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Case Report

Obstetrics and Gynaecology Section

Uncommon Presentation of Triploidy: A Case Report

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ABSTRACT

A 28-year-old woman presented in her first pregnancy was admitted with severe hyperemesis gravidarium. Increased nuchal translucency with cardiac anomaly and omphalocele at the first trimester was observed at the ultrasound examination. Chorionic villus biopsy confirmed triploidy. The combination of type I and type II triploidy patterns were seen together in the second trimester of the pregnancy. Although the symptoms due to increased human chorionic levels occured, at the pathologic investigation there were no molar changes in the placenta. Here we report a case of uncommon presentation of triploidy.

Keywords: Molar pregnancy, Nuchal translucency, Omphalocele

CASE REPORT

A 28-year-old nulliparous woman with 7 weeks of gestation was admitted to our clinic with severe nausea and vomiting. Weight loss and persistent ketonuria necessiated in patient treatment for several times. Ultrasound at 12th weeks of gestation revealed a nuchal fold of 6 mm, ompalocele and Atrioventricular septal defect [Table/Fig-1].

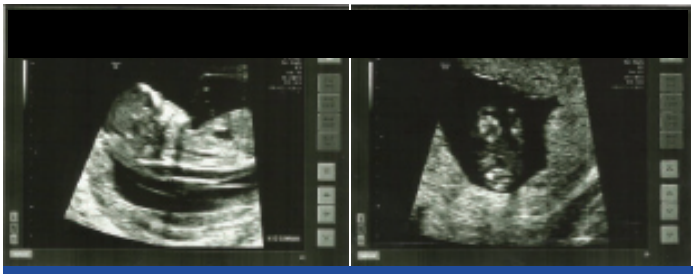
Chorionic villus biopsy confirmed triploidy (69 XXY). At 16th week of pregnancy, the couple took a decision for termination of pregnancy. While planning for abortion, the patient was re-evaluated and the following findings were noted: bilateral theca Lutein cysts in (12 cm diameter), hypertension (160/100 mmHg), 1 (+) proteinüria and symptomatic hyper-thyroidism. On ultrasonographic examination, enlarged placenta and growth resctriction was observed. Following the termination of pregnancy, regression of all symptoms was observed in a few months. On pathologic examination of placenta no molar changes were noted [Table/Fig-2].

Post mortem examination revealed a fetus of 14 weeks size confirming, intrauterine growth reterdation, omphalecele, facial abnormalities including low-set ears and talipes equinovarus [Table/Fig-3].

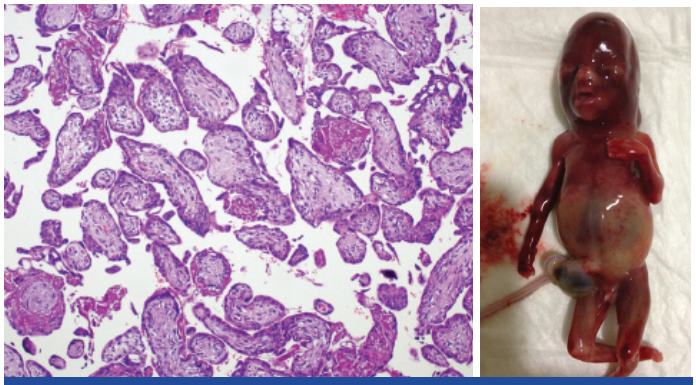
DISCUSSION

Triploidy is a chromosomal disorder that arises at conception with a complete extra set of chromosomes (3n:69) and estimated to occur in 1% of all conceptions [1]. Most of these conceptions results in spontaneous abortion of pregnancy. Common findings include limb anomalies, microcephaly, and neural tube defects [2].

Triploidy can be distinguished into two types according to the parental origin [3]. In type I, where the additional chromosome set is of paternal origin (diandric), the placenta is usually enlarged and partially molar with elevated levels of maternal serum beta human chorionic gonadotropin (β-hCG) and the fetus is relatively well grown with increased nuchal translucency. Partial molar gestations are usually associated with triploidy of diandric origin.



[Table/Fig-1]: Ultrasound at first trimester



[Table/Fig-2]: Pathologic investigation of the placenta **[Table/Fig-3]:** Triploidic fetus

Type II triploidy which the additional chromosome set being of maternal origin (digynic) is the most common type of triplody. It is characterized by a small placenta with decreased levels of β-hCG and asymmetrical fetal growth restriction. Malformed hands, cardiac anomalies and face abnormalities are the common structural defects in both types of Triploidy [4].

In the present case signs of elevated levels of human chorionic gonadotropin and growth restriction were together. The finding of growth restriction in a type I triploidy seems atypical but has been reported [5]. Daniel et al., reported 19 cases of triploidy which was diagnosed by inheritance of DNA micro satellites and by methylation patterns of SNRPN or PW71. The parents blood was unavaliable. The phenotype of the fetuses were divided into two types described by McFadden and Kalousek. Three of six fetuses with Type I triploidy showed mild to moderate growth retardation. The study suggested that the fetal triploid 'Type 1' definition be modified to 'well grown to moderate symmetrical IUGR' to allow for such variation [6].

Unlike other common chromosomal abnormalities, Triploidy may affect the mother with varying degrees of pre-eclampsia Rijsinghani et al., reported seven patients with triploid pregnancies who became preeclamptic. Four of them showed fetal growth restriction [7].

Hyperemesis gravidarum and theca lutein cysts were also showed increased hormone levels in our case. Triploidy cases with preeclampsia are usually with molar changes in the placenta [8] In our case,

no molar changes were found in the careful investigation of the placenta. It should be born in mind that placentomegaly without molar degeneration could cause this symptoms.

CONCLUSION

Our case demostrates that characteristic symptoms of type I and type II tripliody may be together and a molar placenta is not essential for increased human chorionic gonadotropin symptoms. Our case and the literature showed that growth retardation and increased human chorionic gonadotropine levels may be together. The definition of Type I Triploidy may be modified according to new data.

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