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Role of antenatal 2D and 3D ultasonography in detection of cleft lip

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Abstract
Ultrasound examination is used for screening of abnormal findings on prenatal screening. Cleft lip with or without cleft palate of the fetus can also be screened by using ultrasonography. Presence of abnormal findings of the fetal lip or palate can be detected accurately and quickly by having a thorough understanding of the embryology of the face. A 30 year-old woman G1P1L0 presented for routine antenatal ultrasound at 19 weeks 4 days according to last menstrual period. Presence of a vertical hypoechoic defect in the upper lip in the paramedian location, not reaching upto the nares is an important sign in the coronal view which aid in the diagnosis. We report a case of unilateral cleft lip without any obvious cleft palate diagnosed antenatally in the second trimester with a brief review of embryology of the face along with critical and practical points in fetal ultrasonographic diagnosis.

Keywords: cleft lip, cleft palate, prenatal ultrasonography, embryology face

1. Introduction
Orofacial clefts are common craniofacial malformations. Cleft lip with or without cleft palate occur more commonly in males, affecting 1:1000 Caucasians, 2:1000 Asians, and 0.3:1000 Africans. However, isolated cleft palate is more common in females with an equal incidence of 0.4:1000 live born in all races [1]. Clefts occur in a ratio of 6:3:1 unilateral left, unilateral right, and bilateral. The cause of orofacial clefts is complex and thought to be multifactorial, representing an interaction between genetics and environment during a critical stage of development [2]. With the increase use of transabdominal 3D ultrasound, frequency of oral clefts being diagnosed antenatally has increased. As oral clefts usually occur in facial areas where the normal embryological fusion of structures did not occur, familiarity of the embryological background will help the ultrasonographer to comprehend and more accurately diagnose these clefts [3].
Cleft lip results from failure of fusion of the frontonasal and maxillary processes, resulting in a cleft of varying extent through the lip, alveolus, and nasal floor. The cleft that does not extend through the nasal floor is termed as an 'incomplete cleft', while a 'complete cleft' imply the lack of connection between the alar base and the medial labial element [4].

3. Materials and Methods
A 30 year-old primi gravida, with non-consanguineous marriage, came to our radiology department for second trimester level II scan. The gestational age of the fetus was 19 weeks 4 days by her last menstrual period. Until then, there were no known problems regarding the mother and the fetus. On ultrasound examination of the face for nose, lips and palate, a vertical hypoechoic defect was seen in the upper lip in the paramedian location on the coronal view. It did not appear to reach upto the nares. (Fig 1) On evaluation of the palate in the mid sagittal and axial views, no gross cleft was seen in the palate in that scan. These findings were suggestive of cleft lip without any obvious cleft palate. On examination by 3D-ultrasound, a similar defect was seen. (Fig 2) Detailed evaluation of the fetus revealed no other associated congenital anomaly.

4. Results and Discussions
At ultrasound, cleft lip is classically appreciated using angled coronal imaging and appearance of a hypoechoic region in the upper lip. This can be combined with similar findings seen in the axial plane or at 3D ultrasound. A cleft of the maxillary alveolus may be more difficult to detect.

During ultrasonography, the fetal tooth buds are highly echogenic once mineralization begins. Therefore, a cleft of the tooth bud–bearing alveolus can be seen as a linear hypoechoic region within the echogenic dental arch. This finding can be useful in cases of unilateral cleft lip. Bilateral complete cleft lip is typically easily visualized because the entire premaxillary segment protrudes from the face and can manifest as an echogenic mass [5]. The degree of proclination, which can be seen in the sagittal view, should be documented in the report because it may assist the surgeon in deciding whether preoperative dentofacial orthopaedic manipulation will be required or not.

Detection of cleft palate at ultrasound can be more difficult. Transverse or coronal imaging of the soft palate may suggest cleft palate. On 2D planar ultrasound images, the uvula is seen as a linear echogenic structure surrounded by parallel anechoic spaces, giving rise to an "equals sign" appearance. Failure to visualize this normal appearance has been termed the "absent equals sign" and may indicate a cleft palate [6]. Use of 3D ultrasound may assist in detection of cleft palate as well as aid in physician understanding and patient and family education. Three-dimensional ultrasound offer the advantages of soft-tissue surface rendering and greater bone detail. Colour Doppler ultrasound can also be used to assess nasal breathing. Flow seen only above the palate suggests that the palate is intact. Abnormalities in fetal swallowing may also suggest the presence of a cleft palate [7].

