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Prenatal diagnosis of chromosomal microarray in fetuses with nuchal translucency ≥2. 5mm

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Objective

To assess the clinical value of prenatal diagnosis of using quantitative fluorescent polymerase chain reaction (QF-PCR) and chromosomal microarray analysis (CMA) for the examination of genomic imbalances in prenatal amniotic fluid samples from fetuses with nuchal translucency (NT) above or equal to 2. 5mm.

Methods

From November 2015 to December 2017, we included 156 amniotic fluid samples from consecutive ongoing pregnancies, with fetal NT ≥2. 5 mm at 11–13+6 weeks' gestation, from Prenatal Diagnosis Center in West China Second University Hospital. All cases were examined with QF-PCR and then analyzed by CMA in those with normal QF-PCR results.

Results

Of the 156 cases, common aneuploidies were detected by QF-PCR in 18 (11. 5%) cases (11 cases of trisomy 21, 3 cases of monosomy X, 3 cases of trisomy 18, 1 cases of trisomy 13). One case of trisomy 21 mosaicism and one case of X/XX mosaicism were confirmed by fluorescence in situ hybridization (FISH). Among the 136 cases with normal QF-PCR results, microarray detected additional pathogenic copy number variants (CNVs) in 5. 9% (8/136) of cases. Two cases would have expected to be detectable by conventional karyotyping because of large deletions/duplications (>10 Mb), leaving six cases (4. 5%; 6/134) with pathogenic CNVs only detectable by CMA.

Conclusion

It is rational to use a diagnostic strategy in which CMA is preceded by the less expensive, rapid, QF-PCR to detect common aneuploidies. CMA allows detection of a number of pathogenic chromosomal aberrations in fetuses with NT≥2. 5mm.