Orthognathic surgery for a patient with trichorhinophalangeal syndrome type I: A case report

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Trichorhinophalangeal syndrome (TRPS) type I is characterized by slowly progressing systemic osseous dysplasia, exhibiting craniofacial and other skeletal deformities. However, there have been few reports describing this syndrome after undergoing orthognathic surgery. In this report, we present a patient with TRPS type I who successfully underwent orthognathic surgery. In addition, we examined the skeletal stability of the patient for 2 years after the surgery. (Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2006;101:E23-7)

Trichorhinophalangeal syndrome (TRPS) type I is a malformation syndrome characterized by distinctive craniofacial and skeletal abnormalities. Patients with TRPS type I have a bulbous pear-shaped nose, a long and flat philtrum, a thin upper lip, sparse slow-growing hair, protruding ears, short stature, brachydactyly, cone-shaped epiphyses of the fingers, and hip malformations.1,2 The usual mode of transmission is through autosomal dominance. Up to now, over 100 cases have been reported. However, as far as we are aware, there is only 1 report describing a TRPS type I patient undergoing orthognathic surgery.3

In this report, we describe the successful surgical treatment (Le Fort I osteotomy for the maxilla and bilateral sagittal split ramus osteotomy (BSSRO) of the mandible) for a patient with TRPS type I. In addition, we present the dental and skeletal stability of the patient after being followed for 2 years after the orthognathic surgery.

CASE REPORT

A female patient born on August 31, 1982, was diagnosed as having TRPS type I by a pediatrician. No prior history of this disease was noticed in her family.

Physical findings of the patient

Her stature was 145 cm and body weight 43 kg. She had a bulbous pear-shaped nose, a long and flat philtrum, a thin upper lip, sparse slow-growing hair, protruding ears (Fig. 1), brachydactyly, and cone-shaped epiphyses of the fingers (Fig. 2). The patient had normal intelligence and physical activity. All laboratory data were within normal limits, and no cardiac, renal, or hepatic disease was detected.

Clinical course

In 1988, mandibular protrusion and occlusal disharmony became apparent. In 1989, she started orthodontic treatment using a chin cap. However, the growth of her mandible was out of control. In 1997, preoperative orthodontic therapy was started. In 2002, she was referred to our clinic (oral surgery) for a surgical correction of her facial deformity. Because she had a severe class III malocclusion (Fig. 3), we planned a double jaw surgery (Le Fort I osteotomy for the maxilla and bilateral sagittal split ramus osteotomy (BSSRO) of the mandible). In March 2003, a maxillary advancement using a Le Fort I osteotomy and mandibular setback with BSSRO was performed to improve her class I malocclusion and profile. The planned maxillary advancement was 7 mm, and the mandibular set back was 7 and 5 mm on the right and left sides, respectively.

Surgical procedure

A Le Fort I osteotomy and downfracture were performed, and the maxilla was oriented to the mandible with an intermediate splint prepared at the time of mock surgery. The maxilla was fixed with 2 titanium miniplates placed on each side of the piriform rim and zygomatic buttress. Next, the bilateral split osteotomies of the mandible were carried out with the additional use of an appliance developed by our group for the repositioning of the proximal segment of the mandible during this double jaw surgery.4 Under intermaxillary fixation (IMF) in a planned occlusion, the bony segments of the mandibular rami were fixed bicortically in the gonial region with 3 titanium position screws on each side. After the completion of the skeletal fixation, the repositioning appliance and IMF were removed, proper occlusion was verified, and the wounds were sutured. Two intermaxillary training elastics (3/16 or 1/4 inch, medium-light) were applied after surgery.

There were no difficulties encountered during the osteotomy and bone fixation. The quality of the maxillary and mandibular bones appeared to be normal.

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Postoperative evaluation

Lateral cephalograms were obtained preoperatively, and 3, 6, 12, and 24 months postoperatively. Changes in the positions of the anterior nasal spine, upper incisors, lower incisors, and pogonion were measured on coordinate axes passing through the nasion (Fig. 4). The movements of these examination points were represented as linear measurements in millimeters on both axes. Anterior and posterior movements were indicated by positive and negative values, respectively, on the x-axis. Superior and inferior changes were indicated by positive and negative values, respectively, on the y-axis. The overjet and overbite were measured on the same coordinate axes and calculated from the x- and y-axis values of upper and lower incisors.

