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Facial and oral findings in trichorhinophalangeal syndrome type 1 (Characteristics of TRPS 1)

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Abstract

Findings are reported on 12 members representing five generations of a large family with a history of trichorhinophalangeal syndrome type 1. Consistent facial characteristics included; bulbous pear-shaped nose, elongated philtrum, tented alae, scant eyebrows in the temporal portion, large protruding ears and a thin upper lip. Oral findings included; congenital absence of teeth, considerable delay in root and crown development and isolated cases of hypoplasia, malocclusion, abnormal tooth shape and extensive caries.

Introduction

Giedion[1] in 1966 first identified a syndrome characterized by a triad of findings: sparse hair, bulbous nose, and short deformed fingers. He coined the term trichorphinophalangeal syndrome (TRPS). Since that time a number of reports[2-8] have described various aspects of this condition and demonstrated that it is inherited in an autosomal dominant fashion. The most consistent features are a peculiar facies with a bulbous, pear-shaped nose, fine scalp hair, sparse temporal portion of the eyebrows, and shortening of the phalanges. These changes in finger morphology are attributed to an early and uneven closure of growth plates.[8,9] Radiologically the epiphyses of the middle phalanges of the second, third, and fourth digits have a cone-shaped appearance.[2,3,4] Frequently, these patients also have short stature and multiple skeletal abnormalities.

The following findings have also been reported: growth and mental retardation, brachydactyly, tented nares, prominent elongated philtrum, narrow palate,[4] micrognathia, mandibular retrognathia, and midface hypoplasia,[2,3,4] hip disease,[10-12] kyphoscoliosis and pectus carinatum,[7] and cardiovascular abnormalities.[4,13]

The present study involved a family that spans five generations, with affected individuals ranging in age from birth to 70 years (Figure 1). This family has been thoroughly evaluated by multiple medical and dental specialists and three published reports have presented specific findings.[14-16] A radiographic study[14] of this family reported a high incidence of hip and other skeletal abnormalities including scoliosis, kyphosis and pectus carinatum. The authors concluded that the syndrome most probably represents a condition which results from abnormal maturation of the epiphyses and growth plates.

A cephalometric study by King and Frias[15] of both affected and unaffected members of this same family reported the following findings in affected individuals: 1) the posterior cranial base significantly shortened and deflected superiorly; 2) the mandibular body and ramus were shortened; and 3) there was a shortening of the posterior face height and steep inclination of the mandible resulting from the anterior and superior positioning of the mandibular articulation with the cranium and the short ramus. The authors stated that, with the exception of the shortened ramus, "the craniofacial alterations associated with the trichorhinophalangeal syndrome can be attribted to a primary defect in endochondral growth of the skull."

In a comprehensive evaluation of this same family, Frias et al.[16] focused on the genetic aspects of the syndrome, noting a remarkable variability in expression. Normal intelligence was found in all affected individuals and short stature in 17 of the 23. They also reported spinal abnormalities, rarely mentioned in previous reports, and recommended that detailed radiologic evaluation of parents and siblings of affected individuals be preformed prior to genetic counseling for these families.

Methods and Materials

The sample consisted of 37 members of a single kindred for a history of TRPS, ranging in age from three to 66 years. Older members of this group were excluded from the final study group since they were endentulous. The 12 subjects included in the study group had clinical and radiological findings consistent with the diagnosis of TRPS type I. They ranged in age from three to 31 years.

