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CLINICAL SECTION

Lowe syndrome: literature review and case report

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This paper is the first to describe dental findings and orthodontic characteristics of a young adult patient with Lowe syndrome. This syndrome is a rare genetic disorder inherited by a sex-linked pattern, involving primarily the kidneys, eyes and nervous system, which also present oral manifestations. This paper also present the results of careful extra and intra-oral evaluations and the findings of panoramic and cephalometric radiographs of an 18-year-old male with Lowe syndrome.

Key words: Orthodontic treatment, Lowe syndrome

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Introduction

Lowe syndrome (LS) also known as oculocerebrorenal syndrome was first described by Dr Charles Lowe and his colleagues in 1952.[1] This syndrome is a rare genetic disorder inherited by a sex-linked pattern, occurring predominantly in Caucasian or Asian males, and involving primarily the kidneys, eyes and nervous system.[2,3] The aetiology is related to malfunctioning of OCRL1 gene in the X chromosome,[4,5] resulting in a deficiency of an enzyme called phosphatidylinositol 4,5-biphosphate.[6] Normal activity of the Golgi apparatus regulated by this enzyme is compromised in LS.[7]

Several reports have described general conditions frequently observed in individuals with oculocerebrorenal syndrome. The combination of ocular (neonatal onset cataract, glaucoma and nystagmus), central nervous system (severe psychomotor retardation and hypotonia) and renal (proteinuria, generalized aminoaciduria and acidosis) manifestations are usually required for the identification of this disorder.[8] Other significant common findings include: rickets, joint hypermobility, scoliosis, dislocated and/or subluxed hips, frontal bossing, thin and sparse hair, protruding ears, high-pitched scream, as well as deviations from norm in height and weight.[9,10]

Even though the LS has been clinically studied in detail, oral manifestations related to this syndrome are scarcely described. The first reports of oral findings in LS patients showed delayed eruption, crowding, constricted palate, taurodontism of the molars, hypoplastic enamel, dental caries and large pericoronal radiolucencies.[11,12] Batirbaygil and Turgut[13] described a case with a transversely deficient palate, multiple eruption cysts and haematomas. Neither hypoplastic enamel, nor delays in eruption of primary teeth were noticed. Another report on a four-year-old male revealed chronic hyperplasic marginal gingivitis, generalized mobility of all teeth, and normal dental development without any apparent carious lesions.[14]

It appears that the present case report is the first to describe dental findings and orthodontic characteristics of a young adult patient with LS.

Case report

An eighteen-year-old male diagnosed with LS was referred by the family physician for evaluation of dental problems and possible orthodontic treatment. There was a history of LS in the patient's family. Although his parents and sisters presented no medical problems, an analysis of his family history (Figure 1) revealed that he had other relatives also diagnosed with such a condition. All of them were deceased before seven years of age.

Extra-oral evaluation revealed a remarkably dolichocephalic face with increased lower facial height, frontal bossing, protruding ears and incompetent lips (Figure 2). Examination of the lateral view of his face showed a severely convex profile, protrusive maxilla and retrusive mandible (Figure 3).

The panoramic radiograph showed taurodontism of the lower first molars, retained maxillary upper deciduous canines, eruption of the maxillary left permanent canine was delayed, and the upper left third molar had a delayed development. Ectopic eruption was observed anteriorly in the lower arch, probably as a result of

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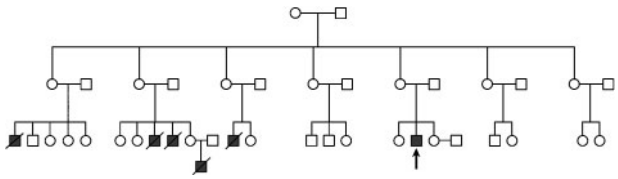


Figure 1 Family pedigree showing the X-linked characteristic of Lowe syndrome. Arrow indicates proband presented in this report

patient's excessive anterior crowing. In addition, the following teeth were absent: UR8, UR6, UL6, LL8, LR8 (Figure 4).

Cephalometric radiographic evaluation revealed a skeletal Class II with a posteriorly located mandible (Figure 5). All measurements analyzing the vertical dimension indicated a significant excess in facial height, although an open bite was not present. The upper incisors were relatively well positioned in relation to the SN plane, and the lower incisors were remarkably retroclined in relation to the mandibular plane. The results of the cephalometric analysis are presented in Table 1.

Intra-oral examination was compromised by the patient's erratic behaviour and his high level of anxiety.

