Diprosopus is the encompassing term for full or partial craniofacial duplication, of which there have been only approximately 100 cases reported in the medical literature.1 Previous publications have found that most infants with complete facial duplication exhibit severe central nervous system defects,2 whereas those with partial duplication have normal central nervous system structure and function. It is also evident that several anomalies, including duplication of the tongue, macrostomia, cleft palate, and orbital hypertelorism, have been associated with facial duplication and documented in those patients.2 Barr3 in 1982 divided diprosopus into 4 types, depending on the absence or presence of duplication of the eyes, nose, pituitary gland, maxilla, and mandible.

Duplication of the mouth is a rare subtype of this condition and is often referred to as partial facial duplication. To our knowledge, there are only 7 reported cases so far in the literature for this curious anomaly.4-10

We report a case of a newborn girl with partial facial duplication and a concomitant dysontogenic cyst. Aside from the unique association with a dysontogenic cyst in the ipsilateral floor of the mouth, this case reactivated the question regarding theories of origin.

Report of a Case

Shortly after birth at a primary care center, a 1-day-old girl was found to have an intraoral mass on the right side of the floor of the mouth causing respiratory and feeding difficulties. She was born at term by means of uncomplicated vaginal delivery to a healthy mother whose 3 previous pregnancies had also ended in live births at term (nonconsanguineous marriage). There was no known family history of any congenital anomalies. She was transferred to a tertiary care center and admitted to the neonatal intensive care unit. At this stage, the newborn exhibited cyanotic episodes associated with increased work of breathing, bradycardia, and arterial oxygen desaturations. The pediatric otolaryngology service was then consulted. On initial bedside clinical assessment, it was noted that the cyst was displacing the tongue to the left side, intermittently obstructing the airway. Because of the risk of impending airway obstruction, the patient was taken to the operating room, and the airway was secured with nasotracheal intubation over a flexible bronchoscope under general anesthesia. Thereafter, the patient underwent magnetic resonance imaging, which revealed a large...
unilocular cyst in the right sublingual space, raising the possibility of a ranula, lymphatic malformation, or duplication cyst (Figure 1).

On the newborn’s fifth day of life, the mass was excised using a transoral approach, preserving the sublingual gland and the lingual nerve. The mass was found to be a cystic lesion extending through a gap in the muscles of the floor of the mouth, down to the level of the right submandibular triangle, simulating a plunging ranula. However, subsequent histological examination revealed a congenital epithelial lined cyst with a partly discontinuous squamous, nonkeratinizing, stratified epithelial lining showing focal replacement by granulation tissue, indicative of a dysontogenic cyst, specifically the epithelioid variety (Figure 2).
While engaged with the airway and the cystic lesion, the team also noted the presence of a complete circle of lip tissue located inferior and to the right of the patient’s normal mouth (Figure 3). This accessory mouth exhibited a fully developed and separate vermilion border, and it surrounded a blind pouch lined with mucosa. On palpation, one could feel a tooth bud. A computed tomographic scan was completed at a later date and revealed a right duplicated hemimandible associated with a separate alveolar component and 4 primary teeth (Figure 4).

Additional diagnostic workup was undertaken to investigate whether concomitant congenital anomalies existed. Examination of the temporomandibular joint and muscles of mastication and audiological assessment had normal results. Investigations included abdominal radiography and ultrasound, chest radiography, kidney ultrasound, cranial ultrasound, newborn metabolic screen, and brain magnetic resonance imaging, all of which had normal results. Prenatal and antenatal histories were unremarkable. Neither of the 2 anomalies had been detected on antenatal ultrasound.

The patient underwent excision of the duplicated mouth and repair of the lower lip by the plastic surgery team. She was observed for 1 year and demonstrated normal feeding and attained normal developmental milestones.

