Case Report

Anomaly of cervical vertebrae found on orthodontic examination: 8-year-old boy with cleft lip and palate diagnosed with Klippel-Feil syndrome

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ABSTRACT We incidentally encountered an anomaly of the cervical vertebrae during orthodontic examination of an 8-year-old boy with cleft lip and palate. Klippel-Feil syndrome was diagnosed by the orthopedic specialist to whom he had been referred for detailed examination. This case report shows the results of axial multislice-helical computed tomography with three-dimensional reconstructions of the cervical vertebrae. Furthermore, we evaluated the frequency of congenital anomalies on lateral cephalogram in patients with cleft lip and palate or cleft palate and non-cleft controls from among all patients in our orthodontic clinic. (*Angle Orthod.* 2010;80:975–980.)

KEY WORDS: Klippel-Feil syndrome; Cervical vertebrae; Cleft lip and palate

INTRODUCTION

Klippel-Feil syndrome (KFS) was first reported by Klippel and Feil[1] and is mainly characterized by congenital fusion of at least two of seven cervical vertebrae in the cervical spine, with limitations to movement of the head or neck, a short neck, and a low posterior hairline.[2] In addition, fusion or anomaly of the vertebrae may be apparent in the thoracic or lumbar spine. KFS is occasionally associated with cleft lip and palate (CLP) and frontonasal malformation.[3] Other findings include several defects, such as deafness, congenital heart disease, and renal anomalies.[4] The incidence of KFS is estimated as 1:40,000–42,000, and 60% of cases occur in females.[5] If the patient with KFS shows high-risk patterns of fusion of the cervical vertebrae and spinal motion, even minor trauma may induce sudden neurological injury or, at worst, death.[6] When the symptoms of a patient with KFS are minor, these symptoms may be overlooked. Thus, the patient may not be diagnosed with KFS and may lead a normal life. Conversely, dentists may incidentally encounter an anomaly of the cervical spine associated with KFS on radiological examinations. This case report describes an anomaly of the cervical vertebrae that was first found on orthodontic examination of an 8-year-old boy with CLP.

CASE REPORT

An 8-year, 1-month–old boy was referred to our private orthodontic clinic from a hospital of plastic surgery with a surgically treated unilateral left-sided CLP. He had been delivered at term after a normal pregnancy and labor. He was his mother's first child and weighed 2825 g, with a length of 46 cm, at birth. At the ages of 6 months, 18 months, and 3 years he had undergone corrective surgeries (cleft lip repair, cleft palate [CP] repair using the ''push-back'' method, and lip and nose revision, respectively) at the previously mentioned hospital of plastic surgery. At the time of first meeting, there was a possibility that he would need to undergo bone grafting in the near future, although it had not been performed to date. The child had no other systemic disorders, and no congenital anomalies were known among relatives on either the maternal or paternal side.

Physical examination revealed a short neck, low posterior hairline, partial limitation of movement of the neck, and protrusion of the lower lip (Figure 1). The child also showed an anomaly of the left ear, although otorhinolaryngological examination revealed normal auditory sensitivity. He was small in stature (116.2 cm) compared to the standard size for a child of his background (8-year-old Japanese boy: 128.3 $\pm$ 5.41 cm), although his body weight (30.4 kg) was

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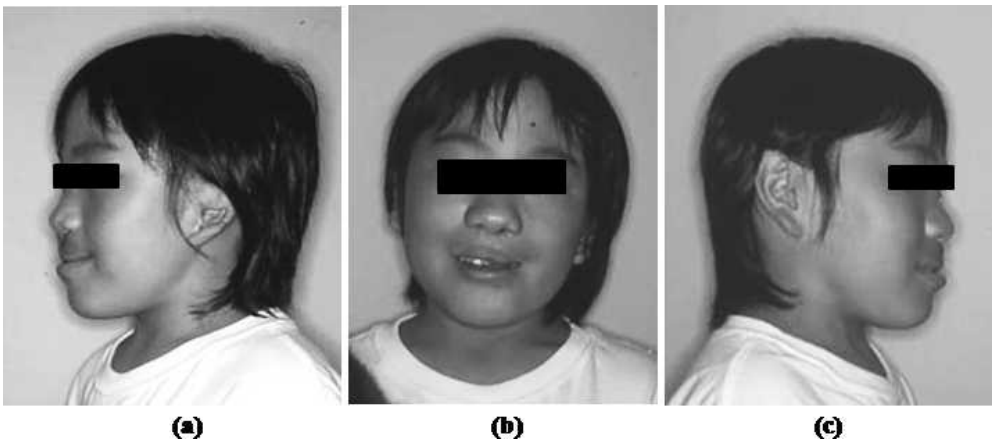

