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Case Report

An insight into the placental morphology in a case of Edwards syndrome

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ABSTRACT

Certain histomorphological features of the placenta can suggest, although not diagnose, the presence of abnormal karyotype in cases of chromosomal anomalies in the fetus. However, data correlating placental histology and abnormal fetal karyotype is scarce. Here, in this article, we wanted to enlighten the readers about the importance of the placental morphology in chromosomal aberrations by presenting a case of a 22-year-old primigravida who at 8 weeks of gestation had a regular prenatal check-up. Over a period of time, the ultrasound study at 21 weeks of gestation revealed a single live intrauterine fetus in a breech position with polyhydramnios, strawberry-shaped skull, and other features. Amniocentesis revealed that the fetus was having Edwards syndrome (trisomy 18). The placental examination showed areas of sclerosis and congestion with a single umbilical artery.

Keywords: Fetal anomalies, Histomorphology, Placenta, Karyotyping, Trisomy 18

INTRODUCTION

Approximately 10-20% of all clinical pregnancies end in abortion. It's etiology is often complex and the majority of early abortions are usually due to chromosomal defects in the embryo. Autosomal trisomy is the most common (61.2%) genetic abnormality followed by triploids (12.4%), monosomy X (10.5%), tetraploids (9.2%), and structural chromosomal anomalies (4.7%).[1] The importance and role of the placenta in fetal cytogenetic abnormalities have been seldom documented in world literature. Here, we describe a case of Edward syndrome (trisomy 18) with emphasis on the utility of the placenta in such a rare chromosomal aberration.

CASE REPORT

A 22-year-old primigravida female presented at 8 weeks of gestation to the gynecological outpatient department for a regular prenatal check-up. Ultrasonography (USG) was performed and revealed a single live intrauterine fetus (SLIUF) and the expected date of delivery was calculated. Next USG was performed at 19 weeks and 1 day of gestation, which revealed SLIUF with a single umbilical artery and variable lie. The amniotic fluid was adequate and the expected fetal weight was 240 grams. The next USG was done when the patient was at 21 weeks of gestation. The USG revealed SLIUF in breech position with polyhydramnios, the fetal skull was strawberry-shaped, and the umbilical cord showed only 2 vessels. There was intrauterine growth retardation and the fetus had an atrioventricular septal defect with a club foot. The menstrual history of the patient included complaints of irregular cycles. Her family history for any congenital abnormalities was non-contributory. Amniocentesis was performed and the fetus was diagnosed with trisomy 18 i.e., Edwards syndrome. The patient opted for medical termination of pregnancy soon afterwards. The fetus was sent for autopsy to the forensic medicine department. The placenta and membranes were delivered completely and sent for histopathological examination. On gross examination, the placenta weighed 100 grams, and the

placenta measured 10×7.5×2.5 cm. The maternal surface had 4 cotyledons and the attached umbilical cord measured 19 cm in length which on the cut section showed the presence of only two vessels (Figure 1). Microscopic examination revealed focal areas of sclerosis and congestion in placental tissue with membranes showing mild chorioamnionitis (Figure 2). Microscopic examination further confirmed the gross finding of the presence of only two vessels in the umbilical cord (Figure 3).

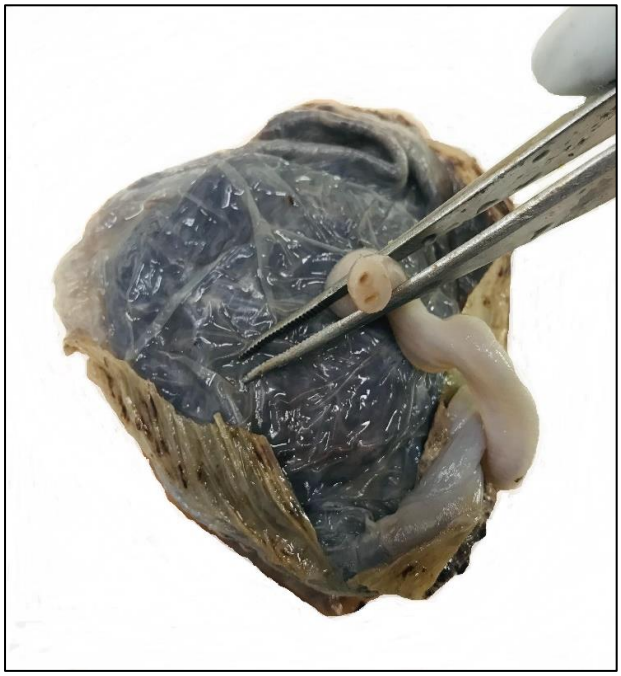

Figure 1: Small placenta with attached umbilical cord showing two vessels.