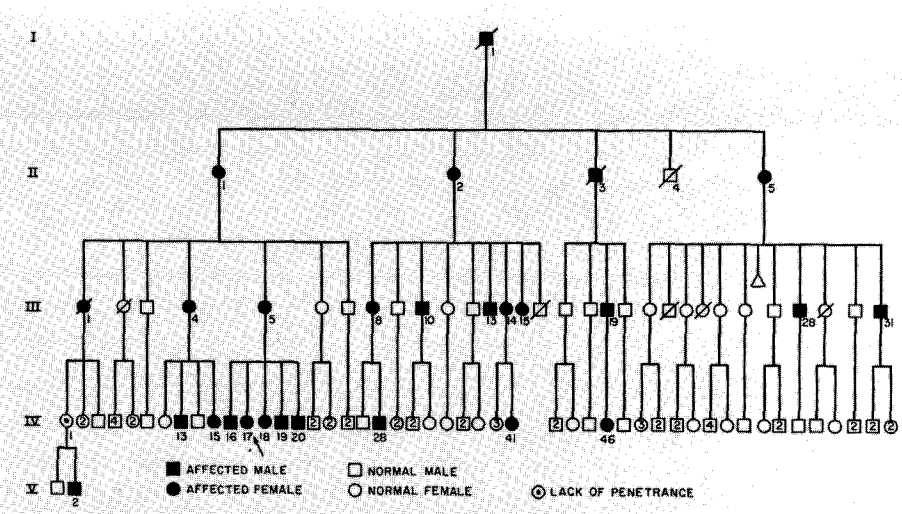

Figure 1. Pedigree of the study family demonstrating autosomal dominant inheritance. Affected individuals shown in black. Diagonal lines indicate deceased individuals. Subject numbers shown below individuals.

Each subject received a visual dental examination during which a series of 2 x 2 color slide photographs (full face, profile, anterior intraoral, maxillary arch and mandibular arch), and a cephalometric and panographic x-ray were obtained. Cephalometric findings are reported in a previous study.[15] The records were arranged in a large notebook alphabetically according to the patients's last name. Information on affected and unaffected individuals was recorded in a separate folder.

A score sheet containing the areas to be examined was developed to include all findings previously reported. Areas shown on Table 1 were evaluated by two examiners (the first and second authors). In addition, the dental ages of seven subjects were calculated from panoramic x-rays using a technic to assess crown and root development described by Nolla.[17] Both examiners evaluated the records and scored each area. The second examiner had no knowledge of whether the patient was an affected or unaffected subject. The results of the two examiners were compared and differences reconciled by joint review of the records in question.

Findings

Table 1 lists the facial and oral characteristics of this population. Ten of 12 subjects had a bulbous nose. The median eyebrows appeared normal in nine subjects, while the temporal portions of the eyebrows were scant in 10 of the 12 subjects. Seven had a straight profile and nine had sparse hair. A thin upper lip was seen in 10 subjects, large laterally protruding ears in 10, and a horizontal groove on the chin in eight of the 12 subjects.

Intraoral findings varied. Eleven of 12 subjects had an ovoid arch form and nine had a deep hard palate. None of the subjects had supernumerary teeth; however, seven of the 12 had congenitally missing teeth. Table 2 identifies the specific teeth missing in each subject and shows that the mandibular second premolars and third molor were the most commonly absent.

A number of additional findings were also noted. Two subjects, IV-1 and IV-13, had anterior open bite; three subjects, IV—15, IV-17 and IV-18, had anterior crossbite and in one subject, IV—17 the crossbite extended into the posterior segment and involved all posterior teeth as well. Two subjects, IV—17 and IV-28, had enamel hypoplasia involving multiple teeth.

Dental age, using a radiographic technic described by Nolla,[17] was calculated on seven subjects between the ages of three and 17 years. Only one subject, IV—17, exhibited a dental age consistent with her chronologic age. The other six showed a delay in dental development ranging from ten months to three years. Subject IV-16 showed a 36-month delay; subject IV-18, showed an 18-month delay; subject IV—19, a 19-month delay; subject IV-20, a 16-month delay; subject IV-41, an 18-month delay; and subject V2, a 10-month delay. Some of these same subjects had congenitally missing teeth; however, the dental age calculations were made as if the teeth were present and developing normally.

Summary

Patients with TRP syndrome type 1 had a characteristic facial appearance (Figure 2, 3 and 4) with a bulbous pear-shaped nose, a long philtrum and tented alae. The temporal portion of the eyebrows are scant, the hair somewhat thin and the upper lip line narrow. They tend to have short stature and slight body build. All of the subjects in this study were of normal intelligence.

