

Bilateral cleft lip and palate associated with increased nuchal translucency and maternal cocaine abuse at 14 weeks of gestation

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Summary

A case of bilateral cleft lip and palate associated with increased fetal nuchal translucency detected at 14 weeks of gestation in a cocaine abusing pregnant woman is presented. There were no other associated structural or chromosomal abnormalities. We propose that systematic examination in both the sagittal and parasagittal plane of the fetal profile and recognition of the characteristic ultrasound appearance of a premaxillary protruding echogenic mass should make detection of this type of cleft relatively easy at the moment of the first trimester scan. First trimester diagnosis of cleft lip and palate can facilitate the parental decision-making process on continuing or terminating the pregnancy and should open the perspective of fetal surgery.

Key words: Orofacial cleft; Nuchal translucency; Cocaine abuse.

Introduction

Bilateral cleft lip and palate results from failure of the fusion of the primary with the secondary palate. Two types of bilateral cleft lip are recognized on prenatal ultrasound – with or without premaxillary protrusion [1]. Premaxillary protrusion represents abnormal alveolar and gingival tissue resulting from uninhibited growth of the premaxilla caused by lack of continuity between bony, gingival, and labial structures.

Case Report

A 21-year-old woman, G2P1A0 presented at 14+0 weeks of gestation for a routine first trimester scan. Her medical history was notable for daily intranasal cocaine use. Ultrasound examination revealed a viable fetus with biometry consistent with the gestational age and increased nuchal translucency thickness for the gestational age (3.3 mm). An echogenic premaxillary mass protruding over the fetal nose was demonstrated in the mid-sagittal plane of the fetal profile (Figure 1). On axial views of the fetal face the hard palate was found to be severely affected (Figure 2a) and the cleft was also easily identified (Figure 2b). The rest of the fetal anatomy survey was unremarkable. Amniocentesis was performed and revealed a normal fetal karyotype 46, XY. After extensive counseling with the local maxillofacial-team, the parents opted for termination of pregnancy which was performed at 17 weeks' gestation. Pathological examination confirmed the prenatal diagnosis (Figure 3).

Discussion

It has been suggested that cocaine exposure could result in an increased incidence of facial clefts through a mechanism involving vasoconstriction, but the actual

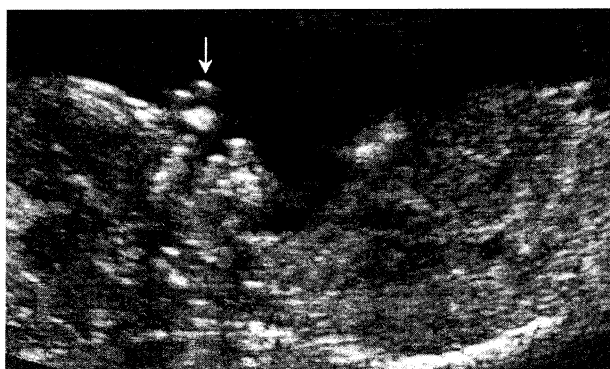


Figure 1. — Mid-sagittal view of the fetal profile at 14+0 weeks of gestation. Note the premaxillary protrusion (arrow).

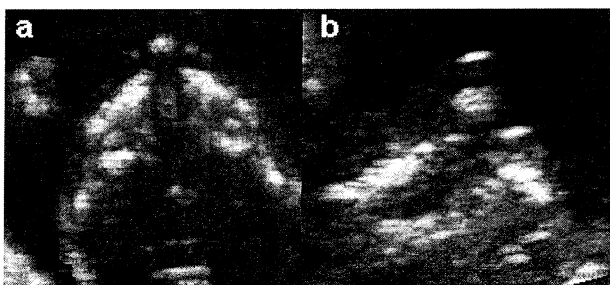


Figure 2. — Axial views (a, b) of the fetal face at 14+0 weeks of gestation. Note the lack of continuity of the hard palate (a) and the presence of the cleft itself (b).

association between cocaine exposure and facial clefts is still unresolved [2]. The presence of an echogenic mass protruding over the fetal nose is recognized as a typical ultrasound finding for bilateral cleft and palate with premaxillary protrusion in the second and third trimester of pregnancy [3]. Since the ultrasonographic appearance is



Figure 3. — Pathological examination at 17+1 weeks of gestation.

characteristic the diagnosis should be assumed even if the cleft is difficult or impossible to visualize on axial and coronal views. Although the cleft originates at around seven weeks of gestation, the premaxillary protrusion develops after 11 weeks of gestation only.

Contrary to the cleft itself, a premaxillary protrusion could be more easily detected at the moment one is looking for nuchal translucency and nasal bone ossification, which could enhance early pregnancy detection rates of facial clefts [4]. Only a few reports have been published concerning the first trimester prenatal diagno-

sis of cleft lip/palate and it is clear that the earlier diagnosis influences the decision-making process regarding the choice between continuing the pregnancy and preparing dedicated neonatal care versus termination of pregnancy [5]. Early detection of orofacial clefts could allow for fetal surgery to be performed before the transition from fetal (scarless) to adult-like healing [4].

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