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Fetal Triploidy Syndrome – A Case Report

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Abstract
The fetal triploidy syndrome presentations include intrauterine growth retardation, hydrocephalus, oligohydramnios, and cystic changes of the placenta. Although the majority of triploid conceptions abort spontaneously in the first trimester, rarely they progress to term. Determination of the karyotype is important for the management of a pregnancy with a live fetus, and has implications for genetic counseling of subsequent pregnancies. We present a case of fetal triplody syndrome with large placental cyst and hydrocephalus.
Key Words: *Triploidy, Antenatal ultrasonography, Placental cyst, Congenital malformations, Karyotype.*

Introduction

Triploidy is a chromosomal abnormality where three instead of two haploid sets of chromosomes are present. In other words, in humans there are 69 instead of 46 chromosomes. Triploidy is a rare syndrome estimated to occur in about 2% of all pregnancies.[1] Majority of triploidy syndromes abort spontaneously in first trimester and rarely progress beyond that. The triplody are associated with multiple congenital anomalies and is not compatible with long term survival.[2]

Case Report

A 24 year old female, G2P0+1, presented to emergency department in 37 weeks of gestation with labor pain. She had history of headaches in this pregnancy. She had received treatment for hypertension elsewhere and had documented proteinuria. She had one history of spontaneous abortion at 12 weeks of gestation previously. Antenatal ultrasound was not done yet. She had blood pressure of 152/90 mm of Hg, pulse rate of 132 per minute with no respiratory distress at admission. On pelvic ultrasound, a large hypoechoic area was noted arising from fetal

surface of placenta. In view of fetal distress, emergency LSCS was done. A hydrocephalic male baby was delivered. Placenta with membranes was taken out. On inspection, the placenta delivered had a large fluid filled cyst of dimension 15×6×5 cm (figure 1).

Figure 1. Large placental cyst

Mother was hemodynamically stable post operatively. Baby had hypertelorism, syndactyly and polydactyly in all the four limbs .Computed tomography of head of the neonate showed hydrocephalus with aqueductal stenosis. His echocardiography and USG abdomen were normal. In view of large placental cyst, polydactyly, syndactyly, hydrocephalus, hypertelorism and significant obstetric history, the diagnosis of fetal triploid syndrome was made. Karyotype was not done because of technical difficulties. Parents were counseled and the baby was referred to higher center for further evaluation and management.

Discussion

Three different mechanisms may produce triploidy: 1) nondisjunction in the first or in the second meiosis of spermatogenesis, resulting in an extra set of paternal chromosomes (diandry); 2) nondisjunction in the first or in the second meiosis of oogenesis, resulting in an extra set of maternal chromosomes (digyny); 3) double fertilization of a normal egg, resulting in an extra set of paternal chromosomes (dispermy).[3] Triploidy syndrome is characterized by general dysmaturity, muscular hypotonia, large posterior fontanelle, low set dysmorphic auricles, hypertelorism, microphthalmia and coloboma, cutaneous syndactily of the third and the fourth finger, simian crease, micrognathia, major facial anomalies, holoprosencephly, agenesis of the corpus callosum, cardiac malformations, omphalocele, renal hypoplasia and other anomalies, hypospadias and/or maldeveloped external genitalia.4 In our case, the baby had polydactyly, syndactyly, hydrocephalus, hypertelorism. Triploidy can be detected prenatally by cytogenetic analysis of fetal cells obtained by chorionic villous sampling (CVS) or by amniocentesis. Postnatal diagnosis is based on phenotype of the proband with cytogenetic confirmation by karyotyping. In one previous reported series, placental findings were used to diagnose triploid syndromes.[2] Antenatal ultrasonography is an important screening tool for early identification and intervention too, for diagnosis of congenital malformations associated with various chromosomal abnormalities. It may help in reducing the mental trauma and guilt of the parents associated with malformed baby, unlike in our case.

As a conclusion triploidy syndrome must be taken in consideration if minor or major abnormalities in the first or in the second trimester of pregnancy are revealed on ultrasound. Active screening in the general population should be focused on the detection of all congenital anomalies.

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