Case Report: Patent Mandibular Symphysis with Congenital Absence and Ankyloglossia

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ABSTRACT The mandible develops prenatally as left and right halves (hemimandibles) that meet at a suture in the anterior midline. This suture normally is obliterated in the first year of life. We describe a 4-year-old girl in whom (A) this suture (*symphysis menti*) is only partially fused, (B) the primary lower left central incisor is congenitally absent (and also its permanent successor), and (C) there is pronounced ankyloglossia. These midline problems share a common etiology, namely incomplete fusion of the halves of the first branchial arch. No cause is suggested, but the embryological problem seems to stem from inadequate streaming together of the mesodermal cores of the first branchial arches. Similar cases with the dental and bony aspects of this condition should be identifiable in skeletal remains. *Dental Anthropology* 2009;22(2):54-58.

As is well known, the human mandible develops from the first branchial arches, and it ossifies before birth as separate left and right hemimandibles that meet ventrally at the mandibular symphysis (*symphysis menti*). Overviews of the embryology of the mandible are provided in Arey (1965), Corliss (1976), Scheuer and Black (2000), and most textbooks on mammalian embryology. At birth (Fig. 1), the mandibular midline is patent, though this suture normally fuses and is obliterated during the first year of life (Fig. 2). After symphyseal fusion, the mandible is rigid, and masticatory forces from the working side are transmitted through the chin to the balancing side. The need for resistance to torsion has been cited as a cause of development of the uniquely human chin (*e.g.*, Sicher, 1947; DuBrul and Sicher, 1953; Schwartz and Tattersal, 2000), which occurs principally in adolescence (Ricketts, 1972).

Rarely, the mandibular symphysis fails to fuse, and the present case report describes such an anomaly in a 4-year-old girl who also exhibits congenital absence of a primary incisor in combination with ankyloglossia. Persistent patency of the *symphysis menti* is an easily observed condition in skeletal material, so this report may be of interest to skeletal biologists. Moreover, it may stimulate readers to share similar findings.

CASE DESCRIPTION

The subject is a healthy American black girl who was 4.0 years of age at examination. She was seen in a pediatric dental setting for routine restorative work. Examination revealed 19 primary teeth with apparent congenital absence of the primary mandibular left central (tooth 71 in the FDI system; tooth O in the Universal system). Decay was seen clinically on the occlusal surface of all 8 primary molars and interproximally on bitewing radiographs. Mesial caries can be seen radiographically on the primary left maxillary central incisor (tooth F).

The girl was treatment-planned for stainless steel crowns on the 8 primary molars, a mesial lingual resin on tooth F, and a lingual frenectomy. Due to her young age, her multiple treatment needs, and her acute situational anxiety, it was recommended that the procedures be performed under general anesthesia. However, the patient moved out of state before treatment could be performed.

The prominent frenum that ties the tip of the tongue to the floor of the mouth (and limits tongue mobility) is

Fig. 1. Occlusal view of the left and right hemimandibles of a near-term infant showing (A) the independent development of the two halves of the formative mandible and (B) the rough surfaces where they meet at the ventral midline.

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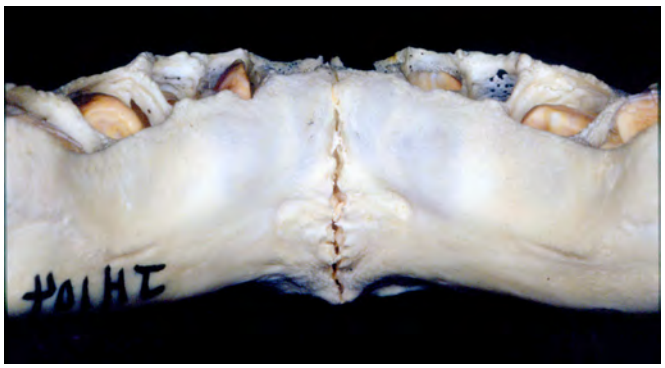

Fig. 2. Lingual view of a deceased infant's mandible showing incomplete but on-going fusion of the hemimandibles.