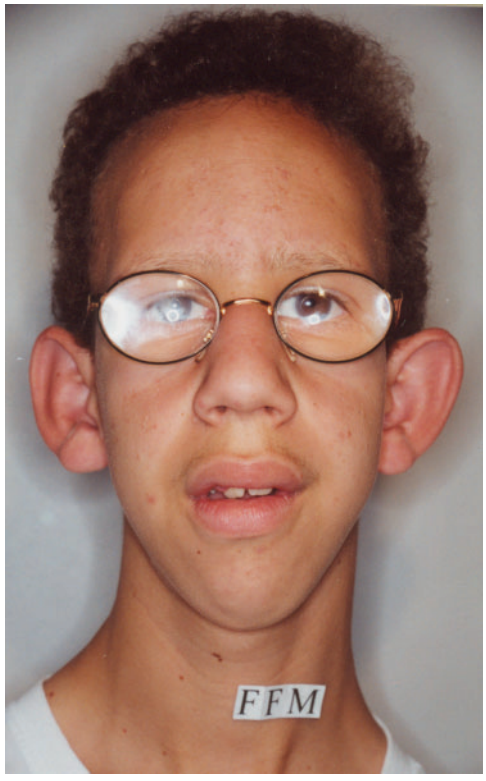


Figure 2 Frontal extra-oral photography showing long facial height, frontal bossing and protruding ears

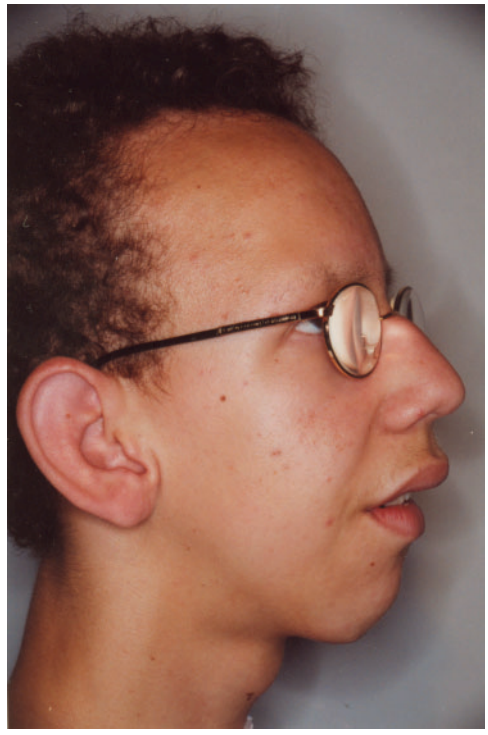


Figure 3 Lateral extra-oral photography: severely convex profile and incompetent lips

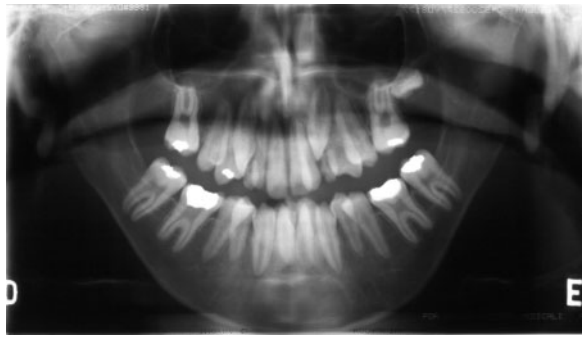


Figure 4 Radiographic image of over-retained deciduous canines, impacted permanent teeth and taurodontism of the molars

Table 1 Summary of patient's cephalometric evaluation.

Measurement	Population norm	Patient
SNA	81°	80°
SNB	78°	72°
ANB	+3°	+8°
NAP	+2°	+15°
FMA	22°	58°
Gonial angle	125°	160°
Y-axis	59°	74°
Upper 1–SN	104°	95°
Upper 1–AP	+3 mm	+10.5 mm
IMPA	90°	70°
Lower 1–AP	+2 mm	+6.5 mm
Upper 1–lower 1	131°	132°

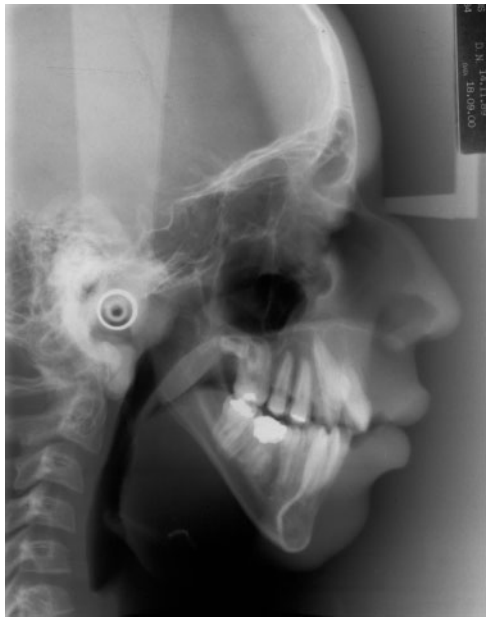


Figure 5 Cephalometric radiograph illustrating the dolichocephalic appearance, with steep mandibular plane angle, short mandibular ramus and obtuse gonial angle

