



## Early prenatal diagnosis of cleft lip and its potential impact on the number of babies with cleft lip

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### INTRODUCTION

Isolated cleft lip is generally considered to be a cosmetic minor malformation which should not influence the continuation of an otherwise normal pregnancy.

However, in our previous experience<sup>1–3</sup> most parents of an affected fetus are inclined to terminate the pregnancy if the malformation is detected early in the pregnancy before the development of fetomaternal bonding.

### Background

Despite decades of intensive investigation into the cause of one of the most common congenital malformations, cleft lip and palate, the pathogenesis is still not clear,<sup>2,4</sup> and there seems to be both genetic and environmental components in its aetiology.<sup>2</sup> The incidence varies between 1 per 1000 live births in whites and 1.7% among Japanese.<sup>2,4</sup> In 75–80% of cases the cleft lip is unilateral, more common on the left, and in two thirds of cases it is associated with a cleft palate.<sup>2,4</sup> It is twice as common in male fetuses.<sup>2</sup> The genetic component seems to involve an autosomal recessive major gene with reduced penetration.<sup>2,4</sup> By the fourteenth gestational week the facial contour of a developing fetus is virtually complete,<sup>2</sup> so detection of the anomaly should be attempted as early as the late first and early second trimester. Cleft lip and palate may be associated with other fetal malformations in 7–13% of cases, in some as part of one of many syndromes.<sup>2,4</sup>

### Prenatal diagnosis

Although cleft lip is detectable by transvaginal sonography,<sup>1–3</sup> in most cases it is undetected as most isolated anomalies affect patients with no previous history of fetal malformations. Sonographic assessment of the mouth and lips of the fetus is not usually included as a standard part of an anatomical sonographic scan for detection of fetal malformations,<sup>1</sup> and transabdominal sonographic examinations, which are usually done after 18 weeks' gestation, have not been successful in detecting cleft lip and palate.<sup>1,6,7</sup> Transvaginal sonography has gained worldwide popularity in the last decade because it

bypasses the subcutaneous tissue that separates the transducer and the fetus on transabdominal ultrasound.<sup>1,8</sup> Its resolution is superior to that of the transabdominal ultrasound because of the higher frequency (6.5–7.5 MHz for transvaginal compared with 3.5–5 MHz for transabdominal).<sup>1,8</sup> We started using the transvaginal approach in 1987 for the early prenatal diagnosis of fetal anomalies.<sup>8</sup> Because of its high resolution and proximity to the fetus it successfully detects 95% of the fetal malformations at 14 to 16 weeks' gestation.<sup>8</sup> Cleft lip has been identified as early as the eleventh week of gestation.

In northern Israel most gravid patients routinely undergo transvaginal sonography at 14–16 weeks' gestation,<sup>8</sup> and during the last 10 years we have done over 24 000 scans and detected 15 cases of cleft lip; in 14 cases the pregnancy was terminated. Thirteen of these 15 cases had cleft lip (with or without cleft palate) when examined postabortally. In two cases the pregnancy was terminated by dilatation and curettage (D&C) which made the postabortal examination impossible. We had only one false negative scan, in which the newborn had a small (<1 mm width) isolated cleft lip without cleft palate or any other associated anomalies. We are not aware of any false positive scans. Most of these cases were in low risk patients, with no previous medical history of fetal anomalies, consanguinity, exposure to drugs, irradiation or any known teratogens.<sup>8</sup>

Fourteen of the 15 parents voluntarily chose termination of pregnancy although they had been told that it is difficult to calculate the size of the cleft lip exactly. The decision was made after the couples had consulted other parents of children who had been born with cleft lip and had undergone surgical plastic repair.<sup>1</sup> In a small survey among 17 families who had a child with congenital cleft lip, all the parents stated that they would choose termination in a subsequent gestation if cleft lip was detected in the early second trimester. Eight plastic surgeons and 15 maxillofacial surgeons have also claimed they would have preferred termination to delivery of an affected newborn.

A demonstrative case report concerns the parents of a child who was born with a cleft lip and had undergone successful plastic reconstruction with favourable results. They came for transvaginal screening for the detection of possible fetal anomalies at 15 weeks' gestation. They declared that they are grateful

for their previous child and were happy that they had not come for sonographic screening in their previous pregnancy as the cleft lip might have been detected and they would probably have decided on termination. When asked what they would do if a cleft lip was detected in the current gestation they said they would choose termination. The only couple who continued the pregnancy of the 15 cases in which cleft lip had been detected prenatally said that they would decide on termination in a subsequent gestation in which the fetus was affected by a cleft lip.

## CONCLUSIONS

Cleft lip can be accurately detected or ruled out by transvaginal sonography<sup>1-3</sup> at 13-16 weeks' gestation. Cleft palate cannot be always detected or ruled out prenatally. The exact severity of the cleft lip and the size of the fissure is difficult to assess accurately. The cleft lip can be estimated only roughly. This malformation is considered to be severe among those who have had previous personal experience—parents of an affected child, the patients themselves, their closest family, and physicians who look after such patients. Most of them would decide on termination in a subsequent gestation with a similarly affected fetus if the diagnosis was made in the early second trimester before fetomaternal bonding has taken place.

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