

Prenatal and postnatal diagnosis of cleft lip and palate

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Orofacial clefts are one of the most common congenital malformations. The prevalence of cleft lip and palate (CLP) has been reported to be 0.48/1000 (Baumler M et al, 2011). The genetic risk of malformations is more elevated when the clefts of palate and lip are associated than with cleft lip is alone (BergÈ SJ et al, 2001).

Prenatal ultrasound techniques are used to display both the normal and the pathological fetal lip and palate (Platt L et al, 2006). It's very important to verify the accuracy of ultrasound technique in predicting clefting of the fetal lip and hard palate and other congenital anomalies. In the current study we present a case report of a woman with prenatal diagnosis of cleft lip and palate: we compared the prenatal ultrasound predictions with postnatal clinical findings on examination of the newborn's palate.

A 33-year-old woman, with a family history of cleft lip and palate, was referred for routine ultrasonic examination during her second trimester of pregnancy. Ultrasonography (USG) showed a single live foetus of a gestational age of 20+4 weeks; biparietal diameter and head circumference were adequate for the week of gestation. In frontal view it was possible to identify a bilateral cleft of the upper lip. In median sagittal view was depicted also a cleft of hard palate. After genetic counseling, the patient decided for an elective termination of the pregnancy. A stillborn female fetus was delivered with a weight of 350 g. Infantogram and gross autopsy confirmed the lip and palate cleft, but the autopsy revealed also an hidden form of spina bifida.

A detailed scan of fetal anatomy between 20 and 32 gestational weeks detect majority of cleft lip and palate, but a few of congenital abnormalities are still difficult to diagnose antenatally. It's therefore necessary to improve the accuracy of ultrasonic examination in order to provide the correct informations for a possible termination of pregnancy. However, this last must be preceded by genetic counseling and diagnosis for recurrence risk.

References

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Key words

Cleft lip, palate lip, ultrasonography, fetal morphology.