CASE REPORT

Congenital lateral cleft palate with unilateral craniofacial microsomia and lateral ankyloglossia

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SUMMARY

Craniofacial microsomia is a group of anomalies that occur due to developmental defects in the first and second branchial arches during the embryological period. The disease has various craniofacial and extracraniofacial presentation patterns. Lateral cleft palate is distinct from the most common palate clefts because it is located lateral to the uvula. The presence of a lateral cleft palate has been very rarely reported in the literature, and the aetiopathogenesis of the disease is still not well understood. We aimed to report a case of the coexistence of lateral cleft palate and craniofacial microsomia and discuss the aetiopathogenesis of these diseases.

BACKGROUND

Craniofacial microsomia (CFM) is a group of deformities characterised by various degrees of hypoplasia of the bones and soft-tissues in the craniofacial area, especially in the mandible and outer ear. The maxilla, zygoma, temporal bone, cervical vertebrae, masticatory muscles, tongue and palate can be affected. The location of involvement and severity of the disease may lead to varying problems, such as hearing loss, facial asymmetry, chewing and swallowing disorders, and velopharyngeal insufficiency (VPI). CFM is the most common head and neck anomaly after cleft lip and/or palate (CL/P), and its prevalence is 1 in every 5600 births.¹

Cleft palate (CP) is a congenital abnormality characterised by fissures in the midline between the oral and nasal cavities anywhere from the incisive foramen to the uvula. It may be accompanied by CL if it is located anterior to the incisive foramen. If it is unrepaired, it may cause respiratory and chewing problems, eustachian tube dysfunction leading to middle ear problems and/or VPI. CL with or without CP is observed in 1.25 in 1000 births, although prevalence differs by race. CP is seen in 3.1 in 10000 births, without any observed racial differences.²

Only three lateral CP (LCP) cases have been presented in the literature, and no aetiological factors have been detected in these cases.^{3–5} We aimed to present a unique case of unilateral CFM and LCP.

CASE PRESENTATION

A 12-month-old female patient presented with ankyloglossia and CP. Her mother had consanguineous marriage. There were no congenital anomalies

in the patient's family history. Her mother had no known diseases and no history of drug use or X-ray exposure during pregnancy. There was no history of trauma to cause the CP, but the patient's mother immigrated to Turkey during pregnancy because of the war in Syria. A physical examination revealed haemifacial asymmetry on the right side and mild cupping on the external ear. The findings of the other systemic examinations were normal, except that hearing loss was detected. Lateral ankyloglossia and mandibular fusion were observed during an examination of the oral cavity. Unlike most cases of CP, the cleft was not on the midline; it was on the right side of the uvula, and the cleft was limited to the soft palate. The right palatal tonsilla and tonsillar fossa were hypoplastic ([figure 1](#)). Maxillofacial tomography revealed hypoplasia of the middle ear, external auditory canal, right mandibular coronoid process and mandibular condyle; loss of aeration and sclerosis in mastoid cells; and a fusion defect in the posterior processes of the cervical vertebrae ([figure 2](#)). The patient was diagnosed with CFM, and she underwent surgical treatment for CP and ankyloglossia.

TREATMENT

The patient underwent operation under general anaesthesia. Following the placement of the Dingman retractor, an epinephrine solution (1/200 000) was injected into the palate and right lateral tongue. The nasal and oral mucosa junctional line of the cleft margin was incised. The tensor veli palatini and levator veli palatini muscles were exposed via bilateral submucosal dissection. The muscles on the cleft side were observed to be hypoplastic. The muscles were released from the hard palate insertion points. The nasal mucosa was sutured. Soft palate muscle repair was achieved with a radical intravelar veloplasty technique ([figure 3](#)). The oral mucosa was sutured. The tongue was released from the mandibular fusion zone. The defect area was repaired with a split-thickness skin graft taken from the lateral aspect of the left thigh.

OUTCOME AND FOLLOW-UP

After the operation, the patient was discharged without any pathology and followed up by the outpatient clinic. At 6 months postoperatively, the affected side of the tongue and soft palate were healthy, and the patient displayed no symptoms.

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Figure 1 Preoperative view of a 1-year-old girl (A) Right haemifacial asymmetry and subcutaneous soft-tissue hypoplasia. (B) Mild cupping in the right outer ear. (C) Normal left outer ear, (D) Right lateral ankyloglossia and lateral cleft palate. (E) Lateral ankyloglossia.

DISCUSSION

CFM is caused by the insufficient development of the first and second branchial arches during the fourth week of pregnancy. This is explained by two most commonly accepted theories: (1) haematoma caused by stapedia arterial bleeding and (2) impaired growth

Figure 2 Three-dimension CT image of the right craniofacial microsomia patient. (A) Short right ramus mandibula. (B) Fusion defect in posterior cervical vertebrae. (C) Right mandibular ramus, coronoid process, condyle and external acoustic meatus hypoplasia (D) Normal left mandibula end external acoustic meatus.

