Case Report

Fetal Diagnosis and Therapy

Fetal Diagn Ther 2009;25:346–348
DOI: 10.1159/000235883

Received: August 4, 2008
Accepted after revision: December 29, 2008
Published online: September 22, 2009

Prenatal Diagnosis of Oral Cyst Caused by Foregut Duplication

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Key Words
Oral cyst · Foregut duplication cyst · Microphthalmia

Abstract
Prenatal diagnosis of oral cystic lesions is rare but is reported more frequently. The diagnosis of sublingual cyst is important because of the potential for airway obstruction. A rare case of a foregut duplication cyst associated with unilateral sclerocorneal microphthalmia is reported. The differential diagnosis and the limitations of the prenatal ultrasound and the postnatal MRI are discussed.

Case Report

In a routine second trimester ultrasound scan of a 23-year-old gravida 3 para 2 patient of Ethiopian origin with an unremarkable family history, the only unusual finding noticed was a 2 × 2 cm cystic structure in the oral cavity (fig. 1a, b). The primary diagnosis was a sublingual ranula. In follow-up examinations, there was no change in the size, shape or echogenicity of the finding and no secondary manifestations of airway obstruction such as polyhydramnios, enlarged hyperechogenic lungs, or flattened or inverted diaphragm. Based on these findings, it was assumed that such a small cyst would not cause airway obstruction. A female infant was born via spontaneous vaginal delivery, at 40 weeks' gestation with an Apgar score of $9^1/10^5$ and a birth weight of 3,450 g. After delivery, a 3-cm sublingual midline mass was noticed, displacing the tongue upward (fig. 2). Right sclerocorneal microphthalmia was noted in ophthalmologic examination. The neonate was able to bottle-feed but not breast-feed. Magnetic resonance imaging showed a 2 × 2 × 2.3 cm lesion, located beneath the tongue. The suggested diagnosis was a ranula or atypical dermoid cyst. There was no lens in the right orbit, suggesting sclerocorneal microphthalmia (fig. 3a, b). At the age of 11 days, feeding became more difficult due to enlargement of the mass and transoral excision was subsequently performed. The mass was found to be a well-defined, unilocular cystic, thin-walled lesion, 2 cm in diameter containing dark serous fluid. Histological sections showed mucosal lining composed of respiratory, gastric epithelium and focally squamous epithelium consistent with a foregut (enteric) duplication cyst (fig. 4). The postoperative course was unremarkable and the infant was discharged breast-feeding and thriving and was referred for ophthalmologic rehabilitation.

Discussion

Prenatal diagnoses of cystic lesions within the oral cavity are rare but are being reported more frequently during routine second trimester ultrasound scans [1, 2]. The increased detection of these lesions is probably due to the performance of routine coronal sections of the fetal

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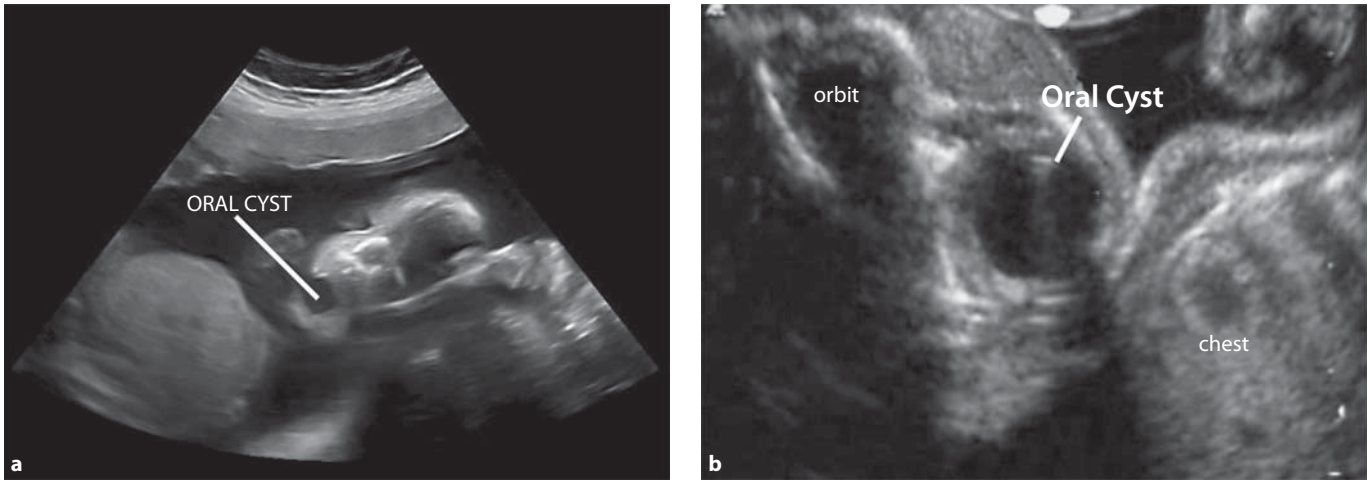

Fig. 1. Coronal sections of the fetal face demonstrating a cystic structure 2 × 2 cm in the oral cavity.