Figure 1. Facial photographs of the patient: (a) Left; (b) frontal; (c) right.

within the standard range (8-year-old Japanese boy: 27.7 $\pm$ 5.48 kg). No mental retardation was apparent.

Intraoral examination revealed surgical scars between the upper left central incisor and upper left deciduous canine. Part of the tooth that seemed to be the upper left deciduous lateral incisor was found adjacent to the scar (Figure 2). A few occurrences of secondary caries were found, although decayed teeth had been treated previously. The patient showed an anterior cross bite with a Class III molar relationship on both right and left sides.

Panoramic radiograph revealed congenital absence of the upper left lateral incisor (Figure 3a). Cephalometric analysis using lateral cephalogram (Figure 3b) showed normal maxillary and mandibular positions (SNA, 77.0°; SNB, 76.0°). The maxillary incisors were lingually inclined (U1 to SN plane, 100.5°). Based on the results of these examinations, the patient was diagnosed with functional Class III malocclusion with an anterior cross bite due to lingual inclination of the central incisors.

Examination of the lateral cephalogram from this patient incidentally revealed anomalous cervical vertebrae in the cervical spine. The shape of the spinous processes (Figure 4a) clearly differed from that of an 8-year-old boy with whom we regularly dealt (Figure 4b). The shapes of spinous processes for the third and fourth cervical vertebrae (C3 and C4) were obscure, and the spinous processes of the fifth and sixth cervical vertebrae (C5 and C6) appeared to be fused. The child's parents had never been informed of any anomaly of the cervical vertebrae at the hospital where he had undergone surgeries. We referred this patient to an orthopedic specialist for detailed examination of the cervical vertebrae. Axial multislice-helical computed tomography with three-dimensional recon-

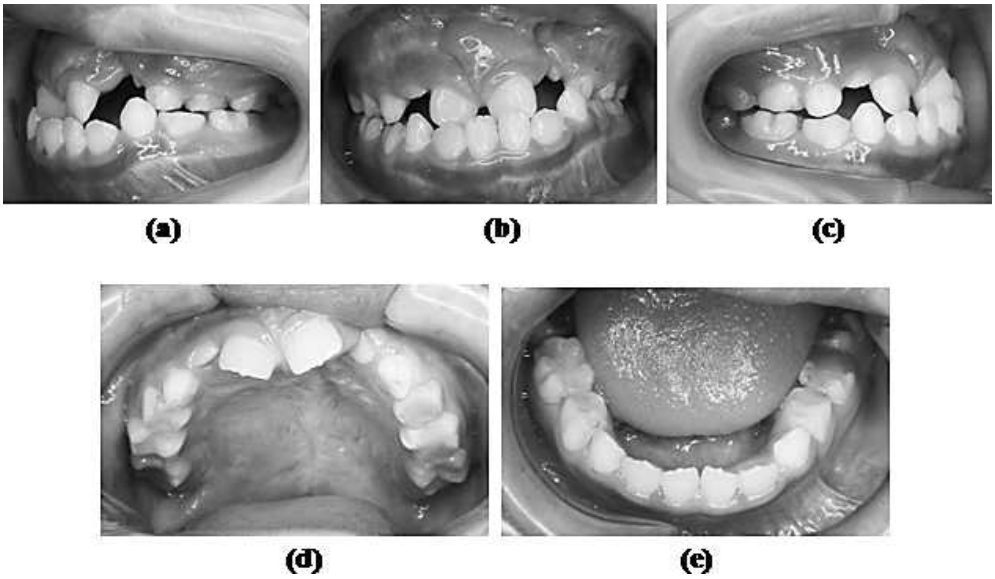

Figure 2. Intraoral photographs of the patient: (a) Left; (b) frontal; (c) right; (d) upper; (e) lower.