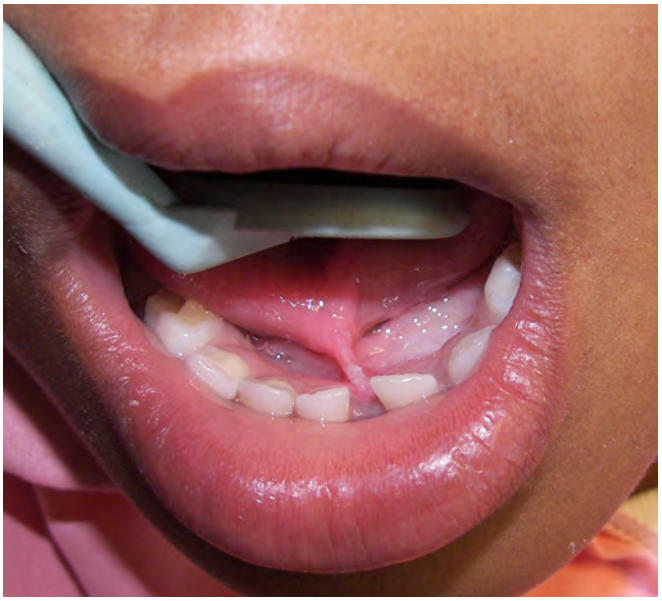

Fig. 3. Extraoral photograph of the girl showing the partial ankyloglossia with prominent soft tissue attachment (frenum) encroaching on the space of the missing left central primary incisor.

an obvious feature in this girl (Figs. 3 and 4), but what caught our attention was the persistent mandibular suture that is evident on X-ray (Fig. 5). The hemimandibles are effectively fused together, but patent remnants of the suture are evident on the cranial and caudal aspects of the midline, and the open suture extends at least half a centimeter down through the mandibular alveolus.

Mandibular symphysis

Textbooks routinely note that the suture between the two hemimandibles fuses "within the first year of life," though we have been unable to find more definitive statistics. Molleson and Cox (1993) studied the Spitalfields collection and found that the two hemimandibles were always separate before 3 months of age, but most had initiated fusion by 6 months of age. At 4 years of age, the girl's suture described here is clearly delayed, if indeed fusion is still ongoing.

Ankyloglossia

The tongue develops from the presumptive floor of the mouth (branchial arches I and III), and endoderm immigrates around the developing tongue during post-conception week 5. The apoptosis (selective cell death) of this endoderm is necessary for the mobile region of the tongue to be freed from the base (Fig. 5). Some of these cells persist in the midline and form the frenulum of the tongue, which is the membranous strand that ties the anterior, mobile portion of the tongue to the floor of the mouth. It is not uncommon for this tissue (the lingual frenum) to be prominent in infants and children, which can limit tongue mobility. This typically is of little concern because the frenulum regresses and stretches with age, particularly during infancy (*e.g.*, Wright, 1995; Lalakea and Messner, 2003). However, the prominence and extent of the ankyloglossia in this girl (Figs. 3-4) clearly is outside of normal limits. This is obvious (Fig. 4), where the girl is incapable of protruding here tongue because it is tethered to the floor of the mouth, with the frenum being continuous with the lingual gingiva (Fig. 3). On the other hand, the girl's labial vestibule (*i.e.*, separation of the lower lip from the gingival ridge) is normal, and there is no hint of notching or clefting of either lip.

True ankyloglossia (fusion of the whole tongue to the floor of the mouth) is a particularly rare event—to the point that clinician's commonly use "ankyloglossia" to refer to the lesser "tongue tie" condition, where it

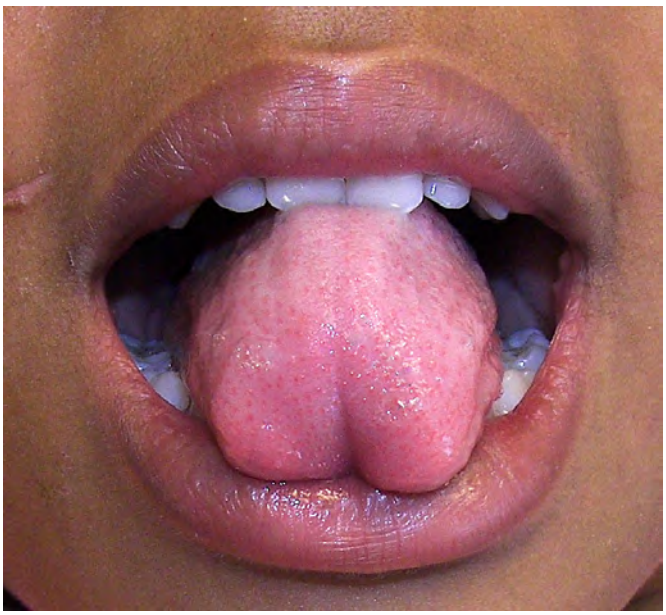

Fig. 4. Extraoral photograph of the girl showing how her ankyloglossia prevents normal tongue movement.

