Trichorhinophalangeal syndrome type I (TRPS I) is a rare complex genetic disorder characterized by sparse, slow growing hair, a bulbous pear-shaped nose, clinobrachydactyly and radiographically cone-shaped epiphyses, especially the second through fourth fingers and toes and deformities of interphalangeal joints resembling those of rheumatoid arthritis (1,2). In addition, there is moderate short stature and, in adult life, moderate joint discomfort in both large and small joints. Trichorhinophalangeal syndrome type II (TRPS II) or Langer-Giedion syndrome is characterized by sparse hair, short stature, bulbous nose, cone-shaped epiphyses, and multiple cartilaginous exostoses. All patients have a de novo deletion affecting 8q24.1, which most often is visible by cytogenetic analysis, and many patients are mildly to moderately retarded (3). In contrast to TRPS II, patients with TRPS I do not develop multiple exostoses, and most of them have normal intelligence and normal chromosomes. Trichorhinophalangeal syndrome type III (TRPS III) seems much rarer than the other types. It differs from TRPS II by the presence of normal intelligence and absence of exostoses and from TRPS I by the severe shortness of all phalanges and metacarpals (4). This report presents a 9.5 year old girl who had the characteristic features of TRPS I.

Case Report
A 9.5 year old girl was admitted to our clinic because of a 4-year history of painless swelling of her fingers. Her past history was otherwise unremarkable. Her parents were not from related families and appeared normal. There were no similar symptoms among relatives or siblings. Her weight was 26.6 kg (25-50p), height 131 cm (25-50p), arm span 124.5 cm and sitting height 73.5 cm. Ratio of upper and lower segment was 1.34. Her pulse rate was 100 bpm and arterial blood pressure was 100/80 mmHg. She had sparse and fine hair and the facies was distinctive, characterized by a bulbous, pear-shaped nose, prominent ears, a long philtrum, a high-arched palate and micrognathia (Figures 1 and 2), along with broadening and slight flexion of the proximal interphalangeal joints of the second to fourth fingers and deviation of the phalangeal axis, with no limitation of motion (Figure 3). She had brittle toenails, hypoplasia of the fourth and fifth toes and a short first toe (Figure 4). Her mental ability was not impaired. X-ray skeletal investigations showed cone-shaped epiphyses in the proximal phalanx of the first digit, in the middle phalanges of the others (Figure 5), and also in the toes (Figure 6). The results of routine laboratory studies were normal. Chromosomal analysis of the patient revealed normal karyotyping (46, XX) with no evidence of deletions or translocations.
Trichorhinophalangeal syndrome is an uncommon and complex genetic disorder that was first described by Giedion in 1966 (5). A hereditary and a sporadic form

Figure 1. With a bulbous, pear-shaped nose, prominent ears, the typical facial appearance of the case was seen.

Figure 2. She had sparse and fine hair.

Figure 3. Along with broadening and slight flexion of the proximal interphalangeal joints of the second to fourth fingers and deviation of the phalangeal axis, the deformities of the fingers of the case are seen.

Figure 4. Brittle toenails, hypoplasia of the fourth and fifth toes and short first toes of the case.

Figure 5. X-ray skeletal investigations showed cone-shaped epiphyses in the proximal phalange of the first digit, in the middle phalanges of the others.
were recognized, representing, respectively, TRPS I, TRPS II or Langer-Giedion syndrome and TRPS III (Table 6). TRPS I is determined by an autosomal dominant gene of variable expressivity. Autosomal recessive transmission is rare. Our patient demonstrated the characteristic presentation of the syndrome. She had sparse and fine hair. Even though the cuticular pattern was not studied by scanning electron microscopy, alterations of the cuticular pattern have been previously described (7,8). She was the only affected patient with TRPS I in the family. She did not have growth failure or mental deficiency. In addition, our finding of normal chromosomes agrees with the majority of the literature on TRPS I. Because 8q24.12 is a very narrow dark band,

