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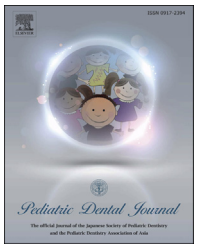

Case Report

Lowe syndrome oral findings: Case report

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ABSTRACT

Lowe syndrome is an X-linked recessive disorder characterized by anomalies of the eye, nervous system, and kidney, and caused by a mutation of the oculocerebrorenal gene (*OCRL1*) on the X-chromosome. Dental manifestations are associated with rickets due to renal dysfunction. A 2-year-1-month-old boy with Lowe syndrome came to our clinic for consultation in regard to mobility of the primary mandibular incisors. Periapical radiograph findings showed alveolar bone absorption and a wide pulp chamber. Dental caries extending to the pulp was detected on the palatal side of the maxillary central incisors. We initially provided brushing and dietary habit instructions, and subsequently performed periodontal and dental caries treatments. Repeated periodontal therapy led to disappearance of mobility of the affected teeth at the age of 3Y3M.

1. Introduction

Lowe syndrome (oculocerebrorenal syndrome) was first reported in 1952 by Lowe et al. [1] as an X-linked recessive disorder. The disease is caused by a mutation of the oculocerebrorenal gene (*OCRL1*) on the X chromosome [2] and its frequency is estimated to be one per 500,000 [3]. Notable characteristics in affected individuals are anomalies of the eye (neonatal onset dense cataract, cataract, nystagmus) and nervous system (hypotonia, mental retardation), and renal tubular dysfunction (Fanconi syndrome) [3]. Most patients develop chronic renal failure and renal tubular dysfunction leads to hypophosphatemia, with dental findings commonly associated with rickets, such as enamel and dentin hypoplasia, absence of lamina dura, wide pulp cavity, and elongated pulp horns [4–6]. However, there are few reports about dental manifestations in patients with this syndrome. In the present report, we describe oral findings and treatment of a Japanese child with Lowe syndrome.

2. Case report

A 2-year-1-month-old Japanese boy diagnosed with Lowe syndrome was referred to the Pediatric Dentistry Clinic of Osaka University Dental Hospital, Osaka, Japan with mandibular incisor mobility. The patient had been diagnosed with Lowe syndrome and was affected by mental retardation, congenital cataracts and glaucoma, and renal tubule dysfunction. There was nothing unusual about his family health history. Our first oral examination revealed that all primary teeth except for the primary second molars had

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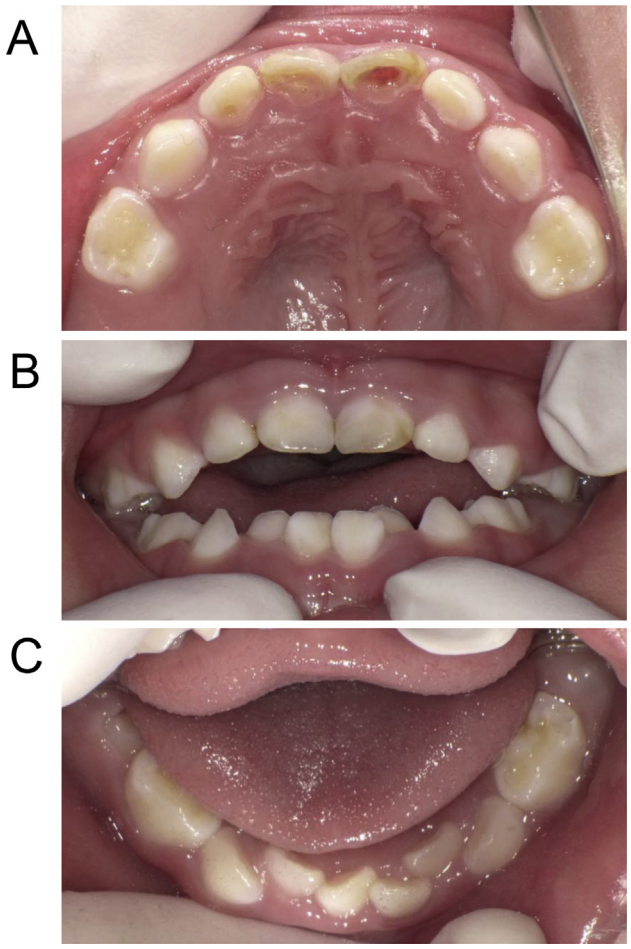

