Orthodontic Treatment for Bloch-Sulzberger Syndrome in Patient with Cleft Lip and Palate

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Abstract

We performed orthodontic treatment, fitted prostheses, and provided restorative treatment in a patient with Bloch-Sulzberger syndrome and cleft lip and palate during the early mixed dentition period. We report the case after a subsequent 6-year retention phase including the period of pubertal growth. A girl aged 8 years 4 months visited our hospital with the chief complaint of crowding of the anterior teeth and anterior crossbite. She had bilateral cleft lip, alveolus, and palate; a Class II molar relationship; winging of both the maxillary bilateral central incisors; and spacing in the mandibular anterior teeth arches. Anterior crossbite comprised 0 mm overbite and −1 mm overjet. The crown diameter was at least one standard deviation smaller than normal in both the deciduous and permanent teeth, and the crowns were slightly peg-shaped. Panoramic radiograph confirmed congenital absence of 21 permanent teeth. Cephalometric analysis revealed poor growth of the maxilla, downward growth of the mandible, and lingual inclination of the maxillary central incisors. The diagnosis was skeletal anterior crossbite with cleft lip, alveolus, and palate, accompanied by hypodontia. Orthodontic treatment comprised an edgewise appliance and an expansion arch to improve crowding and anterior crossbite. The appliance was removed 2 years after treatment initiation, followed by crown restorations of the maxillary central incisors and mandibular deciduous anterior teeth. A metal retainer was then fitted to the maxillary dentition. She was subsequently placed in a 6-year retention phase including pubertal growth, during which occlusal stability and esthetics were maintained.

Key words: Orthodontic treatment — Bloch-Sulzberger syndrome — Cleft lip and palate — Multiple congenitally missing teeth — Anterior crossbite

Introduction

Bloch-Sulzberger syndrome, a hereditary form of ectodermal dysplasia, is characterized by the manifestation of erythema-like streaks and clusters of vesicles primarily on all four limbs and around the trunk at birth or several weeks thereafter. This condition has an
X-linked-dominant pattern of inheritance, and is extremely rare in men\(^\text{11}\). It usually develops as a result of a mutation that deactivates the \(IKK\gamma\) (\(IkB\) kinase \(\gamma\)) gene (also referred to as the \(NEMO\), or \(NF\kappa B\) essential modulator gene), which is located on region 28 of the long arm of the X chromosome (Xq28)\(^\text{15}\). In addition to the symptoms described above, other ectodermal tissue abnormalities may also occur, such as those related to the central nervous system and abnormalities of the teeth, eyes, bone, hair, and nails. Rashes develop frequently all over the body, followed by grayish-brown pigmented spots in the same areas. Thereafter, the pigmented spots gradually disappear, and the prognosis is good\(^\text{11}\). Tooth abnormalities include hypodontia, delayed eruption, peg-shaped teeth, impacted teeth, morphological anomalies, and malocclusion\(^\text{6,8}\). In addition to reports on skin symptoms associated with this syndrome\(^\text{11}\), a number of studies have also described associated dental features\(^\text{2,4,6,7,9,13}\) and corresponding orthodontic treatment\(^\text{1,3,10,14,16}\). Reports on orthodontic treatment for this syndrome when accompanied by cleft lip and palate are particularly limited, however, and, to our knowledge, there are none on the subsequent long-term course of such treatment.

In this case study, favorable treatment outcomes were achieved by performing orthodontic treatment, fitting prostheses, and providing restorative treatment in a patient with Bloch-Sulzberger syndrome and cleft lip and palate during the early mixed dentition period. We report the long-term outcomes of this case, wherein occlusal stability and esthetics were maintained over a 6-year period during which pubertal growth occurred, despite the occurrence of mild regression.

**Case Presentation**

A girl aged 8 years 4 months was referred to our clinic with the chief complaint of crowding of the anterior teeth and anterior crossbite by the Department of Plastic Surgery on the basis of a diagnosis of Bloch-Sulzberger syndrome. She was the second child in her family, and had an older brother. A diagnosis of Bloch-Sulzberger syndrome had been made in no other family member, however. At birth, she had cutaneous erythematous lesions with linear vesication on her lower limbs and axilla. By 10 weeks of age, she had developed similar lesions on her trunk, palms, and soles, leading to a diagnosis of Bloch-Sulzberger syndrome. At the time of the initial examination at our department, however, she exhibited no skin symptoms apart from

![Fig. 1 Pre-treatment facial photographs at age 8y4m](image)
pigmented spots due to sunburn. In addition, she had undergone a cheilostomatoplasty at 6 months and surgery for repair of cleft palate repair at 1.5 years. On presenting to us, she also had mild dysarthria, but no abnormalities of the central nervous system such as mental retardation.