Because of this intra-oral, occlusal photography was not possible. Oral hygiene was relatively good, with his mother showing excellent knowledge of preventive procedures and being very careful in maintaining adequate oral health conditions. There was hyperplasic anterior gingival inflammation in both maxillary and mandibular arches. Hypoplastic enamel was observed in the deciduous canines (Figure 6).

Dental impressions were performed despite the patient's uncooperative attitude and a set of orthodontic models was obtained. The study models revealed severely constricted dental arches, especially the upper,

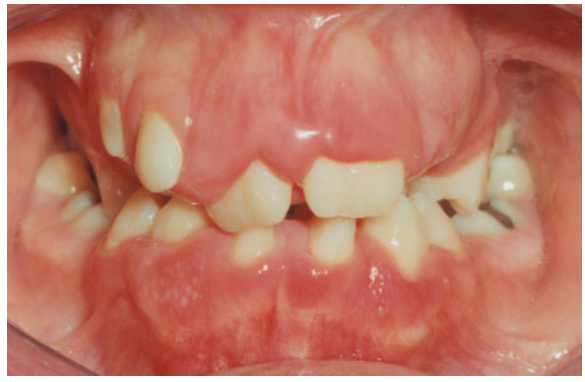


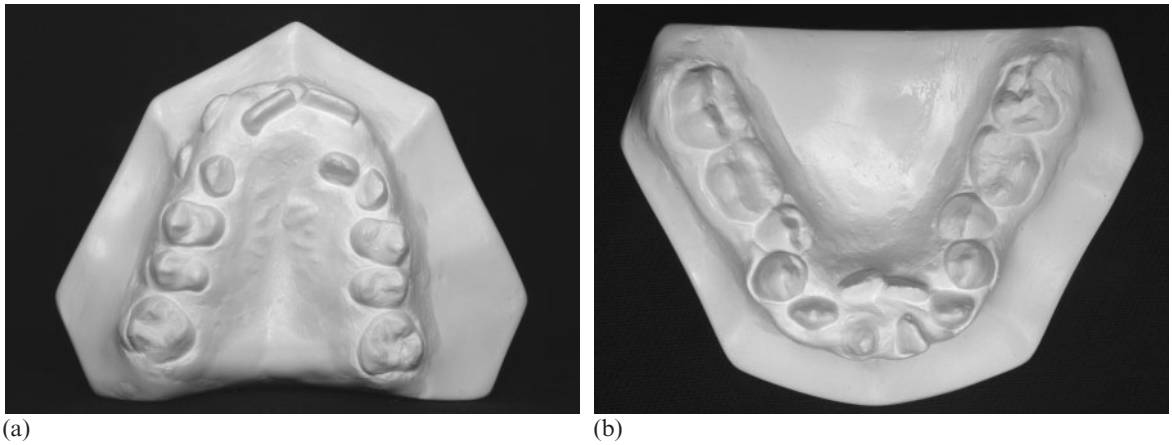
Figure 6 Intra-oral photograph showing hyperplasic anterior gingival and posterior crossbite

resulting in a bilateral posterior crossbite, as well as excessive maxillary and mandibular crowding (Figure 7a,b). Overjet was minimal, and a 3 mm overbite was measured. The molar relation was Class III, probably due to the early loss of maxillary first permanent molars (Figure 8a,b).