Dental findings included congenital absence of teeth, especially lower second premolars and third molars, and considerable delay in root and crown development. Isolated cases of hypoplasia, malocclusion, abnormal tooth shape, and advanced caries were also noted.

The patients in this study did not exhibit supernumerary teeth, narrow palate, or mental retardation. The absence of these features emphasizes the importance of studying a number of different individuals with the same syndrome to accurately identify specific syndrome-related features and not be mislead into thinking isolated, sporadic findings are common for all

Table 1. Facial and oral findings in affected individuals.

	III-15	IV-1	IV-13	IV-15	IV-16	IV-17	IV-18	IV-19	IV-20	IV-41	IV-28	V-2	Total
General													
Age (yrs.)	31	24	17	3	18	16	15	14	7	14	24	7	
Sex	F	F	M	F	M	F	F	M	M	F	M	M	
Normal intelligence	+	+	+	+	+	+	+	+	+	+	+	+	12/12
Short Stature	+	-	-	-	+	+	+	+	+				
Facial Features										+	+	-	8/12
Bulbous nose	+	-	+	+	+	+	+	+	+				
Tented alae	-	-	+	+	+	+	+	+	+	+	+	-	10/12
Long prominent philtrum	+	-	+	+	+	+	+	+	+	+	+	-	9/12
Large laterally protruding ears	+	-	+	+	+	+	+	+	+	+	+	-	10/12
Scant median eyebrows	-	-	+	-	-	-	+	-	+	+	+	-	10/12
Scant lateral eyebrows	+	-	+	+	+	+	+	+	+	-	-	-	3/12
Sparse eyelashes	-	-	-	+	-	+	+	+	+	+	+	-	10/12
Midface hypoplasia	-	-	+	+	+	+	+	+	+	+	+	-	7/12
Micrognatic mandible	+	-	-	-	+	-	+	+	+	+	-	-	8/12
Sparse scalp hair	+	-	-	+	+	+	+	+	+	-	-	-	5/12
Thin upper lip	+	-	+	+	+	+	+	+	+	+	+	-	9/12
Horizontal groove on chin	-	-	+	-	+	+	+	+	+	+	+	-	10/12
Dental										+	+	-	8/12
Congenital absence of teeth	+	+	-	-	-	+	+	-	+	+	-	+	7/12
Convex profile	-	-	+	-	+	+	-	-	+	+	+	-	6/12
Straight profile	+	+	-	+	-	-	+	+	-	-	+	+	7/12
Normal tooth size	+	+	+	+	+	+	+	+	+	+	+	+	12/12
Abnormal tooth size	+	-	-	-	-	+	-	-	+	-	+	-	4/12
Ovoid maxillary arch	+	+	-	+	+	+	+	+	+	+	+	+	11/12
Tapering maxillary arch	-	-	+	-	-	-	-	-	-	-	-	-	1/12
Ovoid mandibular arch	+	+	-	+	+	+	+	+	+	+	+	+	11/12
Tapering mandibular arch	-	-	+	-	-	-	-	-	-	-	-	-	1/12
Deep hard palate	-	-	+	+	+	+	+	+	-	+	+	+	9/12
Class I molar relationship	+	+	-	-	+	-	-	+	-	+	-	+	6/12

Table 2. Congenitally missing teeth.

Subject No.	
III-5	Maxillary left lateral incisor; maxillary and mandibular right 3rd molars
IV-1	All 3rd molars
IV-17	Mandibular left 2nd premolars
IV-18	Mandibular 2nd premolars
IV-20	All 3rd molars and mandibular 2nd premolars
IV-41	Mandibular 2nd premolars
V-5	All 3rd molars

patients with the syndrome.