Facial findings revealed a symmetrical frontal view, downslanting palpebral fissures, strabismus, and external malformation of the ear on the left side. Her lateral profile view revealed a straight facial type (Fig. 1). Intraoral examination revealed bilateral cleft lip, alveolus, and palate; a Class II molar relationship; winging of both maxillary central incisors; and spacing in the mandibular anterior tooth arches. Anterior crossbite presented as 0 mm overbite and −1 mm overjet. Crossbite was observed at the site of the left maxillary teeth C and D. The mandibular midline was displaced 5 mm to the left of the maxillofacial region. Teeth 1, C, D, and E on the left side of the maxilla; 1, 3, D, and E on the right side of the maxilla; and A, B, C, D, E, and 6 on the left and right sides of the mandible had erupted. Moreover, crown diameters were at least one standard deviation smaller than usual in both the deciduous and permanent teeth, and the tooth crowns were slightly peg-shaped (Fig. 2). The occlusal condition during the deciduous dentition period was unclear. Panoramic radiograph confirmed congenital absence of 21 permanent teeth: 2, 3, 4, 5, 6, and 7 on the left side of the maxilla; 2, 4, 5, 6, and 7 on the right side of the maxilla; 1, 2, 3, 4, 5, and 7 on the left side of the mandible; and 1, 2, 3, and 5 on the right side of the mandible. Furthermore, the mandibular first molars were single-rooted teeth (Fig. 3). Cephalometric analysis revealed a skeletal pattern with an SNA of 67°, SNB of 70°, and ANB of −3° anteroposteriorly, and a mandibular plane angle of 30°, as well as a gonial angle of 130° vertically. Poor growth of the maxilla and downward growth of the mandible were also observed. The facial type was a dolichofacial, and the dentition revealed lingual inclination of maxillary central incisors (Fig. 4).

The list of concerns in this case included
(1) bilateral cleft, alveolus, and palate; (2) multiple hypodontia; (3) small crown diameters and a narrow dentition diameter; (4) poor maxillary growth; (5) lingual inclination of the maxillary anterior teeth; (6) crowding of the maxillary anterior teeth; and (7) anterior crossbite. Based on the aforementioned findings, the diagnosis was skeletal anterior crossbite with cleft lip, alveolus, and palate, accompanied by hypodontia.

The treatment objectives were (1) improvement of crowding of the maxillary anterior teeth; (2) improvement of the anterior crossbite; and (3) space management. The plan was to restore the crown morphology of the maxillary and mandibular anterior deciduous and permanent anterior teeth and fit a metal retainer with artificial teeth to improve and maintain esthetics in the maxillary dentition after completion of active treatment.

### Clinical Procedures and Outcomes

Orthodontic treatment involved fitting a sectional edgewise appliance to the bilateral central incisors for alignment, followed by an expansion arch to the band of the maxillary second deciduous molar to anteriorly expand the maxillary anterior teeth. Bone grafting of the cleft alveolus was then performed at 7 months after treatment initiation. One year later, a sectional edgewise appliance was fitted between the mandibular canines to achieve space closure. After 1 year 6 months following eruption of tooth 4 and 7 on the right side of the mandible, a band was fitted to the mandibular molars and the deciduous anterior teeth moved lingually by means of a closing loop (Fig. 5). The edgewise appliance was removed at 2 years after initiation of treatment (Figs. 6, 7). After treatment, a pan-

<table>
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<th>Post Treat.</th>
<th>Post Ret.</th>
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Fig. 4 Tracing and measurements on pre-treatment cephalometric radiograph
Fig. 5  Progressive intraoral photographs during alignment using an E-arch (upper image) and closing of lower space using closing loop (lower image)

Fig. 6  Post-treatment facial photographs at age 12y10m

Fig. 7  Post-treatment intraoral photographs at age 12y10m
oramic radiograph revealed no noticeable root absorption or pathological mobility in the remaining deciduous teeth (Fig. 8). The patient subsequently received crown restoration for the maxillary central incisors and mandibular anterior deciduous teeth along with a metal retainer incorporating artificial lateral incisors in the maxillary dentition at the pediatric dental clinic. At 6 years after initiation of active treatment, mild crossbite recurred and spaces developed between the mandibular deciduous central incisors, although occlusion remained mostly stable and esthetically acceptable. In addition, the remaining deciduous teeth showed no noticeable root absorption or mobility (Figs. 9–11).

A comparison of superimposed cephalometric radiographs and analyses revealed

Fig. 8 Post-treatment panoramic radiograph at age 12y10m

Fig. 9 Post-retention facial photographs at age 18y6m

Fig. 10 Post-retention intraoral photographs at age 18y6m
almost no vertical growth in the maxilla over the course of treatment, but did show forward growth. Furthermore, vigorous anteroinferior growth was observed in the mandible, resulting in slight deterioration of the Class III jaw relationship. However, corrected coverage was achieved by inclining the maxillary anterior teeth labially and inclining the mandibular anterior deciduous teeth lingually to compensate for this. During the retention period, almost no growth or development was observed in the jawbone, and the mandible rotated clockwise. Moreover, almost no change was observed in the tooth axes (Fig. 12).

