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Sonographic Diagnosis of Fetal Facial Cleft : How Should We Approach?

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ABSTRACT

Objective To establish an appropriate sonographic approach for the diagnosis of fetal facial clefting.

Design Prospective observational study.

Setting Department of Obstetrics and Gynecology, Faculty of Medicine, Chulalongkorn University.

Subjects From September 1996 to September 1997, 94 pregnant women were recruited. Fetal biometry and anatomic survey were performed. Fetal facial scanning was first scanned with conventional coronal plane. The transducer was then rotated to a more axial plane at the level of the upper lip to detect any defect presented. Fetal outcomes were obtained from neonatal results or autopsy findings.

Results A coronal plane demonstrated 8 positive cases of facial clefts. Combining coronal and axial planes revealed only 3 positive cases. Neonatal outcomes revealed 3 facial clefts which were consistent with the ones diagnosed using both coronal and axial planes. The sensitivity and specificity of coronal plane alone in detecting cleft lip were 100% and 94.25% respectively. The sensitivity and specificity using both coronal and axial planes were both 100%.

Conclusion Incorporation of an axial plane to a coronal plane is recommended for establishing the diagnosis of fetal facial clefting.

Key words : facial cleft, ultrasound, fetus

Prenatal evaluation of the mouth, upper lip and tongue provides valuable information regarding cleft lip or macroglossia. Cleft lip is the most common congenital facial deformity detected in newborn accounting for 13 percent of all anomalies.⁽¹⁾ Antenatal diagnosis of a facial cleft may be helpful for counseling of couple so that the birth of the fetus becomes a process of confirmation rather than an unexpected event. Besides, facial cleft can be associated with over hundred syndromes which can lead the physician to a more definite diagnosis of fetal anomalies.⁽²⁻⁶⁾

Sonographic technique in detection of facial cleft has been proposed. The most recommended plane of scanning is a frontal or coronal plane, although a false positive diagnosis based on visualization of a normal philtrum (the midline groove of the upper lip) could be possibly made. Statistics regarding sensitivity and accuracy of antenatal detection of cleft lip are not yet available. This study is aimed to investigate the most appropriate approach to establish the diagnosis of fetal facial clefting sonographically.

Materials and Methods

The study group consisted of 94 pregnant women scanned in our division from September 1996 to September 1997. We recruited only women with singleton pregnancies who had transabdominal scans and who later delivered at our hospital. Fetal biometry and anatomy were scanned in a usual fashion. Fetal facial scanning was performed including a conventional coronal plane to obtain a concomitant fetal nose and upper lip (Figure 1). The transducer was then rotated to a more axial plane at the level of upper lip to detect any defect presented (Figure 1). Any patient whose facial images could not be adequately obtained at the first visit will be rescheduled for another scan until an optimal

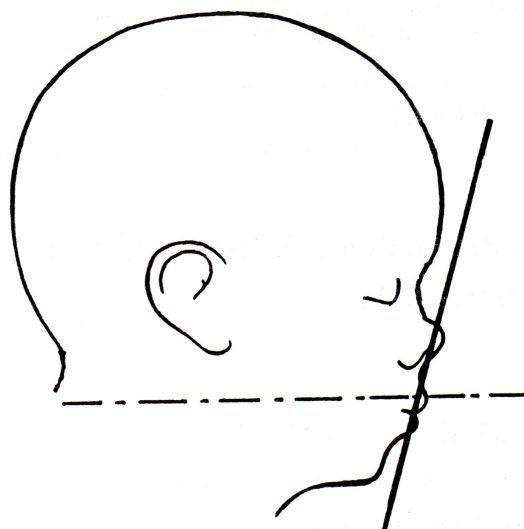


Fig. 1. Drawing showing a conventional coronal scanning plane (dark line) and an axial scanning plane (broken line).

imaging could be obtained.

Scanners included 5 staff members in the Division of Maternal-Fetal Medicine, Department of Obstetrics and Gynecology, Faculty of Medicine Chulalongkorn University. Equipment employed was the Aloka SSD 2000, Tokyo, Japan. with a 3.5 and 5 MHz curvilinear transducer. Fetal outcomes were obtained post partum from neonatal records or autopsy findings. Descriptive statistics and sensitivity as well as specificity were run on all data where it is appropriate.

Results

Four fetuses in whom follow-up could not be obtained were excluded. This left 90 pregnant women in our study group. The gestational age ranged from 9 weeks to 36 weeks. Additional anomalies detected during the study included 3 hydrocephalus, 2 multicystic kidneys, 2 cystic hygroma, 1 cystic adenomatoid malformation of lung, 1 sacrococcygeal teratoma, 1 univentricular heart, 1 porencephalus, 1 hydronephrosis, 1

holoprosencephalus with median cleft and 2 isolated cleft lips.