Figure 3 Right lateral cleft palate patient view (A) Preoperative. (B) Perioperative, right tensor veli palatine and levator veli palatine muscles. (C) Immediate postoperative. (D) Postoperative 6 months.

and migration of neural crest cells.^{6,7} The disease is usually unilateral; however, bilateral occurrence has been reported in 5%–30% of cases.⁸ The craniofacial and extracraniofacial spectrum of the disease is wide and variable. The progression and the severity of the disease are thought to be directly associated with mandibular deformity.⁹ The affected organs are graded by Vento *et al* according to the orbit, mandible, eye, facial nerve and soft tissue (OMENS) criteria to provide interclinic orientation.¹⁰ Horgan *et al* described the OMENS (+) classification for the assessment of craniofacial and extracraniofacial involvement.¹¹ These classifications are based on the Pruzansky classification system, which grades the mandibular involvement that Kaban *et al* have modified.^{12,13} In the present case, normal eye size and position, short mandibular ramus, mild cupping in the external ear, normal facial nerve and minimal subcutaneous soft-tissue deficiency were found (O₀M₁E₁N₀S₁). There were also CP, ankyloglossia, hypoplasia in the middle ear, and aeration loss and sclerosis in mastoid cells. A fusion defect in the posterior processes of the cervical vertebrae was detected as the only extracraniofacial finding.

Ankyloglossia (tongue-tie) is the result of inadequate degeneration of the anterior two-thirds of the tongue that develop from the first branchial arch.¹⁴ The most commonly seen form is the elongation of the frenulum to the tongue tip. The prevalence varies from 3.8% to 4.8%.¹⁵ Chen *et al* reported that 43.6% of CFM patients had associated tongue anomalies. They also classified tongue anomalies into four groups: Tng 0=normal, Tng 1=midline ankyloglossia or unilateral hypoplasia, Tng 2=lateral ankyloglossia with fusion to the mandible or severely bifid tongues and Tng 3=aglossia.¹⁶ The tongue anomaly of the patient presented in this article was classified as Tng 2. Although coexistence of anterior ankyloglossia and CP due to a TBX22 mutation has been previously reported, the coexistence of lateral ankyloglossia and CP has not yet been reported.¹⁷

Conductive hearing loss is frequently seen in patients with CFM due to anomalies in the outer and middle ear. Despite the fact that the inner ear does not develop from the first and second

branchial arches, sensorineural hearing loss is more frequently seen in patients with CFM than in patients with other craniofacial and congenital anomalies.¹⁸ Although the severity of the hearing loss is directly related to the severity of the involvement of the mandible and ear, it has been reported that 15% of patients with a normal ear phenotype have hearing loss.¹⁹ In this report, conductive hearing loss was present, along with involvements of the mandibular bone, external ear, external auditory canal, middle ear and soft-tissue.

VPI without CP may be seen in patients with CFM due to hypoplasia of the soft palate muscles or inadequate innervation (hypodynamic palate).²⁰ In a study by Funayama *et al*, the hypodynamic palate was reported in 24 out of 48 CFM patients, and moderate VPI developed in seven of these patients.²¹ In these cases, hypoplasia of the palatal tonsil and fossa in combination with CP were considered predisposing factors for VPI.

Beginning in the sixth gestational week, the lateral palatal shelves originate from the maxillary prominence of the first branchial arch by descending obliquely on both sides of the tongue.¹⁴ They then horizontalise and fuse from anterior to posterior at the midline of the tongue by ascending beside the tongue. CP is caused by incomplete development of the lateral palatal shelves.²² Genetic factors, maternal teratogenicity exposure, folic acid deficiency, high altitude and advanced paternal age may be responsible for this anomaly. In this patient, parental consanguinity, young maternal age and possible poor conditions during the war (such as vitamin deficiency and teratogenicity exposure) may be among the risk factors.

A recent article from a single institution published in the *American Journal of Medical Genetics* in 2017 by Cohen *et al* reported 11 cases of midline CP in a cohort of 89 patients.²³ Fan *et al* suggested that CL±P and CFM might be caused by impaired neural crest cell development and migration.²⁴ In the literature, there are only a few reports of the coexistence of CFM and CP, and no association between CFM and LCP has been reported.^{21 25} In this case, the pathogenesis of LCP may be explained as follows: because the palatal tonsil and fossa are located on the cleft side, hypoplasia of these structures may be caused by insufficient development of first and second branchial arches, suggesting that the CP is a part of the CFM.

Erdogan *et al* provided the first report of LCP.³ Then, Isik *et al* presented a case of lateral palatal synechia with LCP.⁴ In the same study, it was suggested that the lateral palatal synechia, which is a residue of the buccopharyngeal membrane, might cause LCP by preventing the anterior movement of the tongue. Finally, Kahraman *et al* stated that the aetiology of LCP is unknown.⁵ In all four

reported cases of LCP, including our case, LCP was located in the soft palate, and the genders of the patients and the laterality of the patients' lesions were similar. In the case outlined in this article, the LCP was thought to be a part of the CFM, which may be explained by different theories. This is the first case reporting the coexistence of LCP and lateral ankyloglossia in CFM. Further genetic studies and case series are needed to clarify whether this coexistence is a new anomaly or part of a sequence.

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Learning points

- Craniofacial microsomia (CFM) is often present with mandibular and external ear involvements. However, craniofacial and extracraniofacial involvement of the disease may occur at different degrees. For this reason, patients should be evaluated systematically in addition to craniofacial examination.
- CFM, cleft palate (CP) and ankyloglossia are considered to be the result of the developmental defect of similar embryological regions. Thus, a detailed intraoral examination should be performed in CFM cases.
- Lateral CP (LCP) has been rarely reported in the literature and the aetiology of the disease is unknown. In this case, it is thought that CFM may play a role in LCP aetiology, unlike CP.

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