<table>
<thead>
<tr>
<th>Highlighted clinical findings</th>
<th>TRPS-I</th>
<th>TRPS-II</th>
<th>TRPS-III</th>
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</thead>
<tbody>
<tr>
<td>Hair</td>
<td>Fine, sparse, brittle</td>
<td>Similar to type I</td>
<td>Similar to type I</td>
</tr>
<tr>
<td>Facies</td>
<td>Bulbous, pear-shaped nose, extended philtrum, grooved chin, maxillary prognathism, mandibular hypoplasia, full medial eye brows</td>
<td>Similar to type I</td>
<td>Similar to type I</td>
</tr>
<tr>
<td>Ears</td>
<td>Frequently lopped, low-set</td>
<td>Similar to type I</td>
<td>Similar to type I</td>
</tr>
<tr>
<td>Teeth</td>
<td>Usually normal</td>
<td>Similar to type I</td>
<td>Similar to type I, “crowded”</td>
</tr>
<tr>
<td>Nails</td>
<td>Broad, flat (spatulate)</td>
<td>Similar to type I</td>
<td>Similar to type I</td>
</tr>
<tr>
<td>Stature</td>
<td>Mildly shortened</td>
<td>Similar to type I</td>
<td>Similar to type I</td>
</tr>
<tr>
<td>Axial skeleton</td>
<td>Usually normal</td>
<td>Spinal pain related to multiple cartilaginous exostoses and/or scoliosis, winged scapulae</td>
<td>Similar to type I</td>
</tr>
<tr>
<td>Long bones</td>
<td>Mildly shortened</td>
<td>Exostoses</td>
<td>Similar to type I</td>
</tr>
<tr>
<td>Joints</td>
<td>Dimpling over MCP joints, premature osteoarthritis</td>
<td>Hyperlaxity, avascular necrosis of hip</td>
<td>Similar to type I</td>
</tr>
<tr>
<td>Hands/ fingers</td>
<td>Shortening, stubbiness, and angulation of fingers (clinobrachydactyly)</td>
<td>Pes planus, foot deformities</td>
<td>Similar to type I; severe shortening of phalanges, metacarpals, metatarsals</td>
</tr>
<tr>
<td>Radiologic findings</td>
<td>Small femoral capital epiphyses; occasionally coxa vara deformity of femur; epiphyseal invagination (cone-shaped); occasionally subluxation of femoral heads, tibia, and radioulnar joints; patellar dislocation</td>
<td>Similar findings with exostoses in metaphyses of long tubular bones, ribs, and vertebrae; avascular necrosis of femoral heads</td>
<td>Similar to type I; no exostoses; severe shortening of phalanges, metacarpals, metatarsals</td>
</tr>
<tr>
<td>Genetic findings</td>
<td>Autosomal dominant deletion of band 8q24.12</td>
<td>Autosomal dominant deletion of band 8q24.11-q24.13</td>
<td>Autosomal dominant</td>
</tr>
</tbody>
</table>
chromosome analysis is normal in the majority of affected individuals (2). However, it should be noted that in cases of TRPS I, microdeletion of the long arm of chromosome 8 (8q24.12), which had previously been associated (9) only with TRPS II, is observed. Therefore, TRPS I and TRPS II may not even be separate entities. TRPS I is a syndrome with a normal life expectancy. All the patients described were at work until at least midlife (10). Attention is usually drawn to the hands during the second half of the first decade because of their deformity rather than because of complaints or symptoms. The diagnostic radiologic features are in the phalangeal ossification centers of the hands. Cone-shaped epiphyses are typically present in the middle phalanges and result in their shortening, together with deformity of the proximal interphalangeal joints. 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 (2). Condensation of bone shadow occurs frequently in the epiphyses of the terminal phalanges.

Progressive osteoarticular changes and degenerative hip disease may necessitate orthopedic care. Howell et al. described 14 patients from 7 families with TRPS I and summarized the features and outlined the clinical significance of the deformities in 1986 (11). The patients’ ages ranged from 13 months to 51 years and deformity of the hands was the commonest presenting sign. There were problems in 1 or both hips in 6 of the 14 patients. For most patients the problems were cosmetic rather than functional. Some had limitation of finger movement and difficulty in wearing rings. The hip disorder was potentially the most serious aspect. The disorder is symptomatically mild, and the main clinical problem is usually that of avoiding confusion with Perthes disease. Naselli et al. reported a pair of monozygotic twin girls with TRPS I, followed from 8.3 to 16.1 years of age in 1998 (12). Both presented poor growth and delayed bone age until about 13 years, followed by marked acceleration of bone age and stunted pubertal height spurt and only one had Perthes-like changes in the right capital femoral epiphyses.

Apart from normal variants, cone-shaped epiphyses can be seen in various disorders (13). Many syndromes that include alopecia and structural abnormalities of the nose and the hands can mimic TRPS I. These include oral-facial-digital syndrome, Larsen’s syndrome, Langer-Giedion syndrome, alopecia-onychodysplasia-hypohidrosis-deafness syndrome, trichoonychodental dysplasia, hidrotic ectodermal dysplasia (Clouston’s syndrome), chondroectodermal dysplasia (Ellis-van Creveld syndrome), and Coffin-Siris syndrome (6). The associated features of these syndromes are important in differential diagnosis.

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