Severe pre-eclampsia in a diandric triploid pregnancy

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Triploidy is a rare chromosomal abnormality in which there is an extra haploid set of chromosomes. It has an estimated prevalence of 1 in 3,300 pregnancies between 11-14 weeks gestation. The majority of affected pregnancies undergo spontaneous demise prior to 20 weeks gestation.[1]

The additional haploid set of chromosomes can be of maternal (dygynic) or paternal (diandric) in origin. These two types of triploidy have different phenotypes, which can be often differentiated on ultrasound (Table 1).

We report a case of diandric triploidy that could not be diagnosed using counting-based NIPT.

Case Report

A 27 year old para 1 was referred with severe hypertension and an ultrasound demonstrating fetal growth restriction at 18+6 weeks of gestation. Antenatal history included a low probability non-invasive prenatal testing (NIPT) for the common trisomies and a 13-week ultrasound, which was reported as normal.

On arrival, she was found to have severe hypertension (190/110 mm Hg), requiring intravenous anti-hypertensive therapy. Associated symptoms include headache, mild dyspnea, right upper quadrant pain and significant lower limb oedema.

Ultrasound findings

Tertiary ultrasound demonstrated a severely growth restricted fetus with multiple anomalies including bilateral echogenic kidneys, sandal gap toes and syndactyly (Figures 1-2). The placental was cystic with a hypercoiled cord (Figure 3). Maternal ovaries were enlarged with multiple theca lutein cysts (Figure 4). Subsequent cytogenetic analysis from the amniotic fluid was confirmatory for diandric triploidy, 69 XXX.

Antenatal & Postnatal Course

Termination of pregnancy was performed on maternal medical grounds, secondary to worsening hypertension and increasing oxygen requirements. The woman made a slow but progressive recovery and was discharged on day 6 with serial quantitative hCG tracking.

References

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2. Park JE, Park JK, Kang MY, Cho I, Baek JC. Counting-based cell-free DNA screening test fails to identify triploidy – A case report. *Clin Case Rep*. 2018;7(1):90-93.

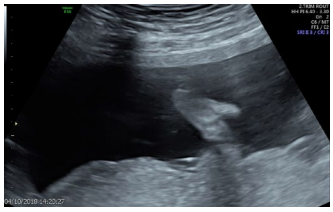

Figure 1

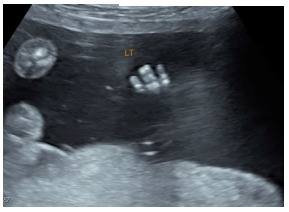

Figure 2

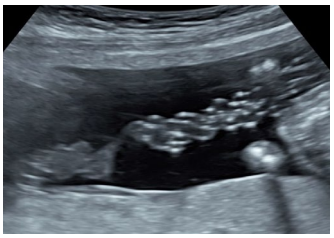

Figure 3

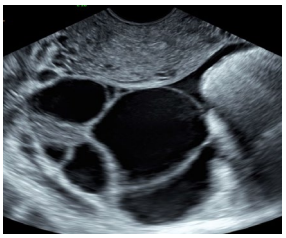

Figure 4

Table 1. Diandric vs Digynic Triploidy

	Diandric	Digynic
Origin	Paternal	Maternal
Fetal growth restriction	Moderate Proportionately normal sized parts	Severe Growth restricted trunk and limbs, normal to large head
Placenta	Large, hyperplastic with molar changes.	Small, non molar.
First trimester screening	↑ NT and hCG Often high risk for T21	Normal NT ↓ hCG and PAPP-A

Conclusion

Diandric triploidy may be associated with severe maternal complications such as pre-eclampsia and persistent trophoblastic disease.

NIPT methods using a quantitative counting approach that relies on comparing the absolute number of sequences might not be able to detect cases of triploidy, when compared with those utilising a single nucleotide polymorphism (SNP)-based approach. [2]

Hence, detailed first trimester ultrasonographic assessment should always be undertaken, even in the context of a low risk NIPT result.

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