Experience of a reference Fetal Medicine Service from Southern Brazil with pregnant women who underwent fetal karyotyping

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Objective

To outline the profile of pregnant women attending a Fetal Medicine reference service in Southern Brazil who underwent fetal karyotyping.

Methods

Data were collected from medical records of pregnant women attending the Fetal Medicine Service of Hospital Materno Infantil Presidente Vargas (HMIPV), Rio Grande do Sul, Brazil, during 8 years.

Results

Our sample consisted of 155 pregnant women. Her ages ranged from 13 to 45 year-old (mean 28. 5 year-old). Forty-one patients (26. 5%) were 35 year-old or more. The main indication for fetal karyotyping was the presence of a fetal malformation (73. 5%). Chromosomal abnormalities were identified in 38 cases (24. 5%). Only 78 patients (50. 3%) had undergone first trimester ultrasound screening and 44 (56. 4%) were considered altered. Echocardiography, as well as magnetic resonance imaging (MRI), were important complementary exams, showing that the morphological ultrasound accuracy for the detection of congenital heart defects was low (22. 6%). We found that at time of procedure, fetuses with karyotypic abnormalities have been more often classified with multiple than isolated defects, when compared to normal fetuses (P = 0. 007348). Fetuses with multiple defects were 1. 5625 times more likely to have a karyotype abnormality. The presence of multiple defects was considered a satisfactory approach for invasive karyotyping (area under ROC curve 0. 624). Higher frequencies of chromosomal abnormalities were seen in fetuses presenting commitment of the following systems: hygroma/hydrops (P= 0. 0219), cardiovascular system (P= 0. 0003) and face and neck (P= 0. 0039). There was a tendency to have a negative association between chromosomal abnormalities and anomalies of the urinary tract (P= 0. 0927).

Conclusion

Fetal echocardiography and MRI could be useful in order to classify in multiple or isolated defects, thus better indicating the invasive procedure. Pregnancy planning allied to a greater access to health care and a better qualification of the ultrasound performing physicians would also help to increase this prenatal detection of malformations and hence chromosomal abnormalities.