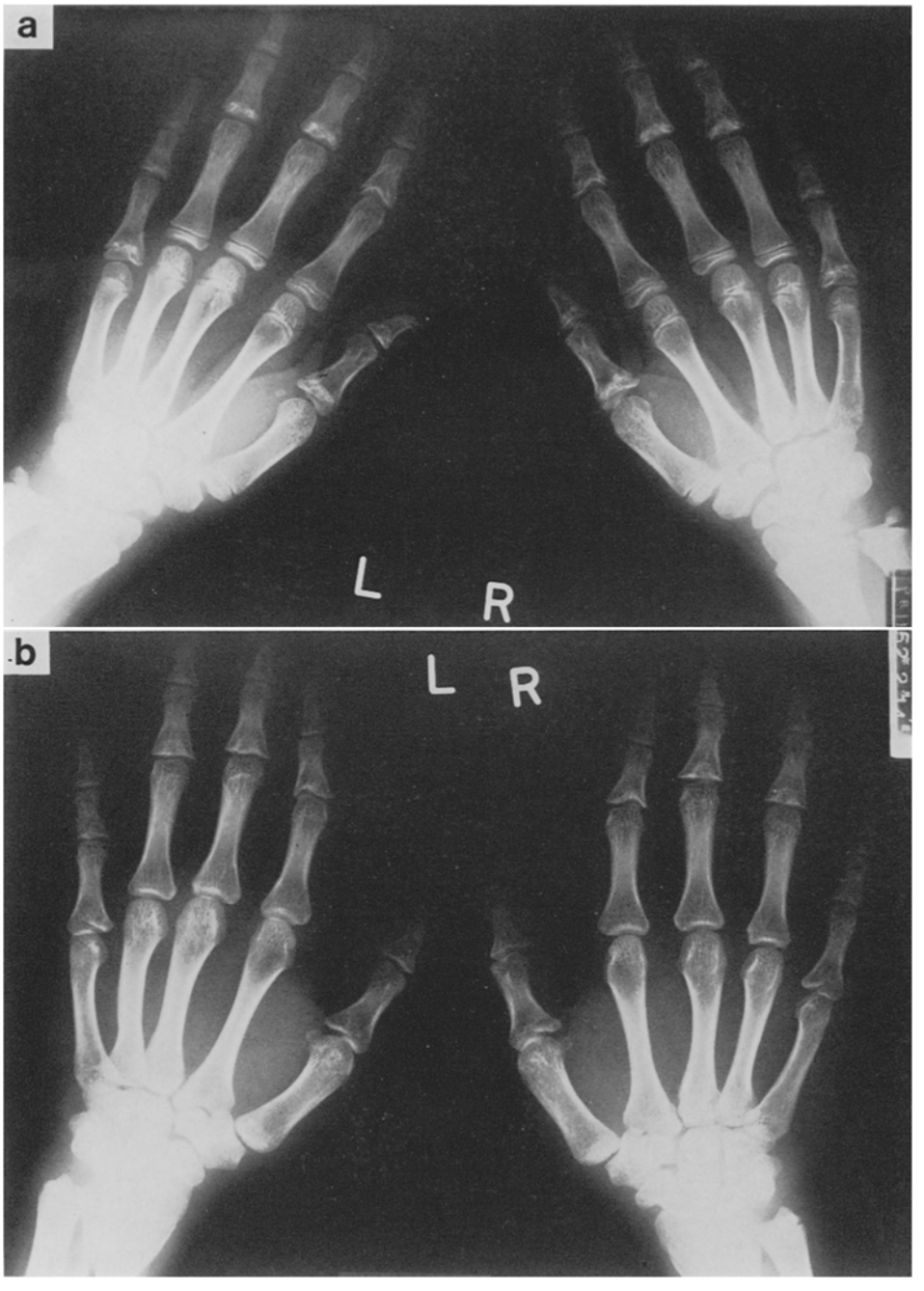

Fig. 3. Dorso-palmar roentgenograms of the hands showing type of 12 cone-shaped epiphyses of the middle phalanges of the index through little fingers and of the basal phalanx of the little finger.
a. at the first consultation, b. 4 years and 1 month later.

