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Case Report

A case of triploidy detected by maternal serum screening in the first trimester of pregnancy

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Abstract

Introduction: Here we report a case of diginyc triploidy, which resulted from the fertilization of a diploid ovum by a single sperm.

Case presentation: A caucasian 24-year-old woman presented in her first pregnancy, naturally conceived. A maternal serum screening and an ultrasound examination was performed to assess the risk of aneuploidy in pregnancy and only the serum screening has raised concerns regarding aneuploidy. After genetic counseling an amniocentesis was performed, showing a 69, XXX karyotype.

Conclusion: Our findings remarked that the first trimester screening at 11-13 weeks' gestation for trisomy is able to detect other chromosomal abnormalities like triploidy.

Introduction

Triploidy is characterized by an extra haploid chromosome set (3n=69), and resulting in lethal chromosomal abnormalities because most triploid fetuses undergo spontaneous abortion before 20 weeks' gestation: the prevalence of triploidy at the 11-14-week ultrasound scan is approximately 1:3300, the incidence of triploidy in liveborn infants is approximately 1:10.000 [1-3]. The origin of the additional chromosomal set could be maternal (digynic) or paternal (diandric).

Case presentation

A Caucasian 24-year-old woman presented in her first pregnancy, naturally conceived. The obstetric history was negative and the medical history proximate and remote showed no comorbidities. The patient was sent to our ambulatory to perform the first trimester screening for trisomy 21-18 at 11-14 weeks' gestation. The screening parameters included nucal translucency (NT), maternal serum free β-human chorionic gonadotropin (β-hCG) and pregnancy-associated plasma protein-A (PAPP-A).

The ultrasound and NT measurement was performed by a certified sonographer: NT was 0.9 mm (Multiples of median = MoM 0.72), the biometrics was normal, and there were no morphological abnormalities. The maternal serum free β-hCG was 3.69 UI/L MoM 0.10 and PAPP-A 0.08 UI/L Mom 0.02. The risk of trisomy 21–18 was calculated using a dedicated software (Ssdwlab 5) and there was a low risk: 1:4830 for trisomy 18 and 1:30414 for trisomy 21. There was only a marked decrease in maternal serum free β-hCG and PAPP-A. After genetic counselling, an amniocentesis was recommended in order to exclude aneuploidies related to serum screening.

Amniocentesis was performed at 17 weeks' pregnancy, twenty millimeters of amniotic fluid sample was collected ; which revealed detectable ultrasound abnormalities: bilateral hydrocephalus, agenesis of the corpus callosum, oligohydramnios and asymmetric growth restriction.

The fetus showed a 69, XXX karyotype; the fetus presented intrauterine fetal demise diagnosed at the 18 weeks' gestation. The chromosomal preparation obtained from fibroblasts confirmed the diagnosis. The postmortem examination findings revealed the intrauterine growth restriction, facial abnormalities. No further pathologic findings were revealed due to fetal maceration.

Discussion

Triploidy is estimated to occur in 1% of all conceptions [4]. The prevalence of triploidy at 12 weeks' gestation is estimated to be 1/3500 compared with 1/30000 at 16 weeks [5] because a large part of fetuses were aborted during the first and second trimester. A recent study published by Engelbrechtsen and collegue [6] showed that in their cohort study, the incidence of triploidy is 1:6614, suggesting that triploidy is not as common as shown by others. However there is a great significance attached to detecting triploidy early in pregnancy, because of the risk of maternal complications such as preeclampsia and persistent trophoblastic disease [7-9]; early diagnosis can contribute to an early decision on possible termination of pregnancy and reduced the psychological impact of carrying a fetus with a letal condition.

Increased maternal age isn't a risk factor in triploidy and there isn't an increased recurrence risk [10].

There are two phenotypes of triploidy depending on the origin of the extra haploid chromosome set: the type I diandric if extra haploid

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chromosome is paternal; type II digynic if extra haploid chromosome is maternal. The different karyotypes result in markedly different phenotypes [11-15].

Type I, the diandric phenotype is characterized by a moderate growth restriction or it is relatively well - grown, proportional body parts and is associated with a large hyperplastic placenta with partial hydatidiform mole changes. First trimester screening usually shows increased NT and levels of free β-hCG in the maternal serum and mildly decreased PAPP-A [16]; the diandric triploid fetuses are often found to have an increased risk for trisomy 21.

The type II, digynic type, which is the most common, is characterized by a small normal looking placenta and severe asymmetrical growth restriction, with a growth restriction of the trunk and limbs and a normal o large head. The fetal NT is normal, with markedly decreased maternal serum total free beta-hCG and PAPP-A. Previous studies have confirmed this serological picture in the two phenotypes [17,18].

Digynic triploid fetuses can be difficult to detect at first-trimester screening. A number of characteristic abnormalities are common in both types and there is no difference between the two phenotypes: the anomalies of the brain, heart and urinary tract, abnormal genitals and syndactyly of the third or fourth fingers and of the toes [11-13,19-22].

In our case, the biochemical findings were comparable with the result s of the previous report of maternal origin: serum free β-hCG was 3.69 UI/L MoM 0.10 and PAPP-A 0.08 UI/L MoM 0.02 was very low. The NT was normal, and the risk of trisomy 21-18 was not increased. Our report remarked that there was a large deviation in the maternal serum and the biochemistry of fetus with digynic triploidy is to be identified during the first trimester screening. In fact the risk of triploidy is not related to maternal age and other sonographic markers don't show substantially different values in euploid fetuses [23,24].

Anyway, in our case there was not a severe intrauterine growth restriction at the first ultrasound scan.

Conclusion

Our findings remarked that the first trimester screening at 11-13 weeks' gestation for trisomy is able to detect other chromosomal abnormalities like triploidy.

Other study demonstrate that maternal serum screening in the first trimester of pregnancy are able to detect triploid fetuses. Our study add a case of digynic triploid fetus, without ultrasound anomalies, but only with anomalies of maternal serum biochemistry.

Competing interests

The author(s) declare that they have no competing interests.

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