

## Prenatal diagnosis of facial clefts and associated anomalies

Sdiri M, Abid C, Lamiri N, Karoui A, Magdoud K, Abouda HS, Zargouni O, Chelly D, Chanoufi M  
Service C, The National Center of Maternity and Neonatology of Tunis., Tunis, Tunisia

### Objective

The aim of this study was to determine the relationship between facial clefts, associated malformations and chromosomal abnormalities.

### Methods

The study was conducted between January 2008 and December 2018. Forty Fetuses with a suspected cleft lip with or without cleft palate, determined by prenatal ultrasound, were prospectively and retrospectively enrolled and evaluated for the nature of the cleft lip or palate and for the nature of the associated anomalies. Additionally, karyotype was performed in 35 of the 40 patients (87.5%). Postnatal outcome was obtained.

### Results

Postnatal follow-up revealed that 29 (72.5%) of these 40 fetuses had an additional structural or syndromic abnormality. None of the fetuses presenting an isolated cleft lip had additional anomalies and all survived. Of the 11 fetuses with prenatally determined 'isolated' cleft lip with or without cleft palate, 3 (27.27%) had an additional malformation identified after delivery. The frequency of additional anomalies and the mortality rate in this population varied with the type of cleft. All fetuses presenting a median facial cleft had concurrent anomalies (particularly of the central nervous system (86%)) and a fatal outcome. Associated defects were more frequent in fetuses with bilateral clefts (68%) than in those with unilateral clefts (38%). Fetuses with a unilateral cleft lip with or without cleft palate had a better survival rate (54%) than those with a bilateral cleft lip with or without cleft palate (35%). The frequency and type of chromosomal abnormalities varied with the type of cleft. The highest rate of chromosomal abnormalities was found in fetuses with median clefts (79%).

### Conclusion

The study revealed a strong relationship between the type of facial cleft, associated malformations, chromosomal abnormalities and fetal outcome. In pregnancies complicated by a cleft lip with or without cleft palate, patients should be informed of the risks of associated anomalies, some of which may be undetected prenatally.