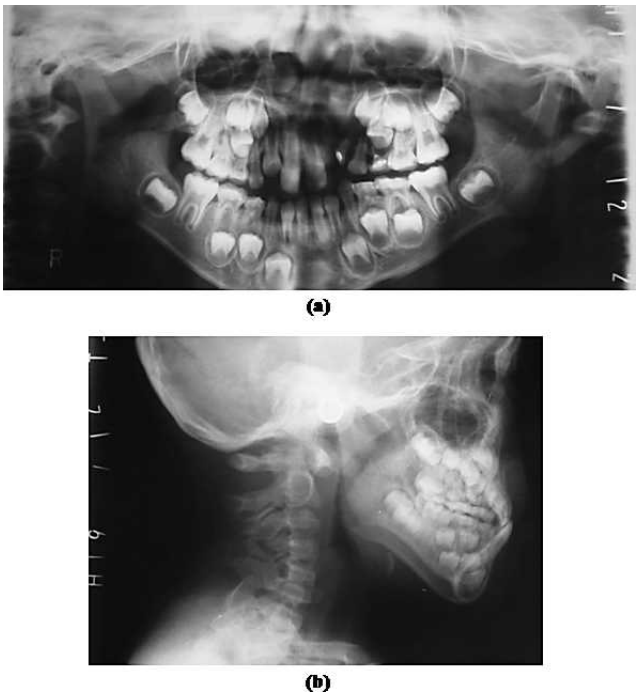

Figure 3. (a) Panoramic radiograph of the patient. (b) Lateral cephalogram of the patient.

structions resulted in diagnosis of KFS. He was found to have no scoliosis, Sprengel's deformity, renal anomalies, deafness, or congenital heart disease, all of which are systemic anomalies occasionally associated with KFS. He received an explanation from the orthopedic specialist that he needed periodic follow-up without therapy, since he presently had no real difficulties in life. However, the possibility that clinical manifestations derived from the neurological disorder might arise in the future was also explained.

Figure 5 shows the three-dimensional reconstructions from computed tomography of the cervical vertebrae for this patient (Figure 5a–c) and a model of normal cervical vertebrae (3B Scientific GmbH, Hamburg, Germany) (Figure 5d–f). Vertebral bodies of C4 to C7 cannot be clearly distinguished from each other, because the spinous processes of all cervical vertebrae except C1 show anomalies. Particularly from the posterior view (Figure 5b), the spinous processes of C2 and C3 seem to be not completely formed, and those of C4, C5, and C6 seem fused. Figure 6 shows characteristic images of tomographic axial sections from each cervical vertebra of the patient. Figure 6 also indicates that the vertebral body and spinous processes of each cervical vertebra are not completely formed.

We began orthodontic treatment using a fixed lingual arch appliance (Figure 7a), as consent for treatment was obtained by the orthopedic specialist. The anterior cross bite was improved using the fixed lingual arch appliance (Figure 7b), and we are awaiting eruption of the permanent canines and premolars in order to place a multibracket appliance.