When a cleft of either the lip or palate is identified at prenatal ultrasound, it is crucial to look for associated anomalies, mainly the midline defects. If associated anomalies are suspected but not seen, fetal magnetic resonance imaging is indicated. Magnetic resonance imaging offers the benefits of revealing associated anomalies and assessing the patency of the airway, both of which may influence delivery plans. One study reported a 4.6% rate of associated cerebral anomalies in patients found to have both cleft lip and palate at antenatal ultrasound.

Unilateral cleft lip and palate can mimic other facial clefts and amniotic band syndromes. However, with good anatomic imaging, other types of clefts will be seen to follow different embryologic lines of fusion, or in the case of amniotic band syndrome will not follow any embryologic pattern. Bilateral cleft lip with a protrusive premaxillary segment can mimic a midline facial mass at ultrasound, raising concern for tumour, encephalocele, or vascular or lymphatic malformation.

Orofacial clefting is multifactorial and has heterogeneous causes. Therefore, proper classification is necessary as different types of clefts may be changeably associated with additional anomalies and chromosomal disorders. An antenatal sonographic classification system has been proposed .This classification shows four types of clefts and their relationship with the primary and secondary palate [8].

Type 1 is isolated cleft lip alone, type 2 is unilateral cleft lip and palate, type 3 is bilateral cleft lip and palate and type 4 is median cleft lip and palate. The so called type 5 cleft is another group of facial clefts associated with the amniotic band syndrome or the limb-body-wall complex and does not follow embryologic patterns but rather shows random types of often large and devastating defects [9].

In our case, a 30 year-old primi gravida, with non-consanguineous marriage, came to our radiology department for second trimester level II scan. The gestational age of the fetus was 19 weeks 4 days by her last menstrual period. Until then, there were no known problems regarding the mother and the fetus. On ultrasound examination of the face for nose, lips and palate, a vertical hypoechoic defect was seen in the upper lip in the paramedian location on the coronal view. It did not appear to reach upto the nares. (Fig 1) On evaluation of the palate in the mid sagittal and axial views, no gross cleft was seen in the palate in that scan. These findings were suggestive of cleft lip without any obvious cleft palate. On examination by 3D-ultrasound, a similar defect was seen. (Fig 2) Detailed evaluation of the fetus revealed no other associated congenital anomaly.

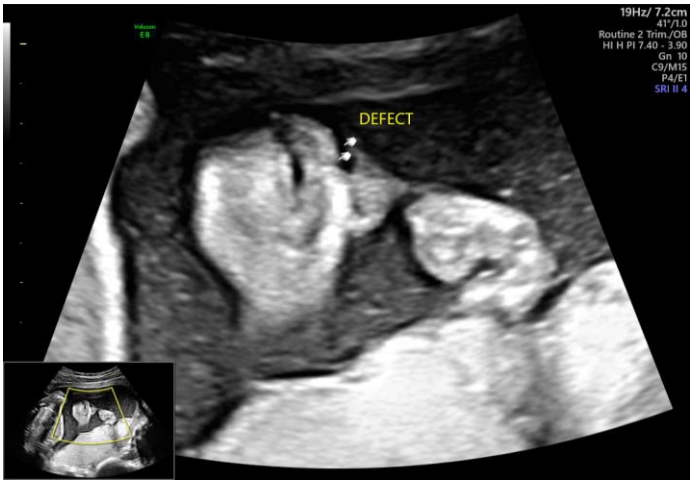

Fig 1: Antenatal 2D Ultrasound Showing Vertical Hypoechoic Defect in Paramedian Location