Discussion

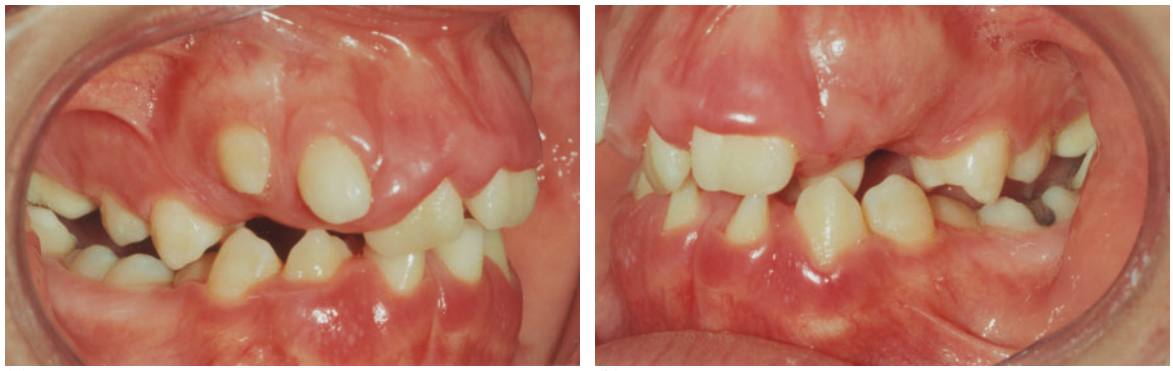
The characteristics of LS patients have been widely studied in the medical literature. However, there are a limited number of articles presenting oral manifestations in individuals with this syndrome. All reports found in the literature assessed young patients with primary or early mixed dentition. This appears to be the first report to describe dental findings and orthodontic characteristics in a young adult male with LS.

The average lifetime of patients suffering from this syndrome varies between five and seven years.[3] However, there have been reports of patients with mild



(a) (b)

Figure 7 (a,b) Occlusal view of orthodontic casts revealing constricted dental arches and severe crowding



(a) (b)

Figure 8 (a,b) Lateral intra-oral photographs. Minimal overjet and Class III molar relation

manifestations of LS that lived through the fourth decade.[8,15] Therefore, it is important for dentists to be aware of oral conditions in such patients.

Extensive caries lesions have been previously described in LS patients,[10,12] and such a problem was present in this patient, but teeth had been already restored. The caring attitude observed in his mother, who reported helping the patient during his oral hygiene procedures, might explain why active tooth decay was not present. Harrison *et al.*[14] reported extensive calculus deposits in a four-year-old male affected by LS. Since only mild calculus formation was noticed, the hyperplasic anterior gingival inflammation found in this patient appears to be related to the excessively malpositioned teeth, as well as to oral breathing habits. Based on these findings, one can assume that if an adequate oral hygiene protocol is implemented, caries and periodontal problems can be minimized in LS patients.

Delayed eruption of permanent teeth was a common feature to all patients analyzed in both reports describing individuals with LS in the mixed dentition.[11,12] Since we first examined our patient at 18 years of age, it is impossible to know if the eruption pattern had been delayed. However, one can assume that if the eruption was delayed, it did not result in an increased number of impacted teeth. We believe the impactions noticed in this patient are primarily related to the severe anterior crowding. Such problems can also be found in the unaffected population, if orthodontic therapy is not implemented, during the transition from the late mixed to the permanent dentition.

Taurodontism in all molars, retained deciduous teeth, and crowding have been previously described as oral findings in LS patients. These features were also noticed in our young adult patient, what could be an indicative of characteristic oral manifestations of this syndrome but taurodontism was observed only in the lower first molars. A constricted maxilla was found in other LS patients.[10,11,13] The transverse deficiency in our patient's maxilla and the excessive anterior crowding were the major occlusal problems encountered.

Abassi *et al.*[3] mentioned that a child affected by LS has a 'characteristic look' resulting from some physical abnormalities, which included: pale skin, prominent frontal bossing, inattentiveness, hyperexcitability, and assumption of bizarre positions due to joint hypermobility and hypotonicity of the muscles.

The patient in this case report presented with a symmetric but severely long face. Both upper and lower facial thirds were significantly increased in height, and the middle facial third had normal length. Marked frontal bossing, anteriorly receded hairline, and hypotonic lips were also noticed. Profile examination confirmed the severely excessive vertical facial height, and also revealed a significantly convex profile, an anteriorly positioned maxilla, an inferiorly–posteriorly located chin button, and protrusive lips.

The cephalometric analysis supported the clinical diagnosis obtained by the clinical examination. The comparison between his cephalometric data and the average population norms helped to quantify the severity of his skeletal craniofacial deviations. The most remarkable finding was the increased vertical facial height. Since there are no other descriptions of cephalometric findings for oculocerebrorenal patients, additional case reports are needed to confirm if these characteristics are common to a number of affected individuals or unique features related to this particular patient.

Conclusions

There have been approximately 200 LS cases reported to date, demonstrating its rare occurrence. Since these

patients present oral manifestations, dentists should be aware of their characteristics and how they compromise patient's dental health conditions. This case illustrated oral findings and orthodontic characteristics of LS in a patient who presented with their permanent dentition. Further case reports must confirm if these findings are characteristic of patients with this syndrome, or isolated to this particular individual.

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