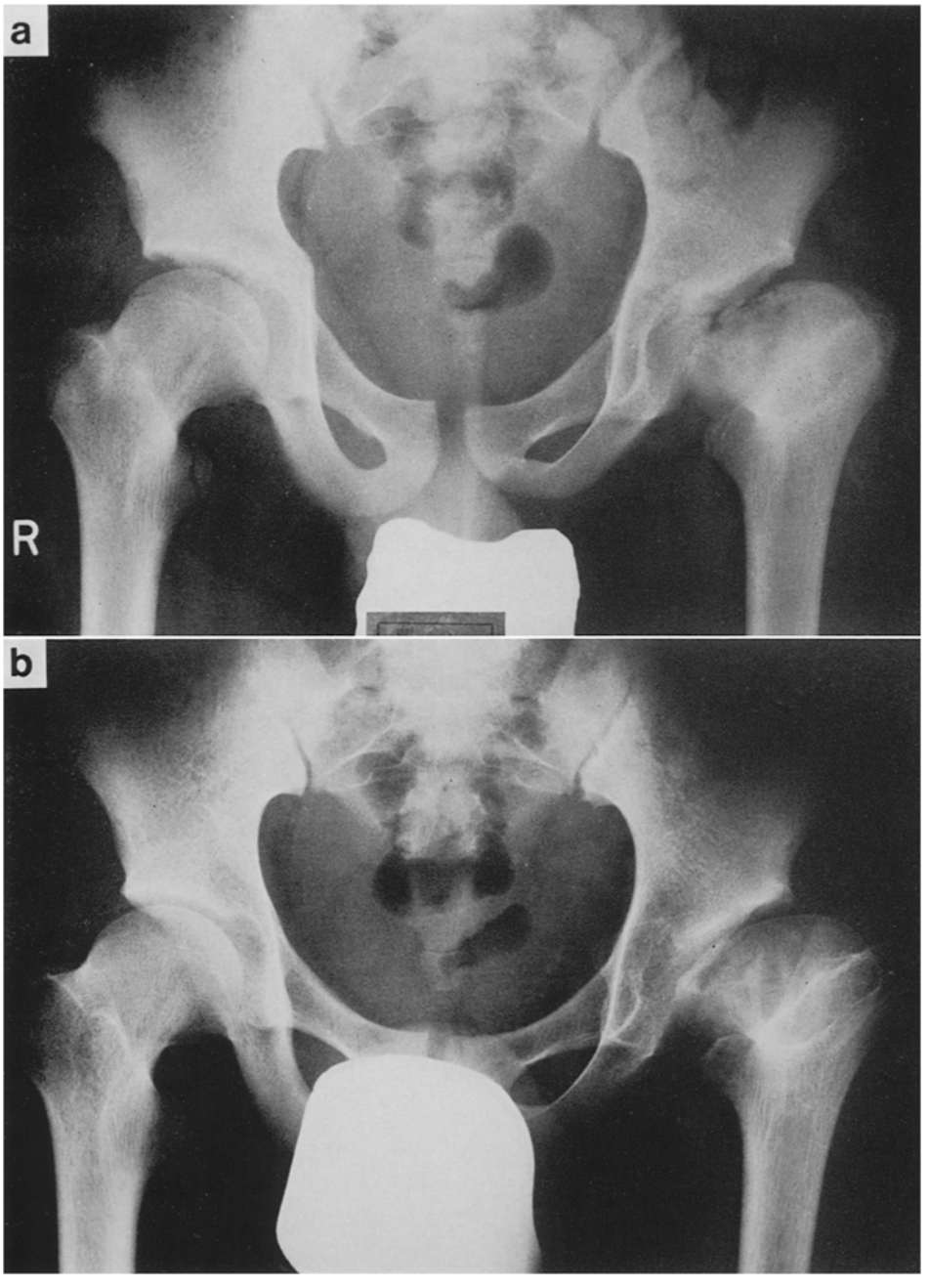

Fig. 4. Antero-posterior roentgenograms of the hip joints showing Pethes-disease-like bone change in the left hip joint.
a. at the first consultation, b. 4 years and 1 month later.

