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Assessment of the gap between the fetal nasal bones at 11 to 13 + 6 weeks of gestation by three-dimensional ultrasound

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KEYWORDS: first trimester; nasal bone; screening; three-dimensional ultrasound; trisomy 21

ABSTRACT

Objective *To detect the presence of a gap between the fetal nasal bones at 11 to 13 + 6 weeks of gestation and to verify if this gap could lead to the erroneous diagnosis of absent nasal bone.*

Methods *Three-dimensional (3D) ultrasound was used to assess the fetal nose in 450 singleton pregnancies, immediately after two-dimensional (2D) evaluation of the nasal bones and screening for chromosomal defects by a combination of maternal age and the measurement of fetal nuchal translucency at 11 to 13 + 6 (median, 12) weeks of gestation. A 3D volume of the fetal face was acquired and then analyzed using the multiplanar mode. A sequence of transverse views was used to confirm the presence or absence of the nasal bones and when they were present any visible gap between them was measured. A perfectly mid-sagittal plane was then examined to determine if the nasal bone was visible or not.*

Results *In 421/450 (93.6%) cases the nasal bone was present during 2D ultrasound. Using the multiplanar mode of 3D ultrasound, in 83/421 (19.7%) fetuses a gap between the nasal bones could be demonstrated and in 36/83 (43.4%) cases the nasal bone was found to be absent in the perfect mid-sagittal view. In 29/450 (6.4%) cases the nasal bones were absent during the 2D scan. In the 3D assessment there was absence of both bones in 25/29 (86.2%) cases and absence of one of the two bones in 4/29 (13.8%) cases. Chorionic villus sampling demonstrated that the fetal karyotype was normal in 404 and abnormal in 46 cases, including 31 cases of trisomy 21. There was absence of one or both nasal bones in three (0.7%) of the chromosomally normal fetuses, in 19 (61.3%) with trisomy 21 and in seven (46.7%) with other chromosomal defects.*

Conclusions *At 11 to 13 + 6 weeks of gestation there is a gap between the nasal bones in about 20% of fetuses, and in about 40% of these cases in the perfect mid-sagittal plane the nasal bone may erroneously be considered to be absent. Copyright © 2005 ISUOG. Published by John Wiley & Sons, Ltd.*

INTRODUCTION

There is a high association between absent fetal nasal bone at the 11 to 13 + 6 gestational weeks scan and trisomy 21[1,2]. It was estimated that an integrated sonographic and biochemical test at 11 to 13 + 6 weeks, which combines examination of the nasal bones and measurements of the fetal nuchal translucency (NT) and maternal serum free β-human chorionic gonadotropin and pregnancy-associated plasma protein-A, can identify more than 95% of trisomy 21 fetuses for a false-positive rate of less than 5%[3].

Evaluation of the nasal bone requires well-trained operators to obtain a mid-sagittal view of the fetal profile[4,5]. However, the bony part of the nose is actually composed of two bones, which meet in the midline, and at 11 to 13 + 6 weeks the suture may not be completely closed. It is therefore theoretically possible that in a truly mid-sagittal view these bones may not be visible and a false-positive diagnosis of absent nasal bone made.

The aim of this study was to evaluate the fetal nose with three-dimensional (3D) ultrasound to detect the presence of a gap between the two nasal bones, and to verify if a sagittal plane passing through this gap could lead to the erroneous diagnosis of absent nasal bone.

METHODS

A 3D volume of the fetal face was acquired before fetal karyotyping by chorionic villus sampling (CVS) at 11

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to 13 + 6 weeks of gestation in singleton pregnancies that had been evaluated for the risk of trisomy 21 by a combination of maternal age and fetal NT[6]. In 450 cases the 3D volume was obtained with the fetus in the mid-sagittal plane and the transducer being parallel to the nose. In conventional two-dimensional (2D) ultrasound, which was carried out before the 3D evaluation, the nasal bone had been classified as present (thicker and more echogenic than the overlying skin) in 421 cases and as absent (complete absence or less echogenic than the overlying skin) in 29 cases. All 3D examinations were carried out transabdominally (RAB 4–8L probe, Voluson 730 Expert, GE Medical Systems, Milwaukee, WI, USA), by sonographers with extensive experience in 3D ultrasound, and who were blind to the results of the 2D evaluation.