DISCUSSION

In this report, anomalous cervical vertebrae of the patient were first identified on orthodontic examination. Finding such an anomaly in an orthodontic clinic rather

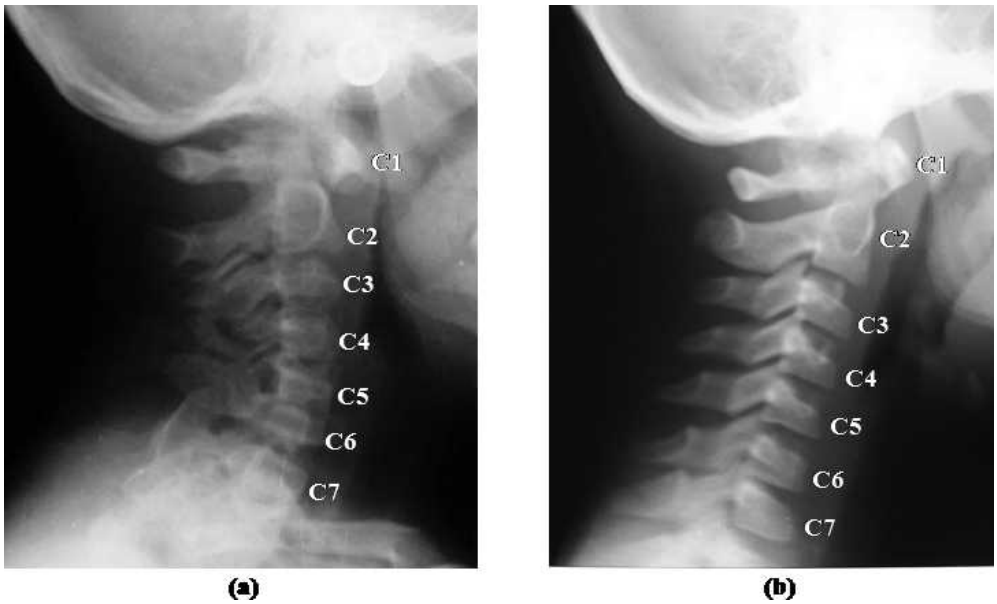

Figure 4. Magnified views of cervical vertebrae on lateral cephalogram. (a) The patient in the present case. (b) A normal boy the same age as the patient in the present case. C1–C7 indicates first through seventh cervical vertebrae.

