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Early Prenatal Diagnosis of Triploidy With Rare Ultrasound Findings

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Introduction

Triploidy, a rare chromosomal abnormality, occurs in approximately 1% of conceptions. Many triploid conceptions are aborted spontaneously in the first trimester. Triploidy associated with fetal development and survival beyond the first trimester is rare. The prevalence of triploidy at 16 to 20 weeks of pregnancy has been estimated to be 1 in 5,000.

Many malformations have been reported in affected fetuses, including holoprosencephaly with median cleft lip, gastroschisis, encephalocele, myelomeningocele, syndactyly, clubfeet, heart defects, and renal agenesis.[1] The rare manifestations include cyclopia with proboscis which is found in 1: 40,000 triploid fetuses.[9]

Case Description

Case 1: A 24 year old female presented with 12 weeks 6 days of amenorrhoea by LMP for nuchal translucency scan with no complains of vaginal bleeding, lower abdominal pain and any previous abortions. Previous scan done at 6 weeks of gestation showed single intrauterine pregnancy with normal cardiac activity.

Imaging findings: USG was performed by transabdominal and transvaginal route. Single intrauterine gestation with fetal heart rate 151 beats per minute was seen. Nuchal translucency was increased measuring 2.6 mm. Fetus showed slightly enlarged head with cystic area replacing bilateral cerebral hemisphere with thin rim of cortex at periphery. Falx was seen anteriorly in midline. Posterior fossa structures seen, thalamus seen in midline. In the face (coronal view) there was an echogenic rounded structure seen in midline at the level of orbits likely proboscis. Single orbital opening and probosis was seen in midline. Abdominal wall appeared slightly edematous. Gestational age was 12 weeks as per USG and 12 weeks 4 days by LMP.

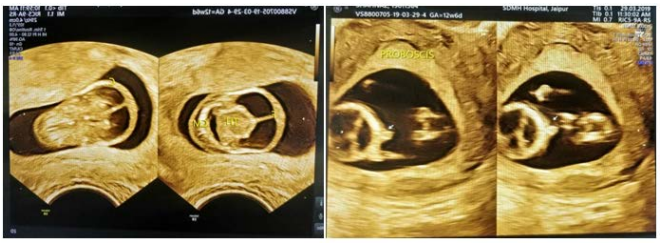

Figure 1 & 2: USG images showing hydrencephalous and proboscis.

Diagnosis

In view of above USG findings like proboscis with cyclopia, hydrencephaly, changes of hydrops foetalis suspicion of chromosomal abnormality like triploidy was considered. The patient underwent medical termination of pregnancy and the products of conception were sent for chromosome profiling. The gross specimen confirmed the USG findings of proboscis and cyclopia. Chromosomal profiling showed abnormal 3 copies of all 46 chromosomes suggesting triploidy.

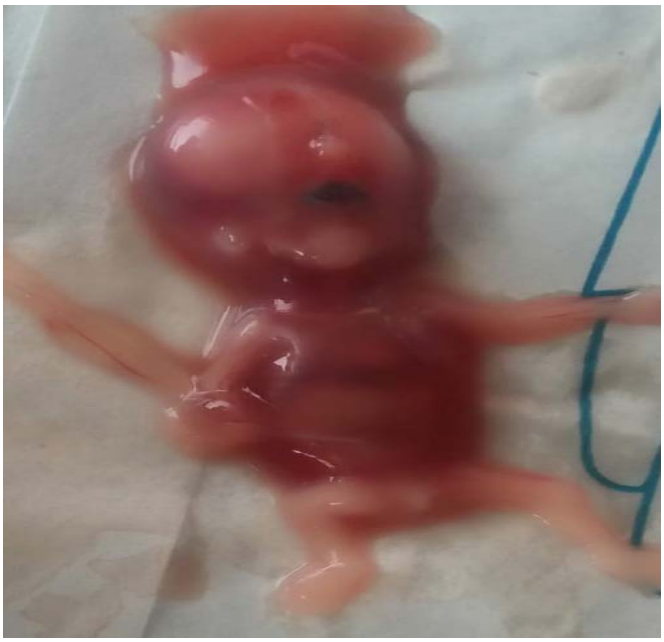

Figure 3: Gross specimen confirmed the USG findings of proboscis and cyclopia.

Discussion

Triploidy is a rare genetic condition with adverse fetal outcome. The triploidy in this report was detected by identification of multiple anomalies on routine USG scan at 12 weeks of gestation. Although anomalies vary between different cases, the fetus in this case had significant US findings, resulted in elective termination of pregnancy.

The most distinguishable sonographic feature of fetal triploidy is IUGR with a reported incidence of 55–100% in different case series, and with an average growth lag (difference from that expected by dates) of 2–6 weeks.[2,3] Asymmetrical growth restriction is observed in triploid fetuses with the trunk being more severely compromised than the head at 11-13 +6 weeks of gestation. [4] The occurrence of IUGR is potentially evident as early as 10 weeks' gestation.[5] In this case however IUGR was not observed.

CNS anomalies are the second most common sonographic features following IUGR, with a reported frequency of approximately 50% of triploid cases.[6] The reported associated CNS anomalies include ventriculomegaly, Dandy–Walker malformation or variant, agenesis of the corpus callosum, holoprosencephaly (HPE), interhemispheric cyst, posterior fossa cyst, encephalocele, neural tube defects, and others.[7]

Holoprosencephaly is a complex developmental abnormality of the brain arising from failure of cleavage of the prosencephalon. The incidence is not known because milder forms may be unrecognized. Cyclopia, which is included in the term holoprosencephaly, has been reported to occur in 1out of 40,000 births.[9]

Increased fetal NT, characterized as a nonspecific and transient fetal anomaly, has been significantly associated with chromosomal abnormalities including triploidy. Early detection of triploidy leads to the identification of more than 65% of triploid fetuses with increased fetal NT.[3] The fetal translucency in this case was increased.

Facial anomalies seen in prenatal triploids include microcephaly or relative macrocephaly, low-set ears,

micrognathia, hypertelorism, facial clefts, cyclopia, proboscis, single nostril, and others.[8]

Conclusion

With the advancement of ultrasound equipment, the use of prenatal ultrasound has greatly improved the detection of triploid fetuses in obstetric practice. The increased knowledge of different sonographic features is helpful in detecting triploid fetuses throughout gestational life. Although several diseases have phenotypic overlaps with triploidy, major sonographic features that point towards the possible diagnosis of triploidy are extra-fetal anomalies such as an enlarged and cystic placenta, oligohydramnios, and enlarged maternal ovaries with multicystic change, and fetal structural anomalies such as IUGR, increased fetal NT, CNS anomalies, facial anomalies, genitourinary anomalies, cardiac anomalies, gastrointestinal anomalies, and limb defects. Clinically, several diseases may have phenotypic overlaps with triploidy; therefore, early first trimester sonographic identification of these anomalies in triploid fetuses is important in providing earlier karyotypic confirmation and enabling more reasonable management for pregnant women.

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