Orthodontic treatment of a patient with Lowe syndrome

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This article describes the orthodontic treatment of a patient with Lowe syndrome. The objective of the treatment was to improve the patient’s dental relationships and consequently his quality of life. This was achieved by maxillary expansion and extraction of the mandibular central incisors and maxillary deciduous canines. The teeth were aligned and leveled with a fixed orthodontic appliance. Satisfactory results were obtained at the end of treatment, with substantial improvement in dental esthetics, occlusal function, and facial profile. (Am J Orthod Dentofacial Orthop 2011;140:562-8)

The oculocerebrorenal (OCRL) syndrome of Lowe, also called Lowe syndrome, is a rare X-linked recessive disorder, distinguished by a triad of organ system abnormalities: ocular disease, such as neonatal-onset cataracts; mental retardation; and renal dysfunction (online Mendelian Inheritance in Man #309000).1 It was first described by Dr Charles Lowe et al2 in 1952. It occurs predominantly in white and Asian males.3,4 The etiology is related to malfunctioning of the OCRL1 gene on the X-chromosome,5-7 resulting in a deficiency of an enzyme called phosphatidylinositol 4,5-biphosphate.8,9 The normal activity of the Golgi apparatus, which is regulated by this enzyme, is compromised in patients with Lowe syndrome.10

Several reports have described general conditions observed in persons with oculocerebrorenal syndrome.1,11 The combination of ocular (neonatal-onset cataracts, glaucoma, and nystagmus), central nervous system (severe psychomotor retardation and hypotonia), and renal (proteinuria, generalized aminoaciduria, and acidosis) manifestations is usually required for identification of this disorder.12 Other significant common findings include rickets, joint hypermobility, scoliosis, dislocated or subluxed hips, frontal bossing, thin and sparse hair, protruding ears, high-pitched scream, and deviations from the norm in height and weight.13-15

The oral and dental manifestations described in the literature include taurodontism, delayed eruption, enamel hypoplasia, gingival hyperplasia, pericoronal radiolucency, palatal constriction, crowded teeth, vertical facial growth, Class II skeletal malocclusion, underdeveloped mandible, and impacted permanent teeth.1,11 The articles available in the literature are concerned only with reporting the clinical manifestations; none address treatment. In an attempt to fulfill this void, the objective of this article was to report the orthodontic treatment of a patient with Lowe syndrome.

DIAGNOSIS AND ETIOLOGY

An 18-year-old man diagnosed with Lowe syndrome was referred by his family physician for evaluation of dental problems (Figs 1 and 2). There was a history of Lowe syndrome in the patient’s family (Fig 3). Although his parents and sisters had no medical problems, other relatives had been diagnosed with the condition; they were all deceased before 7 years of age.

Intraorally, the patient had severe crowding in both arches, gingival hyperplasia, Class III molar relationship, posterior crossbite, lateral crossbite on the right side, constricted maxillary arch, and absence of some teeth. Overjet was normal, and overbite was 3 mm. The extraoral examination showed a remarkably dolichocephalic face with increased lower facial height, frontal bossing, protruding ears, and incompetent lips. He had a severely convex facial profile, a protrusive maxilla, and a retrusive mandible.

The cephalometric evaluation showed a Class II skeletal malocclusion (ANB, 8°) good maxillary positioning

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mandibular retrusion (SNB, 72°), vertical facial growth (SnGoGn, 62°; FMA, 58°), retroclined maxillary incisors (1.NA, 14°), and buccally proclined mandibular incisors (1.NB, 27°; 1-B, 8 mm). The facial profile was convex (LS-S, 7 mm; Li-S, 6 mm) (Fig 4).

The panoramic radiograph showed taurodontism of the mandibular first molars, retained maxillary deciduous canines, delayed eruption of the maxillary left permanent canine, and delayed development of maxillary left third molar. Ectopic eruption was observed anteriorly in the mandibular arch, probably as a result of the patient’s excessive anterior crowding. In addition, the following teeth were absent: maxillary right third molar, maxillary right first molar, maxillary left first molar, mandibular left third molar, and mandibular right third molar (Fig 5).

The patient’s oral hygiene was relatively good. His mother showed excellent knowledge of preventive procedures and had carefully helped her son maintain adequate oral health conditions. Nevertheless, he had hyperplastic anterior gingival inflammation in both arches, and hypoplastic enamel was observed in the deciduous canines (Fig 1).

**TREATMENT OBJECTIVES**

The treatment objectives for this patient were to correct the posterior and anterior crossbites, correct the buccolingual inclination of the mandibular posterior teeth, align the teeth and level with traction of the maxillary canines, establish a Class I canine relationship, and achieve normal overbite and overjet.