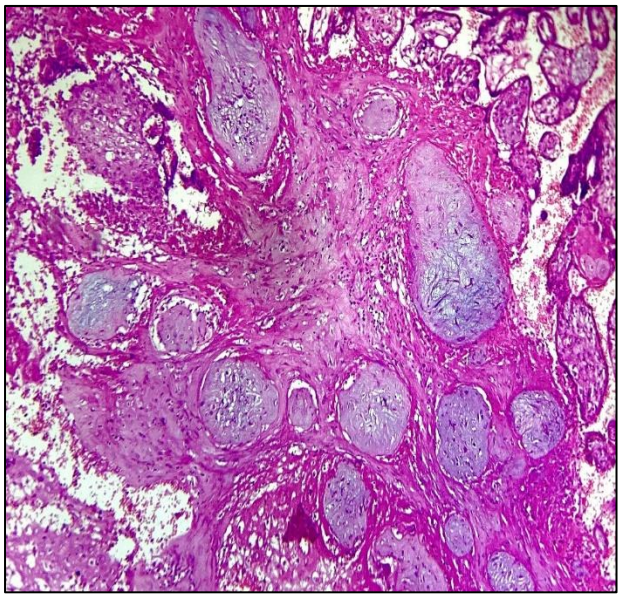

Figure 2: Placental parenchyma showing areas of sclerosis (H and E, ×10).

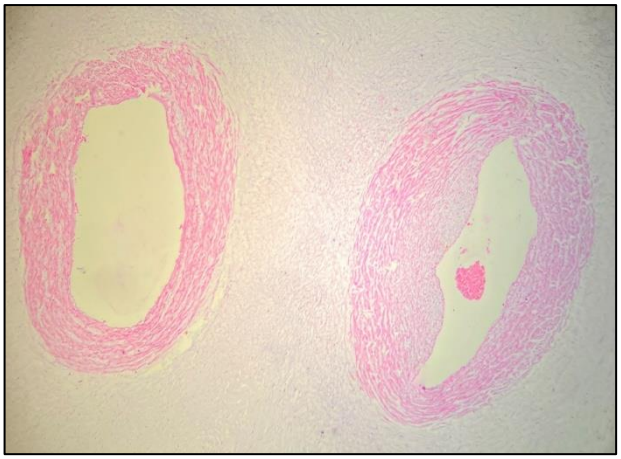

Figure 3: Two-vessel cord (H and E, ×4).

DISCUSSION

Abortion is defined by Ralph Benson in the "Handbook of Obstetrics and Gynecology" as "the termination of pregnancy before the fetus is viable" while WHO defines it as "the expulsion or extraction from its mother of a fetus or embryo weighing less than 500 grams." About 10-20% of pregnancies end in abortion and another 10% are induced illegally.

About 50-60% of abortions occur due to fetal chromosomal abnormalities.[2,3] Maternal age and prior history of abortions are some of the risk factors. Other causes of abortion can be structural uterine abnormalities like leiomyoma and gynecological infections such as pelvic inflammatory diseases, tuberculosis, etc. Several chronic diseases such as anti-phospholipid antibody syndrome and thyroid disorders etc. can also cause spontaneous abortions.[4]

Edwards syndrome (trisomy 18) occurs in 1/5000 births but the incidence is higher in the prenatal period with a high percentage of fetal losses.[5] It had been studied that about 59% of the time trisomy 18 results from maternal meiosis II error.[6] It is characterized by failure to thrive, development retardation, abnormally shaped skull, low set, malformed ears, etc. Congenital heart diseases like ventricular septal defects are also seen. Postnatally, 60% of trisomy 18 children die within 2 months, and more than 95% within a year. In our case, the fetal skull was strawberry-shaped and the fetus had an atrioventricular septal defect with a club foot.

The evaluation of chromosomal abnormalities is usually done through a combination of radiology and biochemical markers. Corresponding histological examination of the placenta or chorionic villi is seldom performed.[7] Therefore, there is limited literature correlating specific placental histomorphology with chromosomal abnormality.[8] In clinical settings where fetal malformations are absent or missed in radiology, placental morphology can potentially help in favoring or ruling out

cytogenetic abnormalities. It is generally believed that placental histomorphology is normal in most cytogenetically normal fetuses although the placental features may vary with regard to shape and size.[9]

Histomorphological features of placentas of fetuses with triploidy (such as in cases of hydatidiform mole) are well studied and include the presence of two populations of villi; normal and hydropic with hydropic villi showing scalloping, trophoblastic inclusions and hyperplasia.[10]

Few researchers have suggested that the presence of villous hydrops with prominent individual intravillous cytotrophoblast cells is associated with abortions due to autosomal trisomies; hydropic villi with infarction and hemorrhage are seen in cases of tetraploidy and hypoplastic, hypovascular fibrotic villi are associated with monosomy X.[11] In trisomies, many features can be seen like a single umbilical artery, delayed maturation, decreased vascularity, increased villous stromal cellularity, and small placentas.[8,12,13]

According to Honore et al, cystic and dilated chorionic villi with hydropic change are seen in the placenta of trisomy 18 patients while Shah et al, have documented placental membrane necrosis along with focal pseudovillous papilliform cytotrophoblastic proliferation, large and cellular chorionic villi with convoluted outlines as its features.[8,11] In the present case, the placenta was small-sized, weighing only 100 grams. There was the presence of a single umbilical artery. However, microscopy of the placenta in our case had non-specific findings such as focal areas of congestion and sclerosis. So, it is being postulated that all the findings may not be seen in every placenta belonging to the fetus with cytogenetic abnormalities. However, placental histomorphology plays an important role, similar to USG, as it can clue us on when to go for fetal karyotyping via chorionic villi sampling or amniocentesis.

CONCLUSION

Our case underscores the importance of placental evaluation especially in cases of abortions. The placental morphology in such scenarios can be an important predictor and indicator of a karyotypically abnormal fetus. However, a deeper understanding of the genesis of specific placental pathology in complex and diverse chromosomal abnormalities is required in the near future.

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