Prenatal Genetics, Reproductive Genetics

AB006. Chromosome analysis in placenta with fetal anomaly

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Background: Confined placental mosaicism (CPM) represents a discrepancy in karyotype between the cells in the placenta and the cells in the fetus. The observation of multiple, chromosomally distinct cell lines in chorionic villus samples (CVS) is not an unusual finding and occurs in 1–2 per 100 samples. Some reports stated CPM is related to fetal growth restriction, but the relationship between CPM and fetal anomaly is still unclear. In this study, we focused on the fetus with anomaly and looked into placenta karyotyping.

Methods: We have recruited 96 cases with fetal anomaly and analyzed the karyotype from their placenta. Chromosome analysis was done in the range of 400–550 bands. We counted minimum of 30 metaphases total, from two or more independent cultures. Analysis was done with minimum of 5 metaphases from total (10 for anomaly case), representing two or more independent cultures.

Results: Eighty-nine out of 96 cases were analyzed. Seven were ended up unsuccessful culture due to infection and less alive cells after maceration due to intrauterine fetal death (IUFD). Seventy-four cases (92.7%) were diagnosed as normal karyotype and abnormal karyotype other than mosaicism (aneuploidy, abnormal structure) was shown in 11 cases (11.8%). Mosaicism was shown in 3 cases (3.2%) and pseudomosaicism was shown in 5 cases (5.3%). The case with mosaicism, clinically diagnosed as sacrococcygeal teratoma, fetal growth restriction and monochorionic diamniotic (MD) twin.

Conclusions: This study showed we have more cases with discrepancy of karyotype within a placenta more frequently than the previous report regarding to CVS at first trimester. It is still difficult to make clear statement which anomaly would be related to CPM.

Keywords: Confined placental mosaicism (CPM); fetal anomaly; karyotype

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