is just the persistent midline fibrous band that limits tongue mobility (which can interfere with chewing, swallowing, and speech). For example, Kotlow (1998) proposed a 5-grade scale to score the extent of ankyloglossia, ranging from a normal range of function (grade 0) up to "complete" ankyloglossia (grade IV) where less than 3 mm of the ventral tip of the tongue is mobile. This system may be useful clinically, but it ignores the developmental scenario where apoptosis (selective cell death) fails altogether and the tongue remains fused to the floor of the mouth.

Clinically, treatment of ankyloglossia (*i.e.*, excision of the frenum) seems unwarranted in most cases. Treatment should be limited to cases with documented speech, functional, occlusal or periodontal problems. The tongue is always short at birth, but, with growth, the tongue becomes longer and thinner at the tip. Many cases are self-correcting (due to frenum stretching and tongue growth), which accounts for the comparatively low frequency of ankyloglossia in adults.

Congenital absence

Figures 3 and 6 show that the lower left primary central incisor is absent. This tooth normally emerges around 6 to 8 months of age (Tanguay *et al*., 1984), and there is no suggestion from inspection of the alveolus that it might have been exfoliated in this 4-year old. Moreover, the mother stated that this tooth was never present, so we conclude that the tooth is congenitally absent. The claim for absence of the primary left central incisor is supported by the congenital absence of its permanent successor (Fig. 6). Since a primary tooth's successor develops from a lingual offshoot of the primary tooth bud (*e.g.*, Avery, 1994), the absence of a primary tooth greatly increases the risk of its successor also being absent (*e.g.*, Grahnén and Granath, 1961).

We suspect that it more than coincidental that the prominent frenum (Fig. 3) is located right at the site of the incisor's congenital absence. It is speculative, but the developmental disorder that failed to remove the presumptive frenum from the ventral midline may also be responsible for the incisor's agenesis (or aborted development).

Dahlberg (1945, 1951) probably was the first to describe the reversal of the morphogenetic field in the mandibular incisors, where the central incisor is smaller, and more variable metrically and morphologically than the lateral incisor—but he provided no interpretation of the reversal, which is unique (all other fields exhibit greater variability of the distal tooth). Other studies (reviewed in Endo *et al*., 2007) suggest that simple hypodontia is tied to craniofacial issues of development, such as short cranial base and maxillary lengths, mandibular prognathism, and diminished anterior facial height. Kjaer (1980) suggests that the poorer vascularity at the *symphysis menti* enhances the variability of the

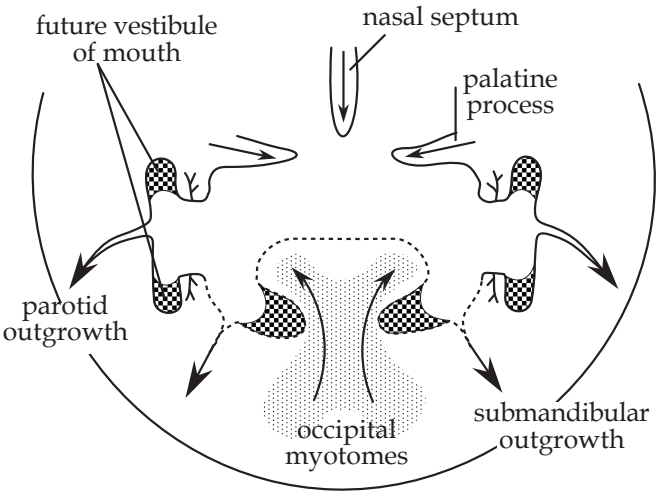

Fig. 5. Schematic cross-section of the developing embryonic mouth showing the pathways taken by the formative tissues. The epithelial surface shown as a solid line is ectodermal; the epithelial surface marked as a dashed line is endodermal. The cross-hatched epithelial area degenerates, forming the vestibule of the mouth and freeing the tongue from the floor of the mouth. Modified from Snell (1975).