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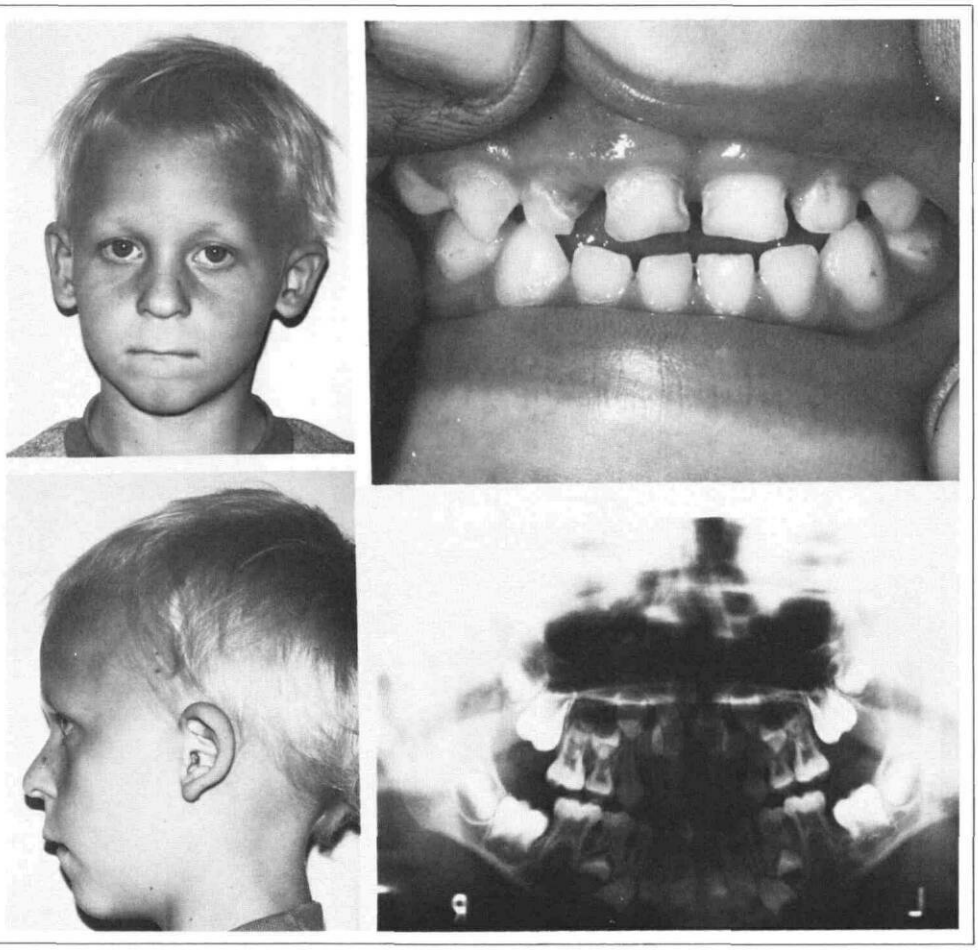
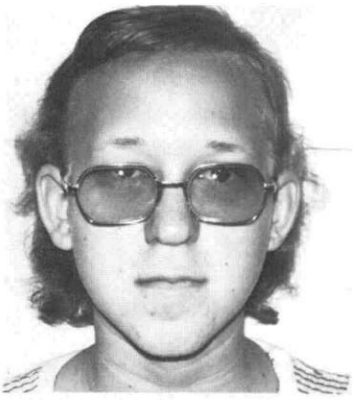
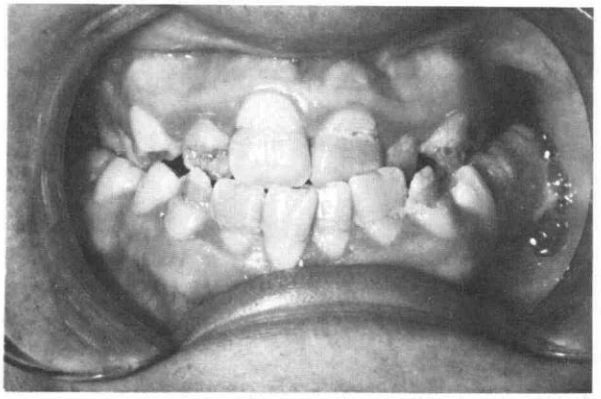


Figure 2. Subject IV-20. Age 6 years, 2 months when records taken. Note facial features, scant hair, extensive anterior caries, anterior open bite, missing lower second premolars, and delayed dental development.