Due to the fetal awkward positions, the facial coronal plane could not be obtained optimally at the first scan attempt in 6 cases (6.7%). The axial plane on the upper lip could not be adequately viewed in 13 cases (14.4%) at the first attempt. All these cases were successfully demonstrated at the follow-up scan, 2 to 4 weeks later.

Sonographic facial examination in coronal plane revealed 8 positive cases of cleft lips. On employing both coronal and axial planes, only 3 positive cases were diagnosed. Other 5 false positive cases with coronal plane were found to

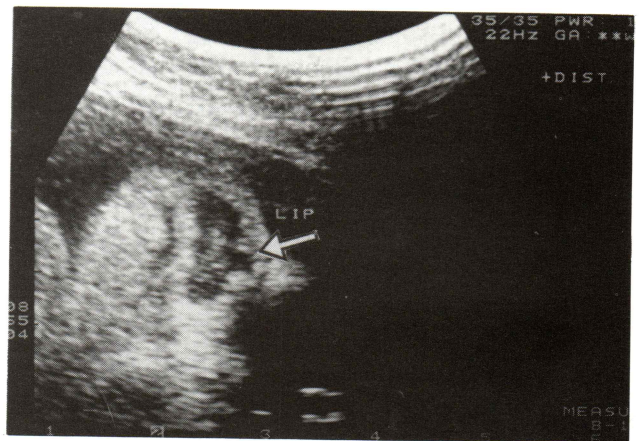


Fig. 2. A coronal plane demonstrates a false positive cleft on the upper lip (arrow) which later demonstrated to have intact upper lip with an addition of axial scan.

Table 1. Sensitivity and specificity of combining coronal and axial scanning in diagnosis of facial cleft

	Facial Cleft	No facial Cleft	
Positive facial cleft on ultrasound	3	0	3
Negative facial cleft on ultrasound	0	87	87
	3	87	90

Sensitivity = $3/3 \times 100 = 100\%$

Specificity = $87/87 \times 100 = 100\%$

Table 2. Sensitivity and specificity of coronal plane scanning in diagnosis of facial cleft

	Facial cleft	No facial cleft	
Positive facial cleft on ultrasound	3	5	8
Negative facial cleft on ultrasound	0	82	82
	3	87	90

Sensitivity = $3/3 \times 100 = 100\%$

Specificity = $82/87 \times 100 = 94.25\%$

have intact upper lip with additional axial plane (Figure 2). Neonatal outcomes demonstrated only 3 cleft lips which corresponded to the ones demonstrated by incorporation of an axial plane to a coronal plane. The results showed that the sensitivity and specificity of sonographic diagnosis of facial cleft using combining coronal and axial planes were both 100% whereas the sensitivity and specificity using coronal planes alone were 100 % and 94.25 % respectively. The details of these results were shown in Table 1 and 2.

Discussion

Facial cleft results from failure of fusion of the frontal prominence with the maxillary process during embryogenesis. It may be complete or incomplete, unilateral or bilateral, symmetrical or asymmetrical and approximately two third of fetuses with cleft lip also have cleft palate.⁽⁷⁾ Antenatal detection of facial cleft is important. It is not only essential for parental counseling but also an important guide for a detailed structural survey as it is associated with other structural abnormalities and syndromes.⁽⁴⁻⁶⁾ Craniofacial malformations may be one of features of a number of syndromes with multisystem involvement and their recognition may have clinical relevance. A typical example could be represented by the holoprosencephalic malformation in which anomalies of the brain and face are directly correlated.⁽⁸⁾

Sonographic diagnosis of cleft lip depends primarily on the demonstration of a groove extending from one of the nostril inside the lip in case of lateral cleft and a wide central defect involving the upper lip in case of median cleft.^(8,9) This needs an appreciation of facial topography in multiple planes as well as adequacy of experience in the technique. Most investigators recommended a coronal plane to detect the lesion as it is simple and easy to obtain. Nevertheless,

concern over false positive diagnosis of a normal philtrum of the upper lip could be possibly made if the coronal plane is employed alone. Our study demonstrated a sensitivity and specificity of 100% and 94.25% respectively on employing the coronal plane alone for diagnosis of facial cleft. On the other hand, incorporation of an axial plane would render a 100% of both sensitivity and specificity. In this study the number of fetuses with structural abnormalities is relatively high. This was because the majority of pregnant patients recruited were referral high risk cases from across the country.

It is possible in the very near future that 3-dimensional ultrasound will be the best solution for this diagnostic problem as the appreciation of the facial feature could be achieved with a single image.⁽¹⁰⁾ Nevertheless, economical concern still has to be taken into account in employing this technology for the routine clinical practice. This study indicates that employing an axial plane in conjunction with a coronal plane is more accurate than using coronal plane alone in antenatal detection of facial cleft and should be a more appropriate approach for establishing the diagnosis of facial cleft. However, the sample size of this study was small, further study with a larger population should be conducted to confirm our results.

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