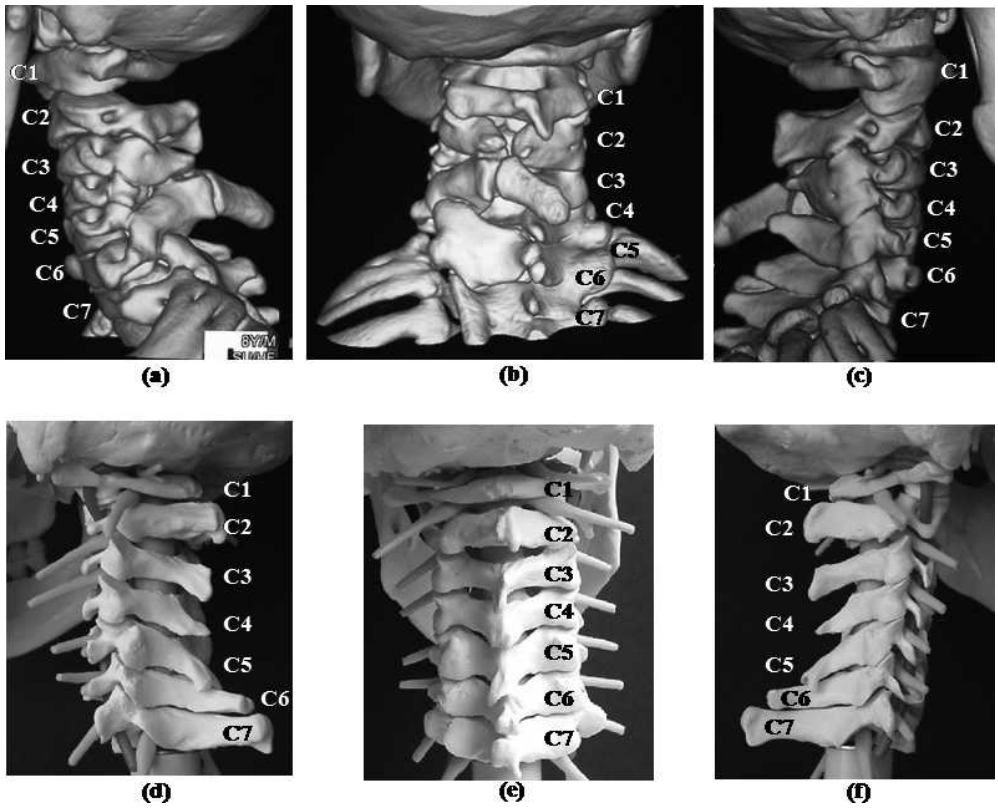

Figure 5. Three-dimensional reconstructions from computed tomography scan of the cervical vertebrae for the patient: (a) left; (b) posterior; (c) right. Anatomical model of normal cervical vertebrae: (d) left; (e) posterior; (f) right. C1–C7 indicates first through seventh cervical vertebrae.

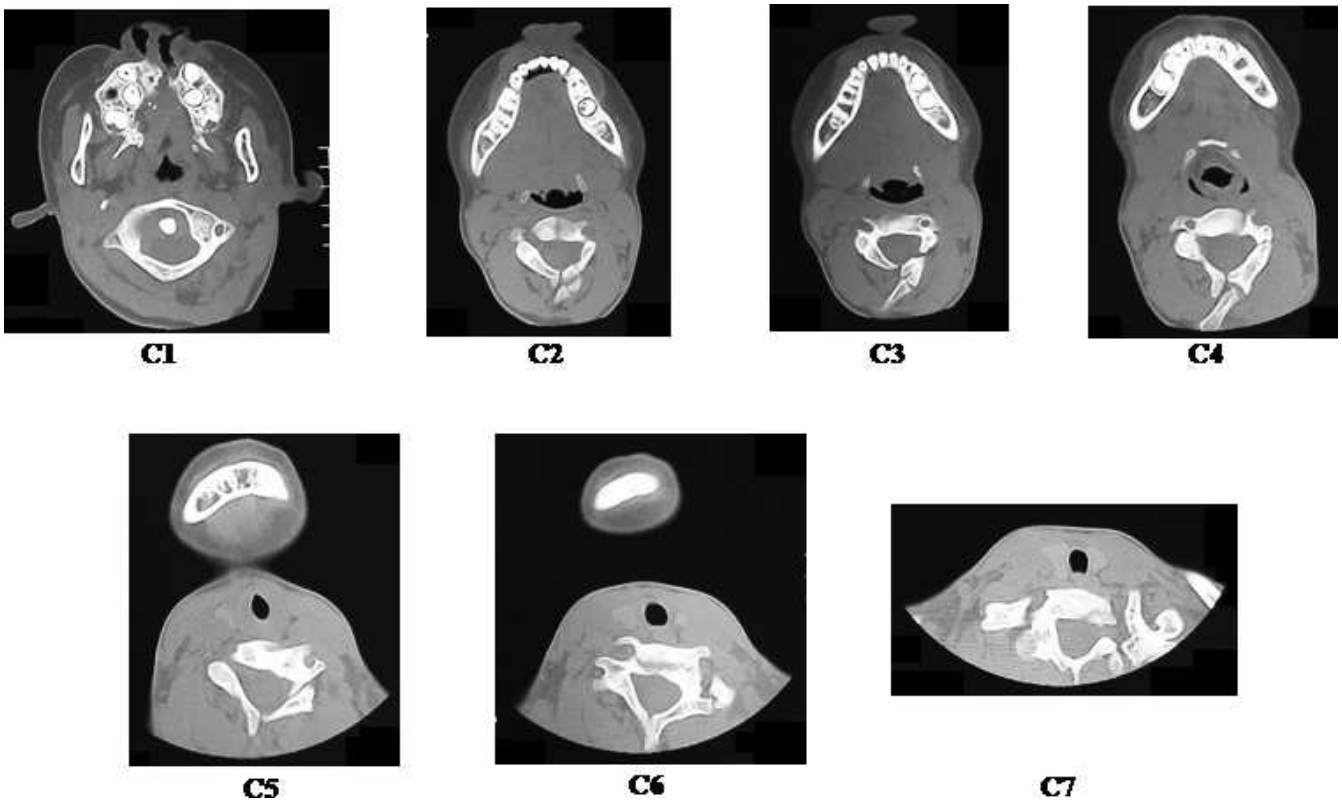

Figure 6. Characteristic images of tomographic axial sections of each cervical vertebra of the patient. C1–C7 indicates first through seventh cervical vertebrae.

