SMFM Fetal Anomalies Consult Series #1: Facial Anomalies

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Introduction: Facial Anomalies

Anomalies of the fetal face can be isolated or a component of a complex syndrome. The syndromic involvement of multiple other organ systems can result in adverse outcomes. Evaluation of the fetal face is a basic part of the sonographic fetal survey, and detection of fetal facial anomalies is a key component of prenatal diagnosis. A

FIGURE 1
Coronal view of nostrils, lips, and nose

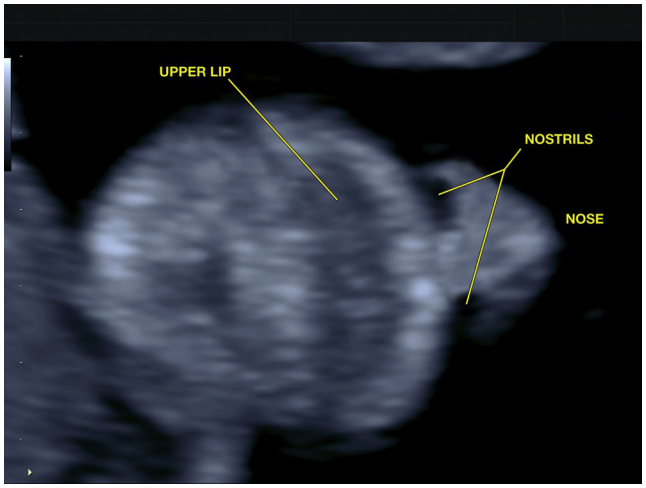

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FIGURE 2
Coronal view of orbits and mandible

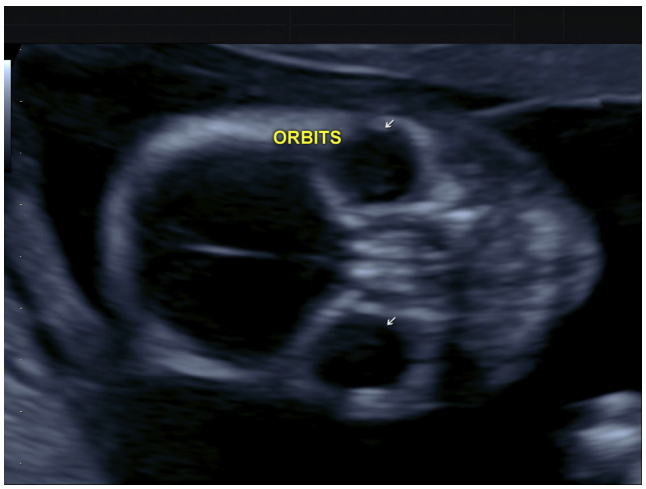

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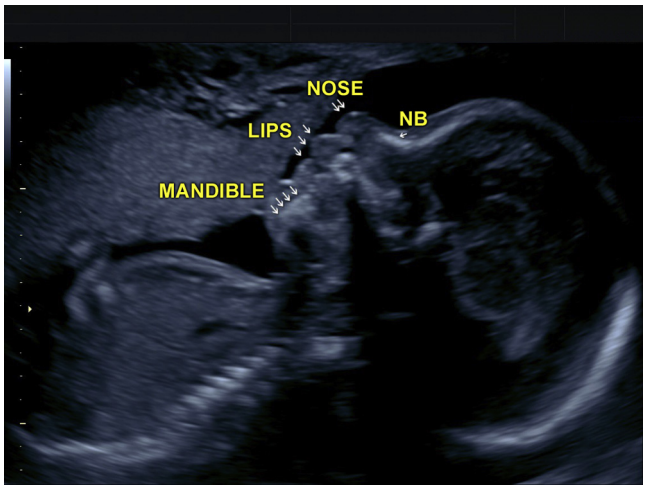
FIGURE 3
Midsagittal view of fetal profile



NB, nasal bone.

