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Case Report

Turner Syndrome Associated with Cystic Hygroma in One Twin of a Dichorionic Diamniotic Pregnancy: A Unique Case Report

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Abstract

We present a rare case of dichorionic diamniotic twin pregnancy, in which one fetus had Turner syndrome associated with cystic hygroma while the other fetus was phenotypically normal. The pregnancy was terminated due to twin A being alive but with cystic hygroma and twin B having missed abortion. Because it was dichorionic diamniotic with discordant gender, karyotyping was done only on the affected fetus, revealing loss of chromosome X-Turner syndrome. This case highlights that in order to gain a better understanding of the underlying genetic pathways, it is important to send tissue samples for investigation, even for fetuses that appear structurally normal. Dichorionic may not be dizygotic.

Key Words: cystic hygroma; dichorionic diamniotic; monosomy x; karyotype; turner syndrome

Introduction:

Turner syndrome is a sex chromosome disorder caused by loss of all or part of an X chromosome. Karyotyping analysis has revealed that turner syndrome accounts for 5–20% of early abortions. [1-4] With Turner syndrome, about 99% of impacted fetuses terminate on their own before birth.5 Based on epidemiological and newborn genetic screening data from Europe, Japan, and the United States, one in 2000 to one in 3000 affected girls are born alive. [6-9] Only the case reports have discussed the occurrence of afflicted twins. As a result, we provide a case study of a dichorionic diamniotic twin pregnancy with one twin having Turner syndrome.

Case report:

A 28-year-old multipara (G3P1L1A1) was conceived spontaneously. The sonographic examination at 8 weeks of gestation showed a twin pregnancy with dichorionic diamniotic. At 18 weeks of gestation, during a routine antenatal checkup, only one fetal heart sound was heard. Hence, an ultrasound was advised. The ultrasound revealed Twin A was alive but had a multiloculated cyst in the posterior aspect of the neck, with sizes of 16mm and 13mm. The color Doppler did not show any flow in the lesion. Suggestive of nuchal cystic hygroma. Other findings included echogenic bowel and oligohydramnios. Unfortunately, twin B had no cardiac activity along with subcutaneous edema. Due to the above ultrasound scan findings, the pregnancy was terminated using mifepristone and misoprostol. The patient delivered both fetuses vaginally.

Upon gross examination phenotypically, the twin A with cystic hygroma was female, weighing 185 g, while the second twin was male, weighing 105 g. Genetic karyotyping of placental tissue from twin A with a cystic hygroma fetus revealed loss of chromosome X, suggestive of monosomy-X, Turner syndrome.

This is a rare case of twins with dichorionic diamniotic, an alive twin with monosomy X, and a phenotypically normal male with a missed abortion.

Discussion:

Cystic hygroma is a congenital malformation resulting from lymph accumulation in the jugular lymphatic sacs due to obstruction of the lymphatic system, most commonly in the fetal neck. Notably, there is an association between cystic hygromas and chromosomal aneuploidy, fetal hydrops, and intrauterine fetal demise. Approximately half of fetal cystic hygroma is associated with chromosomal abnormalities. [10-11]

In the first trimester, cystic hygroma is linked to an increased risk of fetal trisomy, particularly trisomy 21 (Down syndrome), followed by monosomy X (Turner syndrome) and trisomy 18 (Edwards syndrome). [12-13] In the second trimester, a cystic hygroma may clinically present in monosomy X. [14]

In our case, we had a twin pregnancy with dichorionic diamniotic. Since there was a discordant phenotype, we thought it was a dizygotic twin.

Approximately 14% of dichorionic twins are actually monozygotic twins, [15] so some discordant dichorionic twins may actually be monozygotic twins.

Postzygotic nondisjunction in one twin can result in heterokaryotypia for the gonosomes. [16] This condition is evident in male twins where one becomes monosomy X, presumably resulting from the loss of the Y chromosome by nondisjunction early in development. [17] The pathogenesis may be more complicated if a pure 45, X, 45, X/46, XX, or 45, X/46, XY twin is identified with a normal co-twin.

In a comparable case report, where one twin had a cystic hygroma (female fetus) and the other twin was normal (male fetus), both twins unfortunately experienced demise. [18] Upon investigation, it was revealed that one twin had a 45, X karyotype, while the second twin had a 45, XYY karyotype. The determination of zygosity should rely on DNA studies rather than chorionicity.

Even though we just sent the sample from the affected fetus, diagnosing Turner syndrome gave us a great deal of information. In this case, the patient was reassured that Turner syndrome is a sporadic occurrence. It has a low recurrence rate. It is not associated with familial inheritance or maternal age, unlike other genetic disorders such as trisomy's.

Conclusion:

In order to gain a better understanding of the underlying genetic pathways, it is important to send tissue samples for investigation for both fetuses, even though they appear structurally normal. Dichorionic, even with discordant sex, may not be dizygotic.

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