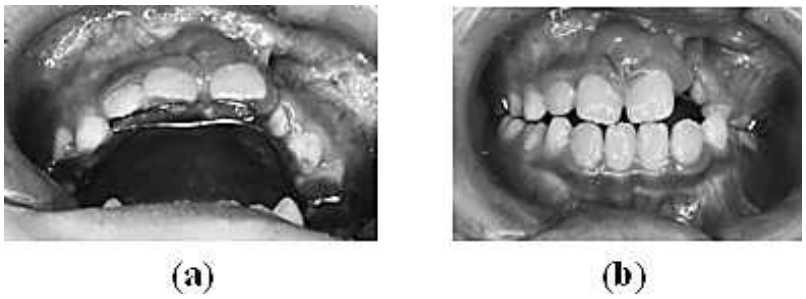

Figure 7. Intraoral photographs of the patient (a) during treatment by a fixed lingual arch appliance and (b) after the removal of the appliance.

than in a medical hospital is probably relatively rare. The patient was diagnosed with KFS by the specialist to whom we referred him.

Since there is a high prevalence of syndromes associated with CLP or CP, it is important to have a comprehensive clinical history and further genetic testing to differentiate syndromic cases from nonsyndromic cases. Furthermore, a careful differential diagnosis of KFS, compared with other syndromes, especially Goldenhar syndrome, Wildervanck syndrome, and Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome, is needed, since many other syndromes may present fusion of the cervical vertebrae as a part of their clinical features. Goldenhar syndrome is characterized by a triad of anomalies consisting of epibulbar dermoids, accessory auricular appendages, and pretragal fistula.[7] It is associated with vertebral, other skeletal, and systemic anomalies. Wildervanck syndrome consists of fused cervical vertebrae, sensorineural hearing loss, and abducens paralysis with retracted globes.[8] CLP or CP is often present in patients with these two syndromes. MRKH syndrome is a congenital condition characterized by vaginal agenesis, with other Müllerian duct abnormalities and normal secondary sexual characteristics.[9] While type I MRKH syndrome is characterized by an isolated absence of the proximal two-thirds of the vagina, type II MRKH is marked by other malformations, including vertebral, cardiac, urologic, and otologic anomalies.[10]

Based on the experience from the present case, using their lateral cephalograms we examined the frequencies of congenital vertebral fusion, a main characteristic of KFS, in 246 patients with CLP or CP and 6578 non-cleft controls from among the 6834 patients seen in our orthodontic clinic from 1978 to 2008. As a result, we found 21 patients, including the patient in this case report, with congenital vertebral fusion (Table 1). Six of 246 patients with CLP or CP (6/

Table 1. Twenty-one Patients with Congenial Vertebral Fusion in our Orthodontic Clinic[a]

Case No.	Gender	Age	Chief complaint	Vertebral fusion	Associated anomalies
1	m	10 y, 7 mo	Crowding	C2 + C3	CP
2	m	10 y, 3 mo	Cross bite	C3 + C4	CP, ASD, PDA, cryptorchism, MR
3	m	23 y, 3 mo	Cross bite	C2 + C3	CLP
4	m	31 y, 6 mo	Cross bite	C2 + C3	CLP
5	m	8 y, 11 mo	Cross bite	C4 + C5 + C6	CLP, VSD
6	m	8 y, 1 mo	Cross bite	C4 + C5 + C6	CLP, KFS
7	m	6 y, 10 mo	Cross bite	Occipital bone + C1	Hydrocephalus, chondralloplasia
8	f	9 y, 10 mo	Maxillary protrusion	Occipital bone + C1	
9	m	12 y, 3 mo	Crowding	Occipital bone + C1	
10	m	8 y, 8 mo	Crowding	C2 + C3	
11	m	9 y, 0 mo	Cross bite	C2 + C3	
12	f	9 y, 3 mo	Maxillary protrusion	C2 + C3	
13	m	10 y, 5 mo	Cross bite	C2 + C3	
14	f	10 y, 8 mo	Maxillary protrusion	C2 + C3	
15	f	12 y, 9 mo	Crowding	C2 + C3	
16	f	33 y, 10 mo	Maxillary protrusion	C2 + C3	
17	f	7 y, 8 mo	Cross bite	C3 + C4	
18[b]	m	8 y, 1 mo	Cross bite	C3 + C4	
19	f	13 y, 2 mo	Crowding	C3 + C4	
20	m	18 y, 0 mo	Cross bite	C4 + C5	
21	f	20 y, 8 mo	Cross bite	C5 + C6	