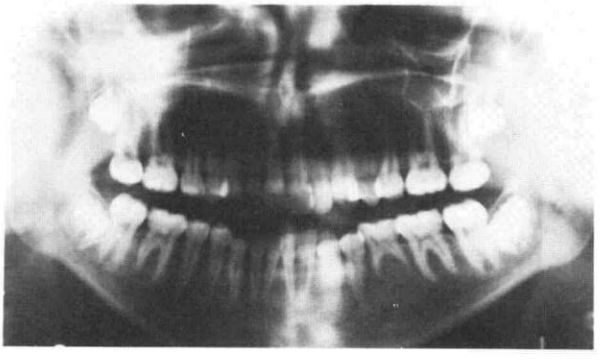
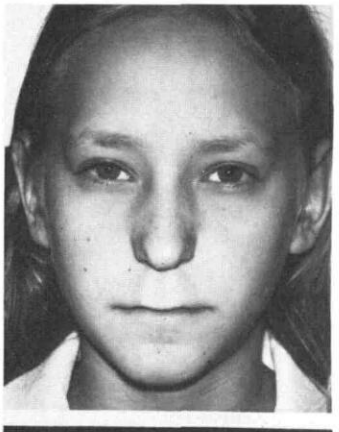
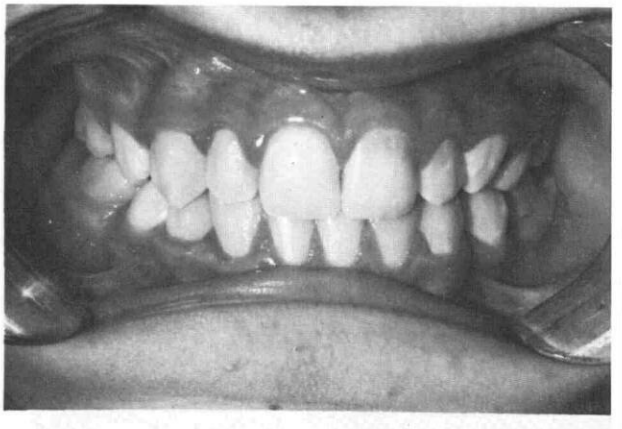
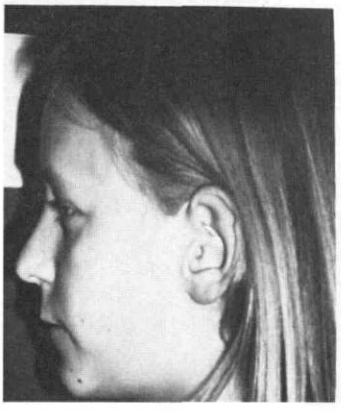


Figure 3. Subject IV-17. Age 15 years, 2 months when records taken. Note facial features, scant hair, enamel hypoplasia, crossbite relationship, missing mandibular left second premolar. Dental development in this subject was consistent with chronologic age.







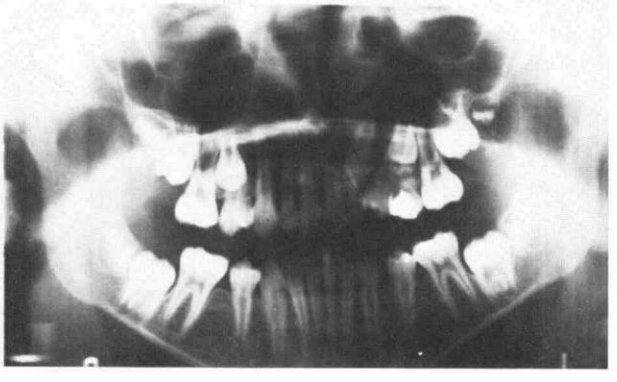


Figure 4. Subject IV-41. Age 12 years, 10 months when records taken. Note facial features, anterior occlusion, missing mandibular second premolars, and delayed dental development.