No complications were observed during the follow-up period. The changes of these examination points are shown in Table I. The postoperative change of each examination point was about within 1 mm. Therefore, we believed that the postoperative dental and skeletal stability of the patient was well maintained. The facial and intraoral photographs of the patient 2 years after surgery are shown in Figs. 5 and 6. Her occlusal condition was well maintained 2 years after the operation.
DISCUSSION

Trichorhinophalangeal syndrome (TRPS) was first reported by Giedion in 1966. Since then, 3 types of TRPS have been reported. TRPS type I is an autosomal dominant disorder characterized by a pear-shaped nose, elongated philtrum, sparse and slow-growing hair, thin upper lip, and bone deformities, especially cone-shaped epiphyses of the phalanges. However, TRPS type I has no mental retardation. In addition, there have been no reports describing abnormality of these examination data in the patients with TRPS I. TRPS type II is called “Langer-Giedion syndrome” and is characterized by loose skin in early childhood and mental retardation. TRPS type III is a serious case of TRPS type I. Patients with TRPS type III have a much shorter stature and more serious brachydactyly than those with TRPS type I. TRPS type III is called “Sugio-Kajii syndrome.”

Recently, the gene of TRPS type I was cloned on the 8q24 human chromosome. Lüdecke et al. screened 51 patients with TRPS type I or III to clarify the correlation between genotype and phenotype concerning bone deformities. They concluded that their evaluation of skeletal abnormalities of patients with TRPS type I mutations revealed various clinical phenotypes. Because TRPS type I shows these various clinical phenotypes, patients with TRPS type I have often been said to be overlooked.

The major radiologic features of TRPS type I are cone-shaped phalangeal epiphyses and premature epiphyseal fusion. This abnormal maturation of the epiphyses can also be observed in other long bones, leading to a growth retardation of stature. In the oral and maxillofacial

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Table I. Postoperative change (mm) of each examination point

<table>
<thead>
<tr>
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<th>3m-6m</th>
<th>3m-12m</th>
<th>3m-24m</th>
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<tbody>
<tr>
<td>ANS (x-axis)</td>
<td>-0.1</td>
<td>-0.2</td>
<td>-0.3</td>
</tr>
<tr>
<td>(y-axis)</td>
<td>0</td>
<td>0.1</td>
<td>0.1</td>
</tr>
<tr>
<td>U1 (x-axis)</td>
<td>0.5</td>
<td>0.7</td>
<td>0.9</td>
</tr>
<tr>
<td>(y-axis)</td>
<td>-0.1</td>
<td>-0.2</td>
<td>-0.2</td>
</tr>
<tr>
<td>Pog (x-axis)</td>
<td>0.7</td>
<td>1.0</td>
<td>1.2</td>
</tr>
<tr>
<td>(y-axis)</td>
<td>-0.2</td>
<td>-0.3</td>
<td>-0.4</td>
</tr>
<tr>
<td>L1 (x-axis)</td>
<td>1.2</td>
<td>1.0</td>
<td>1.1</td>
</tr>
<tr>
<td>(y-axis)</td>
<td>-0.2</td>
<td>-0.5</td>
<td>-0.6</td>
</tr>
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ANS, anterior nasal spine; U1, upper incisors; Pog, pogonion; L1, lower incisors; 3m-6m, change from 3 to 6 months after surgery; 3m-12m, change from 3 to 12 months after surgery; 3m-24m, change from 3 to 24 months after surgery.

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Fig. 3. Preoperative intraoral photographs of the patient: frontal (A) and lateral (B) views of the anterior teeth.

Fig. 4. Cephalometric landmarks and the method used for the analysis of the lateral cephalograms. The X-axis was constructed by drawing a line through the nasion 6 degrees upward from the sella (S)–nasion (N) line, and the Y-axis was drawn as a straight line crossing the X-axis and passing through the nasion (N) point. ANS, anterior nasal spine; U1, upper incisors; L1, lower incisors; Pog, pogonion.
region, patients with TRPS type I are usually reported showing retrognathia of the mandible. However, there are a few previous reports regarding cases of TRPS type I with mandibular prognathism. In one of them, the patient showed a severe relapse after orthognathic surgery, but the reasons for the relapse were not mentioned in the report.

In the present report, a patient with TRPS type I showed mandibular prognathism and underwent a double jaw surgery (Le Fort I osteotomy and BSSRO), in which her postoperative skeletal and dental stability was well maintained 2 years after the operation. Though the reason of the severe relapse described in the previous report is unknown, our patient had no problems after the surgery. This suggests that the prognosis of a patient with TRPS type I undergoing orthognathic surgery is satisfactory when preoperative orthodontic therapy, orthognathic surgery, and postoperative orthodontic therapy are performed properly.
We thank Professor Kimie Oyama and Assistant Professor Michiko Tsuji of Maxillofacial Orthognathics, Graduate School, Tokyo Medical and Dental University, for their helpful discussions and orthodontic treatment for the patient described in this report.

REFERENCES