[a] m indicates male; f, female; CP, cleft palate; ASD, atrial septal defect; PDA, patent ductus arteriosus; MR, mental retardation; CLP, cleft lip and palate; VSD, ventricular septal defect; and KFS, Klippel-Feil syndrome.

[b] Patient No. 18 is the patient described in this case report.

246, 2.44%) and 15 of 6578 non-cleft controls (15/6578, 0.22%) displayed congenital vertebral fusion, representing a significant difference ($P < .01$, χ^2 test). Ross and Londsay[11] also examined lateral cephalograms of 342 children with CLP or CP and 800 non-cleft controls and reported that the frequency of congenital vertebral fusion was 4.01% (14/342) in children with CLP or CP and 0.75% (6/800) in non-cleft controls. That investigation and the present study clearly indicate the high prevalence of CP in patients with congenital vertebral fusion compared to non-cleft controls.

Furthermore, the frequency of anomalies involving the cervical vertebrae reportedly increases with increasing severity of cleft type.[2] However, the relationship between frequency of anomalous cervical vertebrae and the severity of cleft type was not shown in the present study. We need to increase the sample size of patients with CLP or CP who show anomalies of the cervical vertebrae.

In the eighth and ninth weeks of embryonic life, the tongue of the fetus lies between the vertically oriented palatal shelves. In the ninth or 10th week, the head is lifted from the pericardial region, then the mandible and tongue drop to permit the right and left palatal shelves to meet and fuse in the midline, and finally the soft palate is formed. If the fetus has a fusion of cervical vertebrae, the mandible remains compressed against the chest and forces the tongue to continue to lie between the palatal shelves, resulting in incomplete closure of the palate.[12] The high prevalence of CP in patients with congenital vertebral fusion indicates the developmental interactions by which a primary defect in one organ system can lead to secondary defects in contiguous structures.

Of note is the finding that two patients (cases 4 and 9 in Table 1) with congenital vertebral fusion showed associated congenital heart disease. The frequency of congenital heart disease for KFS is reportedly 8.75%, significantly higher than the 0.6% frequency among liveborn children or the 0.2% frequency among schoolchildren.[6] Although the patients in Table 1 (other than the patient in case 18) were not diagnosed with KFS, further study of the associations between vertebral fusion and congenital heart disease is needed.

The present case indicates that important anomalies that may have been missed in the medical hospital can be revealed incidentally on radiological examinations in the orthodontic clinic. Patients with mild cases may not show any severe clinical symptoms at presentation but may develop neurological symptoms secondary to degenerative disc disease of the adjacent mobile segments, spinal instability from hypermobility or from trauma, or spinal stenosis in later decades of life.[13–15] The age at which neurological symptoms appear depends on the level of fusion of the cervical vertebrae.[6] Fusion of the first and the second cervical vertebrae (C1 and C2) tends to produce symptoms in the first decade of life, while fusion of the second and third cervical vertebrae (C2 and C3) is associated with neurological symptoms in the third decade. Most symptoms, regardless of the location of the lesion, appear prior to 30 years of age. Early diagnosis of KFS is critical in determining the risk of other associated diseases at presentation and secondary neurological symptoms in the future.

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