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FIGURE 4
Transverse plane and axial view of lenses, orbits, and nasal bones

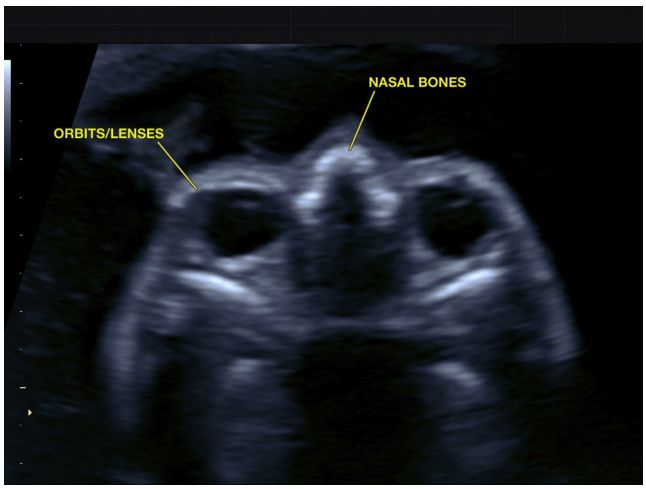

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FIGURE 5
Transverse plane and axial view of tongue

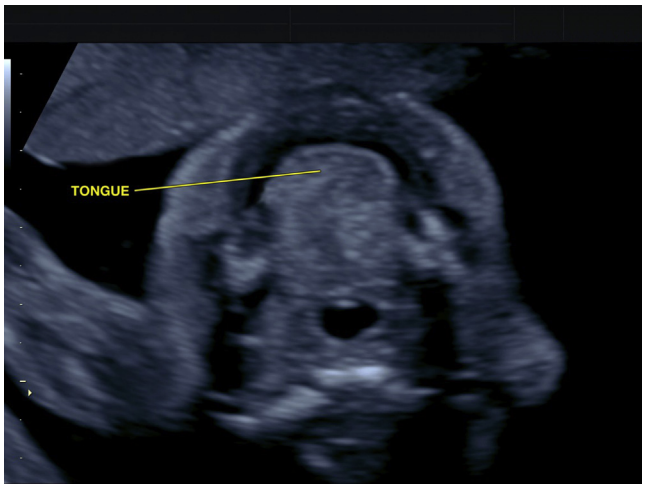

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FIGURE 6
Transverse plane and axial view of alveolar ridge

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standard obstetric ultrasound examination requires only evaluation of the upper lip and is suboptimal for the identification of a range of fetal facial anomalies.[1,2] A detailed obstetric ultrasound examination is more comprehensive and is required if a facial cleft or other facial dysmorphologic condition is suspected on the standard obstetric anatomic scan or if the patient is at risk based on medical or family history. A detailed obstetric ultrasound examination of the face includes the profile, nose, lips, orbits and lenses, palate, maxilla, mandible, and tongue as well as the size and position of the fetal ears, depending on clinical suspicion.[3]

The fetal face should be evaluated in a systematic method with the use of 3 orthogonal planes. This cross-sectional approach to imaging will maximize the detection of facial abnormalities. The coronal view is used to evaluate the integrity of the soft tissue of the fetal lips and appearance of the nostrils (Figure 1). It also allows for evaluation of the fetal lenses and mandible (Figure 2). The midsagittal plane is used to demonstrate the fetal profile, which highlights the appearance of the forehead, presence of the nasal bone, contour of the nose and lips, and position and appearance of the fetal chin (Figure 3). In this plane, one can also evaluate the bones of the maxilla and mandible. The transverse (axial) view can be used to evaluate the fetal orbits and lenses, paired nasal bones (Figure 4), the tongue (Figure 5), and the alveolar ridge that comprises the primary palate (Figure 6). Surface rendering with three-dimensional imaging may be helpful in the demonstration of soft tissue defects. Nomograms are available for various biometric measurements.

If a facial anomaly has been detected and a search for other abnormalities has been completed, the patient must be counseled properly about the ramifications of the findings, further genetic counseling with options for detailed diagnostic testing, prognosis, and further pregnancy management. A multidisciplinary team should be convened to discuss postnatal surgery, genetic evaluation, and alimentary support, if necessary.

This series reviews the sonographic diagnosis, genetic evaluation, and potential treatment and outcome of the following facial abnormalities:

- Absent nasal bone
- Paramedian orofacial cleft
- Micrognathia
- Hypotelorism
- Hypertelorism
- Anophthalmia and microphthalmia
- Median facial cleft

Coding

When coding for fetal facial anomalies (such as fetal cleft lip, fetal cleft palate, micrognathia, microopthalamia, hypotelorism, hypertelorism, or absent nasal bone), the SMFM Coding Committee recommends utilizing the ICD-10 code series O35.8xx0 - O35.8xx9.

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