ABSTRACT

Triploidy presents a chromosomal abnormality in which is characterized by an extra-haploid set of chromosomes (69 chromosomes instead of the normal 46). The incidence of triploidy is extremely hard to calculate since most of the cases result in miscarriage or stillbirth in the first trimester.

CASE REPORT

Hanoi Hospital of Obstetrics and Gynecology received one of such case in 2016 in which the patient of 27-year maternal age with no abnormal medical/pregnancy history.

NIPT (non-invasive prenatal testing) 15 weeks of gestational age returned low-risk while ultrasound images at 15 weeks and 2 days showed a reduction of amniotic fluid but no particular abnormal findings. We performed regular check-up every 2 weeks. One month after, ultrasound images found severe intrauterine growth restriction (BPD: 19th week, AC and FL: 16th week), bilateral ventricle: 9.5 mm, with olygoamnion.

Amniocentesis was performed and followed by Prenatal BoBs and conventional karyotyping techniques. Prenatal BoBs resulted normal 46XX with no microdeletions, QF-PCR showed 3 copies for chromosome 13, 18, 21, and 23 while karyotyping resulted 69XXX after 3 weeks of culture.

In this case of triploidy, ultrasound in the first trimester of gestation showed no particular sign of abnormality but by the second trimester, intrauterine growth restriction was found. Hence, amniocentesis was advised. Due to the priciple of analysis, both NIPT and Prenatal BoBs are not capable of detecting such abnormality of triploidy.

CONCLUSIONS

Through this case, we recognized that most of cases with early intrauterine growth restriction (from the second trimester of pregnancy) should be advised to invasive procedure (amniocentesis) for genetic analysis in which karyotyping should still be prioritized for its ability to accurately identify chromosomal aneuploidies.