Discussion

Facial duplication is a rare anomaly, with an incidence of approximately 1 in 2.5 million births. McLaughlin was the first to describe it in 1948. Facial duplication refers to a spectrum of disease, ranging from partial duplication of isolated facial structures to the complete doubling of all facial structures. In
the developing embryo, the stomodeal plate exists as a depression between the growing brain and the ear. Along with the adjacent branchial apparatus, these develop into the facial structures during the first trimester. The stomodeal structures are most often affected in partial facial duplication. In 1989, Chen and Noordhoff presented 3 cases of duplication of stomodeal structures and reviewed the 18 previously reported cases. In this important work, they proposed a classification system for the duplication of stomodeal structures. This was based on the abnormality present in the true mouth, the developmental completeness of the duplicated lip or jaw, and the location and components of the duplicated tissue:

Type I: duplicated mouth;
Type II: duplication of maxilla–upper lip or mandible–lower lip complex;
Type III: centrally located, poorly developed lip–jaw duplication.

According to this classification scheme, our reported case represents a type I duplication because the duplicated mouth had upper and lower lips, ended in a blind pouch, and contained rudimentary tooth buds. To our knowledge, there have been only 7 reported cases of this type of duplication in the medical literature. Of note, all aforementioned duplications were found on the right aspect of the face except for those of Borçbaken et al., which was located in the left temporal area; Chen and Noordhoff, found on the sphenoid notch; and Morton, which was central in location.

The anomalies most frequently associated with partial facial duplications are cleft palate, lingual duplication, orbital hypertelorism, and macrostomia. Our case is the second known of a partial facial duplication and a concomitant intraoral dysontogenic cyst. Verdi et al reported a comparable case, describing a monozygotic twin exhibiting duplication of the tongue and 2 masses. Both turned out to be dysontogenic cyst-type hamartomas. One was located in the vomer and contained hair, tooth buds, teeth, and squamous epithelium. The second was in the floor-of-mouth mass and contained cartilage, numerous salivary glands, tooth buds, and focal areas of increased vascularity. Several cardiac abnormalities such as ventricular septal defect, double-outlet right ventricle, transposition of the great arteries, and patent foramen ovale have also been identified.

Surgery is considered the mainstay of treatment for this rare condition, its main indication being to achieve a better esthetic outcome. All duplicated mucosa must be removed to prevent development of a retention cyst, which is difficult to accomplish.

The embryogenesis of facial duplication is complex and poorly understood. Barr reviewed the literature of facial duplication prior to 1982 (reporting a 2:1 female preponderance), proposing several suggested mechanisms of embryogenesis. These included splitting of the notochord, duplication of the prosencephalon, duplication of olfactory placodes, and duplication of maxillary and/or mandibular growth centers around the margins of the stomodeal plate. Although these theories offer potential mechanisms of embryogenesis, it must be noted that Barr excluded unilateral accessory mouth from the review. As such, further investigation into the specific embryogenesis of our case is necessary.

Additional theories more specific to duplication of the mouth have been debated. McLaughlin, the first to report duplication of the mandible in 1948, proposed that it was the result of a duplication of the first branchial arch. Later, Bacsich suggested that it was a part of a split notochord syndrome, whereas Davies et al projected that it was a developmental anomaly arising from sequestrated totipotent cells. Verdi et al concluded that facial duplication is less likely to be genetic in origin because only 1 of a pair of monozygotic twins had been born with the duplication, and they attributed it to duplication of the growth centers around the stomodeal plate.

We believe that the theories proposed by McLaughlin and Bacsich are the more likely theories to explain our case. A duplication of the first branchial arch is a feasible proposition because the structures duplicated are all derived from the first branchial arch. Pathological development of the branchial arch may also explain the finding of the dysontogenic cyst because these congenital cysts are thought to be due to sequestration of ectodermal tissue during fusion of the first and second branchial arch. Alternatively, the cyst could be incidental because it is a much more common abnormality than facial duplication. Even this theory is not foolproof, however, because other first branchial arch derivatives were normal in our patient.

Although the split notochord theory is attractive, it would be expected to lead to more major anomalies incompatible with life, explaining the rarity of cases, as well as the more complex craniofacial duplications.

The apparent right-sided preference and female preponderance are yet to be explained. These may be incidental, given the relatively low number of cases in the literature, or may indicate a more complex and multifactorial pathogenesis that is yet to be delineated.

Conclusions

Partial facial duplication is exceedingly rare. There is no agreed-on embryonic basis. Presented here is the second case of partial facial duplication associated with an intraoral dysontogenic cyst. The most plausible theories for embryogenesis include duplication of the first branchial arch.

ARTICLE INFORMATION
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REFERENCES


