Prenatal ultrasound screening for orofacial clefts

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OBJECTIVES
To evaluate the sensitivity and specificity of ultrasound for prenatal diagnosis of facial clefts in a low-risk population and compare it with recent studies.

METHODS
Retrospective review of patients who underwent routine second-trimester prenatal ultrasound screening at a tertiary referral hospital (~5500 deliveries per year), including those referred to the Fetal Medicine Unit, during a 2-year period from January 2012 to December 2013. Patients were identified and antenatal notes reviewed retrospectively.

RESULTS
A total of 9123 low-risk women underwent ultrasound screening. The incidence of orofacial clefts was 1.38% (24 cases). The distribution of clefts was as follows: 4 (17%) cleft lip (CL), 15 (62.5%) cleft lip and palate (CLP), and 5 (21%) cleft palate (CP).

One of the cases with cleft palate had Pierre Robin sequence and 3 of the cases with CL + P had other structural anomalies (cardiac, renal, single umbilical artery), all of them detected antenatally. None of the infants had any syndromic or dysmorphic features. Antenatally 10 (42%) identified as CL, 8 (33%) as CLP, 1 (4%) as micrognathia and 5 (21%) as normal.

Routine amniocentesis was offered in all cases of suspected CL + P and/or with associated structural abnormalities and was performed in one case (result was normal).

CONCLUSIONS
Review of the literature reporting on the accuracy of 2D ultrasound in detecting CL ± P in low risk populations demonstrates a wide variety in diagnostic accuracy.

Most studies report detection rates between 9% and 75%, indicating a considerable number of missed diagnoses of CL ± P. Moreover, the overall reported rate of false-positive diagnosis for prenatal CL ± P in routine screening is low. All of them agree that the detection of isolated cleft palate is difficult.

In our unit the sensitivity of detection for CL ± P was 75%. The percentage of missed diagnoses reached the 21% and no case of isolated CP was detected antenatally.

Overall we can conclude that in our unit the ultrasound screening for CL ± P in low risk population enrols with the general standards set by different recent studies.