For the analysis of the nasal bones, the volume was displayed in the three orthogonal planes that compose the multiplanar mode of the 3D image. The sagittal view showing the fetal profile in this mode was then selected and, as this plane was scrolled, the series of corresponding transverse and coronal images of the fetal face was simultaneously demonstrated. The sequence of transverse views was used to confirm the presence or absence of the nasal bones and, when these were present, a search was conducted for a possible gap between them. If a gap could be seen, its wider length was measured in the transverse plane. This transverse image was then scrolled sideways so that a corresponding sagittal view passing through the gap could be displayed. The purpose of this later evaluation was to determine whether an image of absent nasal bone could be demonstrated when a gap was present.

RESULTS

The ethnic origin of the mothers was Caucasian in 394 cases, Afro-Caribbean in 25, Asian in 20 and Oriental in 11. The median maternal age was 37 (range, 17–45) years, the median crown–rump length (CRL) was 67.1 (range, 45–84) mm, the median gestational age was 12 (range, 11.0–13.9) weeks and the median delta NT (difference between the observed NT and the normal median NT for the same CRL)[7] was 0.8 (range, −0.7 to 18.2) mm.

In 421/450 (93.6%) cases the nasal bone was classified as being present during 2D ultrasound. In 83/421 (19.7%) fetuses a gap between the nasal bones could be demonstrated in the transverse plane of the 3D volume (Figure 1) and in 36/83 (43.4%) cases the nasal bone was found to be absent in the perfect mid-sagittal view obtained in the multiplanar mode. In the 83 cases with a gap, absence of the nasal bone could be demonstrated in all of the 31 fetuses with a gap of 0.7 mm or more, in 5/16 with a gap of 0.6 mm and in 0/36 with a gap of less than 0.6 mm (Figure 2).

In 29/450 (6.4%) cases, the nasal bones were considered to be absent during the 2D scan. In the 3D assessment there was absence of both bones in 25/29 (86.2%) cases and absence of one of the two bones in 4/29 (13.8%) cases (Figure 1).