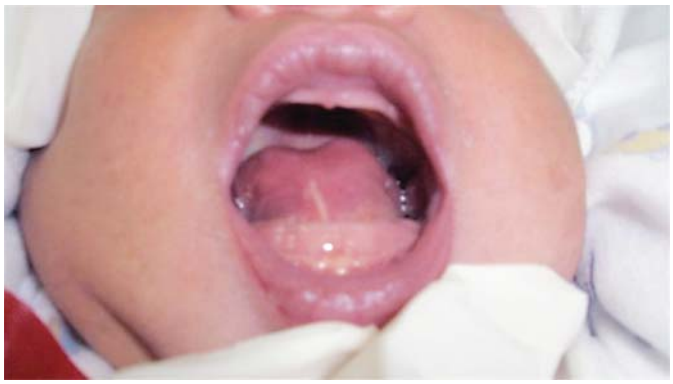

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Fig. 2. Postnatal picture of the oral cavity demonstrating a 3-cm sublingual midline mass, displacing the tongue upward.

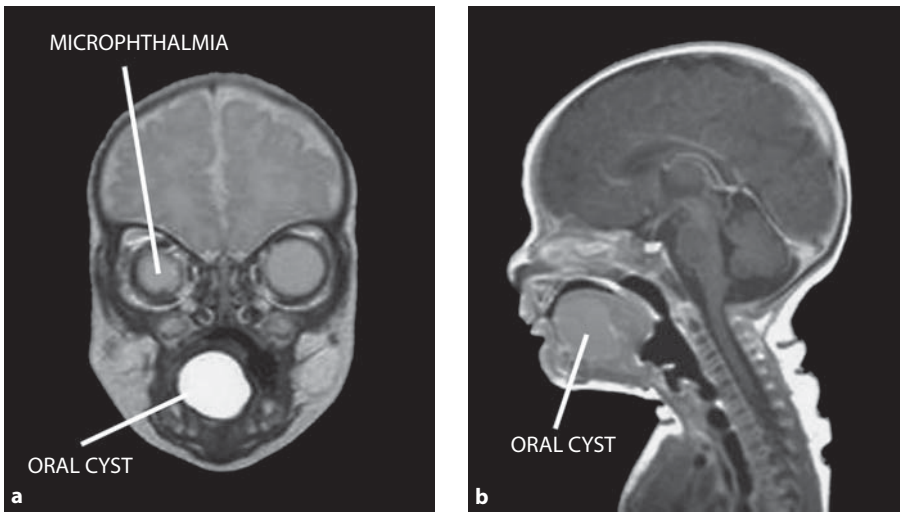

Fig. 3. Postnatal coronal T_2 (**a**) and sagittal T_1W post-Gd (**b**) sections demonstrate the lesion in the floor of the mouth and asymmetry of the orbits and globes.

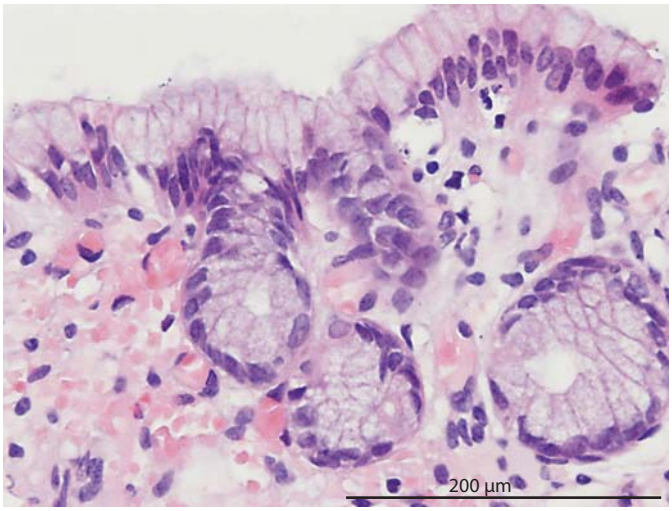

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Fig. 4. Histological section showing mucosal lining of the cystic wall consisting of gastric epithelium.

face in the search of facial dysmorphic features. Foregut duplication cysts in oral cavity are usually isolated findings in otherwise normal fetuses and neonates, but in this patient a right sclerocorneal microphthalmia was missed in the prenatal scans and diagnosed postnatally. Prenatal diagnosis of microphthalmia by MRI was recently reported [3]. The coexistence of unilateral microphthalmia and sublingual foregut duplication cyst in an otherwise normal infant has not been previously described. Insult between the 4th and 8th weeks of gestation can be speculated. Genetic aberrations like mutation in SOX2, which is a key neurodevelopmental gene, was reported in conjunction with microphthalmia and esophageal atresia. In humans, heterozygosity for SOX2 is associated with anophthalmia-esophageal-genital syndrome (OMIM 600992) [4]. Most patients reported had bilateral microphthalmia, but none had foregut duplication. Thus, the combination of the malformations found in our patient may in the future be related to a novel genetic mutation which is currently still unknown. Congenital lesions arising in the floor of the mouth can be difficult to distinguish on antenatal imaging. Foregut duplication cysts in the oral cavity are rare anomalies and few cases with prenatal diagnosis have been reported [2, 5]. These cysts are indistinguishable from dermoids on MRI which is helpful in the preoperative assessment of the location and extent of airway compression, but does not provide a definitive diagnosis [6]. In conclusion, foregut duplication cysts in the floor of the mouth are rare, but should be considered when a cystic mass arises at this site in a fetus. Although isolated in most cases, associated anomalies should be searched for.

Summary Points

- Prenatal diagnosis of sublingual cyst is important because of the potential for airway obstruction.
- Although ranula is the most common diagnosis, other rare sublingual cysts may be considered like foregut duplication cyst.
- Foregut duplication cyst in the oral cavity may be associated with other anomalies like sclerocorneal microphthalmia.

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