Fig. 1 – Intraoral photographs taken at 2 years and 1 month. (A) Palatal view. (B) Front view. (C) Lingual view.

erupted (Fig. 1). Mobility of the maxillary primary central incisors and mandibular primary central and lateral incisors was recognized, with the mandibular primary anterior teeth showing a severe condition. Dental caries was detected in the palatal region of the maxillary primary central and lateral incisors, and pulp exposure was recognized in the maxillary primary central incisors (Fig. 1A), while enamel hypoplasia was seen on the labial side of the maxillary central incisors (Fig. 1B). In addition, the mandibular anterior teeth showed crowding (Fig. 1C). Anterior open bite was identified, which was speculated to be caused by finger sucking. Periapical radiographs showed alveolar bone resorption around the mandibular primary incisors along with enlargement of pulp spaces in the anterior teeth (Fig. 2). His mother reported that he was still being given milk before and during sleeping. Therefore, we gave instructions to her about brushing and dietary habits.

Two weeks later, the patient returned to our clinic with swelling of the gingiva. We performed periodontal treatment and the swelling had disappeared by the next visit 1 week later, at which time we began to perform treatment for dental caries. First, a pulpectomy was performed for each of the maxillary central incisors (Fig. 3), after which diammine silver fluoride was applied to the maxillary lateral incisors to stop caries development. During scheduled follow-up appointments, we performed periodical periodontal treatment at each visit and noted that tooth mobility had disappeared at the age of 3 years and 3 months, when all primary teeth were erupted, and an anterior open bite was detected (Fig. 4). According to his mother, the patient still sucked his finger at that age.

3. Discussion

One of the main manifestations of Lowe syndrome is dysfunction of the renal system leading to rickets, which is commonly associated with dental manifestations seen in affected patents [4–6]. As for dental manifestations of rickets, enamel and dentin hypoplasia, absence of lamina dura, wide pulp cavity, and elongated pulp horns have been generally reported [4–6], which can lead to dental caries, tooth mobility, dental abscesses without caries or trauma, and malocclusion. In the present case, severe mobility of the anterior teeth and swelling of gingiva were also recognized at the first examination, and we speculated that inflammation of the deep periodontal pocket due to poor hygiene had induced the swelling. Fortunately, mobility was stopped by repeated periodontal therapy. Nevertheless, we plan to continue maintenance for periodontal conditions at follow-up visits, since alveolar bone resorption was detected around the affected tooth. In addition, dental caries and enamel hypoplasia were noted on the palatal side of the maxillary anterior

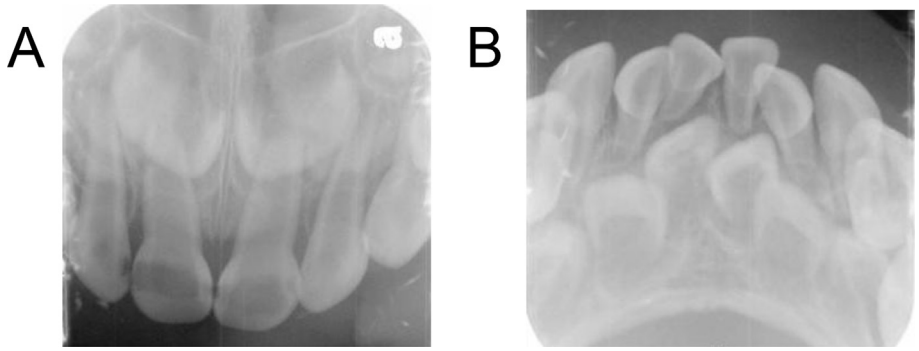

Fig. 2 – Periapical radiographs obtained at 2 years and 1 month. (A) Maxillary anterior region. (B) Mandibular anterior region.