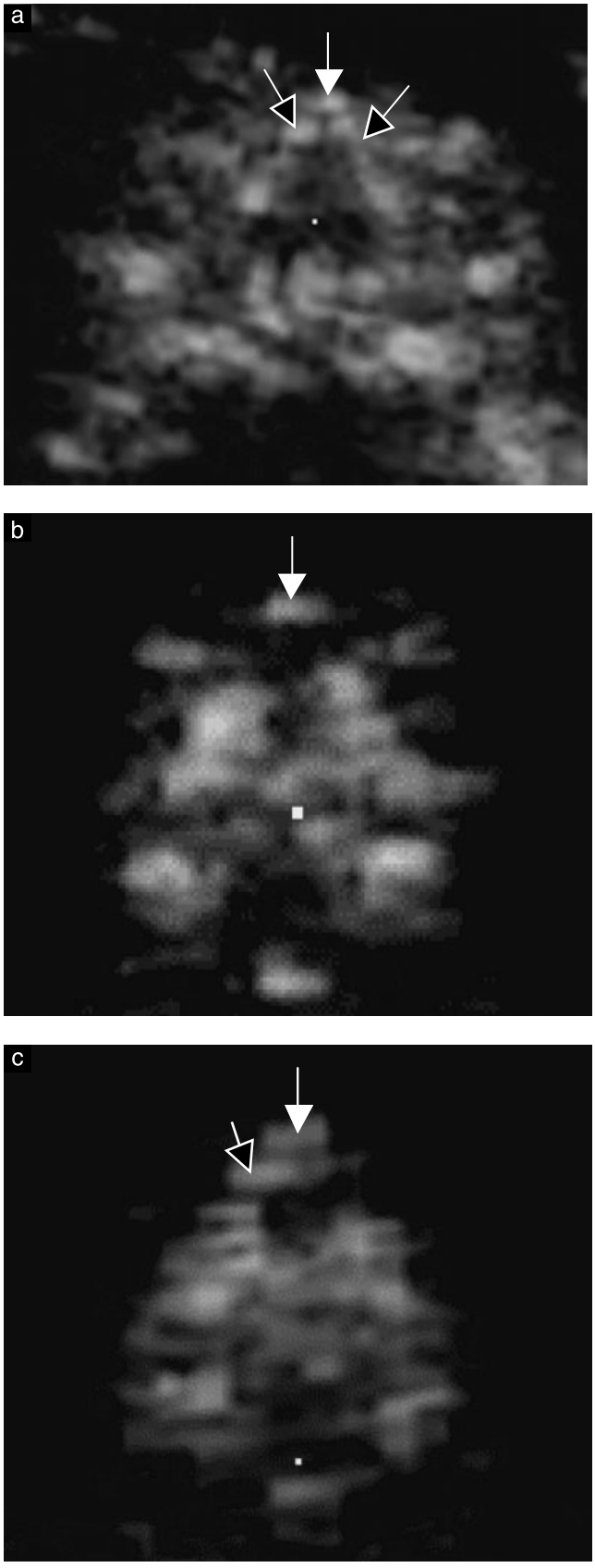

Figure 1 Transverse views of the fetal nasal bones in the multiplanar mode of three-dimensional ultrasound. In (a) there is a small gap between the nasal bones, in (b) there is absence of both nasal bones and in (c) there is absence of one of the nasal bones. White arrows, skin; black arrows, nasal bone.

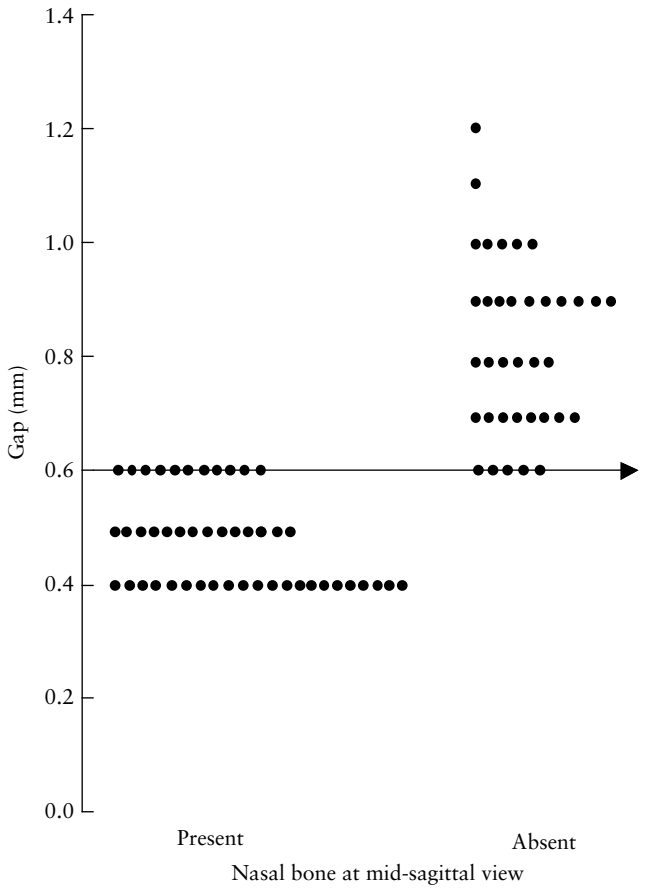

Figure 2 Measurement of the gap between the nasal bones in the transverse view ($n = 83$) according to the presence or absence of the nasal bone in the mid-sagittal plane through the gap.

CVS demonstrated that the fetal karyotype was normal in 404 cases and abnormal in 46, including 31 cases of trisomy 21, seven of trisomy 18, five of triploidy, two of Turner's syndrome and one of Klinefelter syndrome. There was absence of one or both nasal bones in three (0.7%) of the chromosomally normal fetuses, in 19 (61.3%) of those with trisomy 21 and in seven (46.7%) of those with other chromosomal defects (Table 1). In those with present nasal bones there was a gap between the bones in 77/401 (19.2%) chromosomally normal fetuses (median size, 0.6 and range, 0.4–1.2, mm), in 2/12 (16.7%) with trisomy 21 (size, 0.5 and 0.7 mm) and in 4/8 (50%) with other chromosomal defects (size, 0.4, 0.7, 0.8 and 0.8 mm).

