17th World Congress in Fetal Medicine

The misuse of cell-free DNA by obstetricians in cases of abnormal ultrasound findings

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Objective

The introduction of cell-free DNA for the screening of chromosomal abnormalities prompted its misuse in cases of fetal abnormalities detected by ultrasound. Our aim is to evaluate which chromosomal abnormalities would be missed if only cell-free DNA was used in cases of abnormal ultrasound findings.

Methods

We retrospectively analyzed cytogenetic results of patients who underwent invasive procedure test, from December 2013 until March 2018, in a private diagnostic center in São Paulo, Brazil. 83 cases underwent karyotyping (96, 5%) based on ultrasound findings and 3 (3, 5%) based on maternal age.

Results

A total of 86 invasive procedures were performed, 56 (65, 1%) chorionic villus sampling (CVS) procedures and 30 (34, 9%) amniocentesis procedures(AC). Chromosome abnormalities were found in 34 cases (39, 5%). 49 tests (56, 9%)were normal, only 3 cases (3, 5%) had culture failure. The most frequent abnormality was Trisomy 21 58, 8 % (20/34), followed by Trisomy 18 11, 7% (4/34), then Trisomy 13 8, 8 % (3/34) and Monosomy X 5, 8 % (2/34). Other structural chromosome aberrations were observed in 14, 7% (5/34), all of them with major ultrasound findings: 46 XY r(4) (p16q35) ring chromosome 4; 46XX der 4+4: ? chromosome 4 derived from a translocation between 4 and another unknown; 46 XY del (7) (q 32) deletion of long arm of chromosome 7; 46 XY inv (9) (p12q13) pericentric inversion of chromosome 9 and 46 XY t (3, 12) (p25q21)translocation between the short arm of chromosome 3 and the long arm of chromosome 12.

Conclusion

Most of the procedures done were CVS when compared to amniocentesis, that is because in a private center effective early screening can be accomplished. In contrary to what fetal medicine specialists recommend, obstetricians continue to misuse cell-free DNA when anomalies are discovered during scans. Even if we assume that cell-free DNA can detect all the trisomy and Monosomy X cases, almost all the cases of structural chromosomal abnormalities, which constitutes a minimum of 14, 7% (5/34) cases, would be missed. Our data reinforces the indication of invasive procedure in fetus with ultrasound abnormalities.