**TREATMENT ALTERNATIVES**

Two treatment alternatives were considered: (1) maxillary expansion with extraction of the mandibular first premolars and the maxillary deciduous canines;
and (2) maxillary expansion with extraction of the mandibular central incisors and the maxillary deciduous canines.

**TREATMENT PROGRESS**

Based on the evaluation reported above, we opted for maxillary expansion with a hyrax expander in association with fixed orthodontic treatment, including extraction of the mandibular central incisors and the maxillary deciduous canines. Initially, edgewise brackets (0.022 × 0.030 in, Morelli, Sorocaba, Brazil) and a hyrax expander were placed and activated (quarter turn per day) until overcorrection was achieved (Fig 6).

Next, the patient was referred for dental extractions. Fifteen days after the mandibular central incisors and maxillary deciduous canines were extracted, 0.012-in nickel-titanium archwires were placed in the mandible and the maxilla for dental alignment and leveling, and an acrylic splint was also placed to correct the position of the maxillary left lateral incisor. Archwires measuring 0.012, 0.014, 0.016, and 0.018 in were made to finish dental alignment and leveling. The treatment proceeded
with the installation of 0.019 × 0.025-in arches to correct the torque and refine the occlusion. Chain elastics were used to close the remaining spaces, and intermaxillary elastics were used to achieve better intercuspation.

When the orthodontic appliance was removed, circumferential and canine-to-canine bonded lingual retainers were placed in both arches.

TREATMENT RESULTS

The results obtained were satisfactory: occlusal relationships, intercuspation, and Class I canine relationships were all corrected; overbite and overjet were normalized; and the facial profile was improved through a reduction in dental protrusion (Figs 7-11).

DISCUSSION

This article describes the first case of orthodontic treatment in a patient with Lowe syndrome. After conducting clinical, radiographic, and cephalometric examinations, one could diagnose alterations related to this syndrome already reported in the literature: vertical facial growth, lack of arch spaces, prolonged retention of deciduous teeth, and maxillary protrusion.1,3,11

In this patient, the orthodontic treatment was aimed at minimizing the oral problems and providing a better quality of life, thus making oral hygiene easier and reducing both periodontal disease and dental caries.

Tooth extractions were performed before orthodontic treatment. Two options were considered: extraction of the mandibular premolars or extraction of the central incisors. The latter option was chosen not only because the orthodontic mechanics would be simplified because of the patient’s motor impairment, but also because the real risk of root resorption resulting from orthodontic movement in patients with Lowe syndrome is unknown. In addition to the mechanical simplification, mild activations were made at 1-month intervals, because published articles on adults with Lowe syndrome include reports of gross periodontal disease with severe bone loss.12-15

The extractions allowed space to be obtained for tooth accommodation, thus improving the patient’s facial profile. Correction of the Class II skeletal discrepancy was not the main objective of the treatment because the patient’s pubertal growth spurt had already passed, and such a discrepancy could now be corrected only by surgical intervention.
Another condition deserving attention was the gingival hyperplasia at the start of treatment. As described in the literature, anticonvulsant drugs can induce gingival hyperplasia, but this possibility was ruled out. Poor dental hygiene caused by bad positioning of the teeth and oral breathing were the possible causes of the gingival hyperplasia. During the treatment, gingival hyperplasia increased and corrective surgery was needed to permit adequate hygiene. A biopsy at the time of surgery showed the presence of fibrous gingival tissue.

**Fig 6.** During expansion with hyrax appliance.

**Fig 7.** Posttreatment photographs.
Maxillary expansion was performed to correct the posterior crossbite, and expanded archwires were used to correct the lingual inclination of the mandibular posterior teeth. For correction of the anterior crossbite, a 0.012-in nickel-titanium archwire was used, although the bite had to be elevated to allow migration of the maxillary left lateral incisor toward the buccal region. This was achieved by installing an acrylic splint, which the patient wore for 2 weeks until the correction of the maxillary left lateral incisor crossbite had been corrected.

One challenge of treating a patient with Lowe syndrome is related to the patient’s cooperation during the treatment. Despite not being a problem in this case, we purposely simplified the mechanical procedures to avoid problems. The results were satisfactory, improving the dental occlusion as well as the facial and dental esthetics. Tooth extractions also improved the facial profile, with the upper and lower lips showing retrusion amounts of 2 and 1 mm, respectively. This allowed the mouth to close passively and reduced the habitual oral breathing.

**CONCLUSIONS**

Orthodontic treatment of a patient with Lowe syndrome is possible and can provide a better quality of life.

**Fig 8.** Posttreatment dental models.

**Fig 9.** Posttreatment cephalometric tracing.
REFERENCES