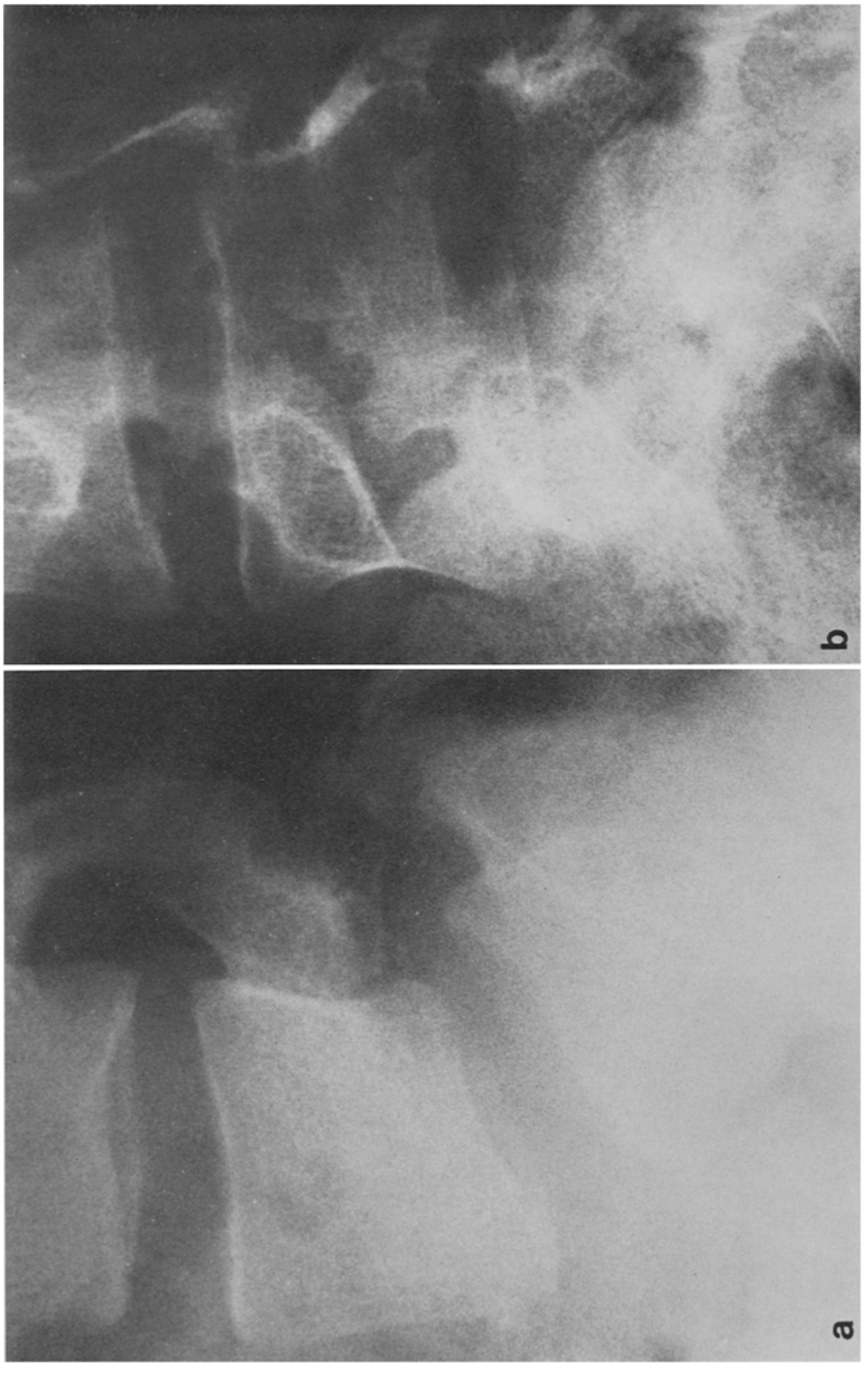

Fig. 5. Roentgenograms of the lumbar spine showing spondylolisthesis of the 5th lumbar spine.
a. lateral view, b. oblique view.