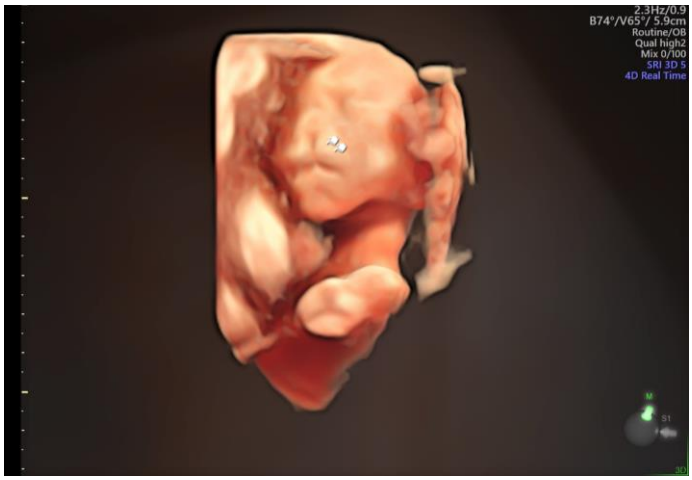

Fig 2: Antenatal 3D ultrasound showing a vertical defect not reaching up to the nares- incomplete cleft lip

5. Conclusion

Imaging of cleft lip, cleft lip with palate and isolated cleft palate necessitates the acquaintance with the clinical entities, treatment plans, and multiple imaging modalities used for diagnosis, treatment planning, and follow-up screening. Making distinction between cleft lip with palate and isolated cleft palate may help in diagnosis of dental and extra facial anomalies. Advances in imaging have made possible the prenatal diagnosis of cleft lip and palate and detection of associated anomalies. Conventional 2D ultrasound can depict cleft lip in multiple imaging planes, while 3D ultrasound can assist in further delineation of the cleft. Knowledge of the embryology of the face adds to the understanding and correctly diagnosing the clefts. Failure of fusion between any of the facial swellings result in facial clefts and can occur either in unilateral or bilateral location, which typically happens at the junction of the lateral incisor and the first premolar teeth. The depth of the cleft may range from the

soft tissue of the lip to a complete cleft of the maxillary bone. Lateralization of the alar base could help the ultrasonographer to diagnose a complete cleft lip with palate. Maxillary protrusion seen with a bilateral cleft lip is highly suggestive of a bilateral complete cleft lip/palate. The embryological fusion of the palatal shelves starts anteriorly and proceeds posteriorly like a zip. An unremarkable uvula, visualised by ultrasound as an "equals sign", suggests an intact normal palate. If the ultrasonographer cannot visualise the middle part of the secondary palate, but the "equals sign" is visible, it is suggestive of an intact palate, which could save the time.

6. References

1. Mulliken JB. The changing faces of children with cleft lip and palate. N Engl J Med 2004;351:745-7.
2. Merritt L. Part 1. Understanding the embryology and genetics of cleft lip and palate. Adv Neonatal Care 2005;5:64-71.
3. Maarse W, Bergé SJ, Pistorius L, van Barneveld T, Kon M, Breugem C *et al*. Diagnostic accuracy of transabdominal ultrasound in detecting prenatal cleft lip and palate: a systematic review. Ultrasound Obstet Gynecol 2010;35:495-502.
4. Tolarová MM, Cervenka J. Classification and birth prevalence of orofacial clefts. Am J Med Genet 1998;75(2):126-37.
5. Nyberg DA, Mahony BS, Kramer D. Paranasal echogenic mass: sonographic sign of bilateral complete cleft lip and palate before 20 menstrual weeks. Radiology 1992;184(3):757-9.
6. Wilhelm L, Borgers H. The "equals sign": a novel marker in the diagnosis of fetal isolated cleft palate. Ultrasound Obstet Gynecol 2010;36(4):439-44.
7. Mittermayer C, Lee A. Three-dimensional ultrasonographic imaging of cleft lip: the winners are the parents. Ultrasound Obstet Gynecol 2003;21(6):628-9.
8. De Milly MN, Alison M, Arthurs O, *et al*. Is fetal cerebral MRI worthwhile in antenatally diagnosed isolated cleft lip with or without palate? Prenat Diagn 2013;33(3):273-78.
9. Nyberg DA, Sickler GK, Hegge FN, Kramer DJ, Kropp RJ. Fetal cleft lip with and without cleft palate: US classification and correlation with outcome. Radiology 1995;195:677-84.