Written informed consent was obtained from the patient’s parents for publication of this case report and accompanying images.

Discussion

Bloch-Sulzberger syndrome was named incontinentia pigmenti by Bloch in 1926, and was later suggested to be a type of ectodermal dysplasia by Sulzberger in 1928. The name Bloch-Sulzberger syndrome, as it is known today, was proposed after it was proven that the accompanying symptoms were not restricted to those of the skin.

Maeda et al. examined 21 cases of Bloch-Sulzberger syndrome reported in the field of dentistry and found that all these studies described tooth anomalies with a high incidence of comorbidity; in addition, hypodontia, abnormal tooth morphology and structure, and delayed eruption of deciduous teeth accounted for the three most common anomalies, similar to past reports. However, none of these studies had described cases of patients with cleft palate. Moreover, Minic et al. conducted a systematic review of dental findings in Bloch-Sulzberger syndrome between 1993 and 2010 and reported a high incidence of dental shape anomalies, hypodontia, and delayed dentition, together with cleft and high-arched palates. Furthermore, approximately 1 in 3 of cases were reported to involve a cleft palate. The present patient had multiple congenitally absent teeth and dental morphological anomalies similar to previously reported cases; however, delayed eruption of deciduous teeth could not be confirmed as the patient’s age was 8 years 4 months at the time of the initial examination. To our knowledge, no other reports of such a case involving cleft lip and palate have been published from Japan.

With regard to maxillofacial morphology, Maahs et al. reported Class II malocclusion as the most common (44.4%), followed by Class III (33.3%), and Class I (22.2%) following cephalographic evaluation of 9 patients with

![Fig. 11 Post-retention panoramic radiograph at age 18y6m](image1)

![Fig. 12 Superimposition of pre-treatment and post-treatment tracings on cephalometric radiographs](image2)

Solid line: pre-treatment at age 8y4m. Dashed line: post-treatment at age 12y10m. Dotted line: post-retention at age 18y6m.
Bloch-Sulzberger syndrome. However, several other reports describe development of anterior crossbite as a result of poor maxillary growth. The present patient also showed a long facial type with a Class III skeletal pattern and anterior crossbite at the time of her initial examination. Moreover, despite an almost complete absence of vertical growth of the maxilla, good anteroinferior growth was observed in the mandible. Similarly, Nonaka et al. reported that the amount of forward growth of the maxilla was smaller than that of the mandible and other regions in Bloch-Sulzberger syndrome. In the present patient, it is likely that the cleft lip, alveolus, and palate, as well as the congenital absence of the maxillary permanent teeth, affected the maxillofacial morphology and pattern of maxillary growth. In terms of change during the retention period, almost no growth or development of the jawbone was observed, despite the patient entering the period of growth and development; however, the mandible did rotate clockwise. This was probably due to tooth eruption owing to the patient’s long facial type, which may have contributed to improving the jaw relationship and occlusal stability of the anterior teeth.

The orthodontic treatment provided in the present patient was considered necessary in terms of both function and esthetics owing to spacing of the dentition due to multiple congenitally absent teeth and skeletal anterior crossbite arising from cleft lip, alveolus, and palate. Moreover, any treatment plan for conservative and prosthetic treatment of spacing due to dental morphological anomalies and hypodontia, as well as any in-depth treatment strategy for the prognosis of the remaining deciduous teeth, must be determined from a long-term perspective and executed in cooperation with other departments. However, if caution is paid to space management and growth and development, the treatment mechanics for skeletal anterior crossbite could be beneficial in patients with cleft lip, alveolus, and palate. Good treatment results have been achieved at our department by means of anterior and lateral expansion of the maxillary dentition with expansion arches and by encouraging forward growth of the maxilla using maxillary protraction appliances in such patients. However, the present patient had multiple congenitally absent teeth, which left only few permanent teeth to act as anchors. Therefore, forward displacement of the maxilla was not included in the treatment strategy. Moreover, only the minimum forward expansion necessary to achieve anterior tooth coverage was performed, as expansion of the dentition exacerbates spacing. If prosthetic and restorative treatments are effective for crown morphological anomalies and spacing, and if planned dental treatment is performed after completion of active treatment, functional and esthetic improvements should be achievable.

At 6 years after active treatment, mild crossbite recurred and spaces developed between the mandibular deciduous central incisors; however, occlusion remained mostly stable and esthetically acceptable. In addition, the remaining deciduous teeth showed no noticeable root absorption or mobility, suggesting that treatment objectives can be achieved with planning and techniques in line with routine orthodontic treatment. Long-term retention management will be continued in close cooperation with the departments of plastic surgery and pediatric dentistry.

**Conclusion**

We performed orthodontic treatment, fitted prostheses, and provided restorative treatment in a patient with Bloch-Sulzberger syndrome and cleft lip and palate during the early mixed dentition period. We reported the case after a subsequent 6-year retention phase that included the period of pubertal growth, during which time occlusal stability and esthetics were maintained.
References


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