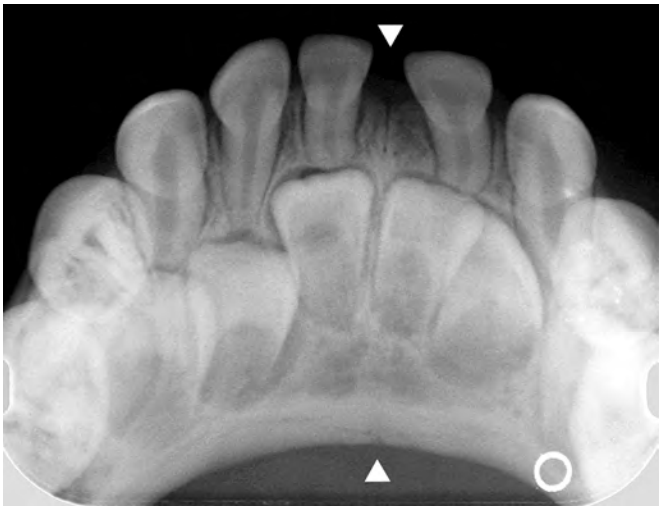

Fig. 6. Occlusal radiograph of the girl's mandibular anterior region. The *symphysis menti* (top arrow) is patent for several millimeters in the cranial region of the midline. A 3-4 mm patency also is visible on the caudal margin of the symphysis (bottom arrow). The primary left central incisor is congenitally absent—as is its permanent successor—but the other three permanent incisors are forming and are completing crown formation. The missing permanent incisor would have been located almost directly beneath the open suture visible between the primary incisors. (Note that orientation is reversed in this radiographic view, so the child's left quadrant is to the right side of the picture.)

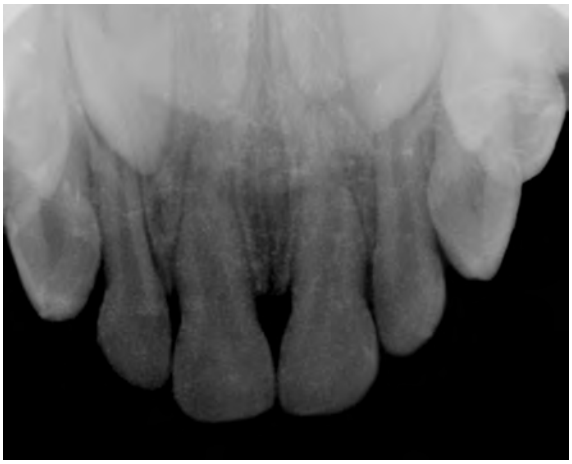

Fig. 7. Occlusal radiograph of the girl's maxilla showing complete tooth formation and eruption of the four primary incisors as well as mineralization of the four succedaneous incisors.

central incisor.

The present case may, however, reflect a local rather than systemic problem, where whatever caused the ankylosis (failure of apoptosis) also caused (A) failure of the incisor to form and (B) arrested fusion of the suture. These defects all involve formation of the first branchial arch, and their common locus at the midline may be due to inadequate mesodermal penetration into this arch's midline (Godbersen *et al*., 1987). Similar cases (reviewed in Eastlack *et al*., 2000) report additional midline defects in various individuals, such as dermoid cysts, ectopic salivary glands, bifid tongue (or aglossia), congenital absence of mandibular incisors, and cleft lower lip. Again, these conditions suggest incomplete fusion of the first branchial arch as the common etiological problem (Gardner and Moss, 2005; Mendis and Moss, 2007).

Syndrome

We initially speculated that this girl's triad of (1) missing central incisor, (2) ankyloglossia and (3) persistent symphyseal suture constituted some sort of midline developmental defect, with incomplete left-right differentiation of the face. The symptoms probably involve a simpler, less dramatic situation. Scrutiny of the girl's maxilla revealed nothing unusual: Both maxillary incisors (primary and permanent) are of normal size and morphology, and the intermaxillary suture is obvious (Fig. 7). These left-right features argue against a problem with embryonic division as found in various sorts of holoprosencephaly (*e.g*., Krauss, 2007; Shiota *et al*. 2007).

OVERVIEW

The case described here has three developmental defects, namely (1) pronounced ankyloglossia, (2) congenital absence of a primary lower incisor (and its permanent successor), and (3) incomplete fusion of the *symphysis menti*. The common etiology of these problems is speculated to be incomplete embryonic fusion of the left and right first branchial arches that should have occurred during week 5 postconception.

These dental and bony anomalies are readily identifiable in the skeletal record, and we would be interested in hearing about similar cases.

ACKNOWLEDGEMENT

We are grateful to our former resident, Dr. Jonathan Gooch, for bringing this case to our attention.

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