Table 1 Prevalence of bilateral and unilateral absence of the nasal bones in chromosomally normal fetuses, trisomy 21 fetuses and those with other chromosomal defects

		Nasal bones (n *(%))*		
Karyotype	n	*Both absent*	*One absent*	*Both present*
Normal	404	2 (0.5)	1 (0.2)	401 (99.3)
Trisomy 21	31	16 (51.6)	3 (9.7)	12 (38.7)
Other defect	15	7 (46.7)	—	8 (53.3)
Total	450	25 (5.6)	4 (0.9)	421 (93.6)

DISCUSSION

This study has demonstrated that in some fetuses at 11 to 13 + 6 weeks of gestation there is a gap between the two developing nasal bones. Furthermore, with the use of 3D ultrasound to obtain a perfectly mid-sagittal plane of the fetal face it is possible to demonstrate 'absence' of the nasal bones when the gap between them is 0.6 mm or more. The fact that in all cases with a gap smaller than 0.6 mm the nasal bone was seen in the perfect mid-sagittal view might be explained by the limit of the lateral resolution of the ultrasound equipment.

None of the cases with a gap was associated with a diagnosis of absent nasal bone in the 2D scan, and in the 29 cases where the nasal bones were classified as absent during 2D ultrasound at least one of the two bones was also absent in the 3D evaluation. These findings suggest that it is extremely unlikely that a false-positive diagnosis of absent nasal bone is made in the presence of a gap. Given the very small dimension of the gaps observed in this study, it is likely that any slight lateral movement of the transducer during the 2D evaluation of the fetal profile would allow the identification of the nasal bones. Indeed, in the original description of the technique for 2D examination of the nasal bone we suggested that once the mid-sagittal view of the fetus is obtained, the ultrasound transducer should be gently tilted from side to side to ensure that the nasal bone is adequately examined[1].

The findings, of the 2D examination and subsequent confirmation by 3D, that there was absence of the nasal bone in 61.3% of the trisomy 21 and 0.7% of the chromosomally normal fetuses are compatible with the results of previous reports. An anthropometric study in 105 patients with trisomy 21 at 7 months to 36 years of age reported that the nasal root depth was abnormally short in 49.5% of cases[8]. Postmortem radiological studies in aborted fetuses with trisomy 21 reported absence or hypoplasia of the nasal bone in about 55% of cases, sonographic studies at 15–24 weeks of gestation reported absent or short nasal bone in about 65% of trisomy 21 fetuses, and studies at 11 to 13 + 6 gestational weeks in a total of 15 822 pregnancies reported absence of the nasal bone in 69% of 397 fetuses with trisomy 21, compared to 1.4% of normal fetuses[9].

We found unilateral absence of the nasal bone in 1/404 (0.2%) chromosomally normal fetus and in 3/31 fetuses with trisomy 21. A postmortem radiological and histological study of 33 aborted fetuses with trisomy 21 at 14–25 gestational weeks demonstrated bilateral and unilateral absence of the nasal bones in eight and two cases, respectively[10]. Goncalves *et al.* examined by 3D ultrasound 26 fetuses at a median gestation of 21 (interquartile range, 19–25) weeks and reported bilateral and unilateral absence or hypoplasia of the nasal bones in 19 and one cases, respectively[11]. Similarly, Benoit and Chaoui examined by 3D ultrasound 20 fetuses at 17–33 gestational weeks and reported bilateral and unilateral absence of the nasal bones in six and three cases, respectively[12].

The present study did not aim to compare 2D with 3D imaging of the fetal nasal bone in their effectiveness in screening for trisomy 21. However, there are two essential findings in the present study that could potentially influence screening. First, the presence of a gap between the nasal bones in about 20% of normal fetuses could result in the erroneous diagnosis of 'absent' nasal bone, thereby increasing the false-positive rate. Second, the presence of only one nasal bone in about 10% of trisomy 21 fetuses could result in the erroneous diagnosis of 'present' nasal bone, thereby decreasing the detection rate. In this respect, it could be argued that 3D ultrasound is essential in effective screening using the nasal bone. However, we have demonstrated that with 2D ultrasound none of the cases with a gap between the nasal bones was classified as 'absent' nasal bone, and none of the cases with unilateral absence was classified as 'present' nasal bone.

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