the epiphyses of the middle phalanges of the index, middle, and ring fingers and of the basal phalanx of the little finger, indicating premature closure of the epiphyseal plates at their middle regions. The metaphyses of affected phalanges were broad (so-called type of 12 cone-shaped epiphysis described by Giedion, 1968). The epiphysis of the middle phalanx of the little finger showed premature complete closure. Brachytelephalangy of the thumb with premature closure of the epiphyseal plate was also observed (Fig. 3a).

At the left hip joint, flattening and broadening of the femoral head with marked ossification disturbance were observed. The femoral neck was very short (Fig. 4a). In the lumbar region, the body of the 5th lumbar spine was displaced toward the anterior showing definite separation between the superior and inferior articular processes (Figs. 5a and 5b).

Four years and 1 month later: In each hand skeleton, the middle phalanges of the index through little fingers and the basal phalanx of the little finger were short accompanied by meta-epiphyseal broadening. Closure of the epiphyseal plates was already complete. The bases of these phalanges displayed an inverted V-shape. Marked ulnar deviation of the index finger was noted at the proximal interphalangeal joint (Fig. 3b). In the left hip joint, closure of the epiphyseal plate was complete and ossification disturbance had also disappeared. There was marked flattening and broadening of the femoral head with marked shortening of the femoral neck (Fig. 4b).

DISCUSSION

In the patients with TRP syndrome reported by the authors *et al.* in the previous paper (1976), the pear-shaped deformity of the nose was not marked except in one patient. The present case, however, showed a marked pear-shaped deformity of the nose and a rather long philtrum. Hypotrichosis of the scalp hair was also very marked. A type of 12 cone-shaped epiphysis described by Giedion (1968) was found at each middle phalanx of the index through little fingers resulting in a generalized brachymesophalangy. The epiphysis of the basal phalanx of each little finger was also cone-shaped. Ulnar deviation of the index finger was observed at the proximal interphalangeal joint.

Giedion (1968, 1969) reported that a Perthes-disease-like bone change of the hip joint was occasionally associated with type of 12 peripheral dysostosis. Silverman (1969), Lemaître *et al.* (1970) and Giedion *et al.* (1973) reported three cases of TRP syndrome which were associated with Perthes-disease-like bone change. To the best knowledge of the author, the present case appears to be the fourth one of this association of symptoms in the world literature. Though the bone changes seen in the hip joint resemble very closely those seen in Perthes disease, it is the author's opinion that this change is not due to a true Perthes disease but to an analogous, severe ossification disturbance of the femoral head as a regional expression of TRP

syndrome. The association of spondylolisthesis with TRP syndrome as seen in this report has not yet been reported.

According to Giedion *et al.* (1973), autosomal dominant transmission of TRP syndrome was evident with 16 males and 30 females in 14 families. The author *et al.* (1976) also reported autosomal dominant transmission of TRP syndrome for two or three successive generations in three Japanese families. However, the three cases of TRP syndrome associated with Perthes-disease-like bone change in the literature presented above were all sporadic as well as the present case. Clinical findings of the head and face, and clinical and roentgenographic findings of the hands in TRP syndrome with Perthes-disease-like bone change were quite identical to those in familial TRP syndrome.

In 1974, Spranger *et al.* classified TRP syndrome into two types, type I and type II, in their book. Hair, nose, and hand changes very similar to those of TRP type I are seen in TRP syndrome type II. In TRP syndrome type II, however, short stature, mental retardation and multiple cartilaginous exostosis are seen as associated findings. They described that genetic transmission of TRP syndrome type I is autosomal dominant with considerable variability of phenotypic expression but that of TRP syndrome type II is unknown. The present case is classified as TRP syndrome type I from the symptoms.

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