Analysis of Ultrasonographic Misdiagnosis of Cephalothoracopagus Janiceps Conjoined Twins: A Case Report

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Article Info

ABSTRACT

Background & Objective: Conjoined twins (CTs) are rare complications of monozygotic twinning. Cephalothoracopagus is the rarest subtype of CTs and occurs once in every 58 sets of conjoined twins or once in every three million births. Here we present such a case and analyze the possible reasons for ultrasonographic misdiagnosis and recommend solutions to avoid it.

Case Report: In this article, we have reported a case of Cephalothoracopagus Janiceps twinning. The ultrasonographic diagnosis was missed at 7 and 12 weeks scans. Increased thickness of nuchal translucency and absent nasal bone was observed alongside with suspected gastroschisis. The patient was referred for further evaluation at 13-14 weeks of pregnancy. Unexpectedly, two alive fetuses were reported fused in the head, thorax, and abdomen. Because of the poor prognosis, counseling was provided for parents and elective medical pregnancy termination was carried out. We have analyzed the possible reasons for ultrasonographic misdiagnosis.

Conclusion: Without applying a standardized scanning of the entire uterus both in a longitudinal and transverse approach in early pregnancy, cephalothoracopagus twinning may be misdiagnosed with a singleton pregnancy. A high level of concerns may raise for conjoined cephalothoracopagus twinning in case of finding a single fetal pole with an irregular body outline and a disproportionally large head in the presence of two separate fetal hearts in early pregnancy. Two fused heads with two brains and two sets of lower and upper extremities do confirm the diagnosis.

Keywords: Cephalothoracopagus janiceps, Conjoined Twins, Ultrasonographic misdiagnosis

Introduction

Conjoined twinning (CT) is a rare complication of monozygotic twinning. The frequency of CT is approximately 2:100,000 gestation, while the true incidence is around 0.5: 100,000 live births (1,2). The incidence is three times higher among female than male twins (2). In humans, monozygotic monoamniotic twins develop when an implanted blastocyst splits at about 9–13 days after fertilization. Failure to undergo a complete spillage of the embryonic disc at or later than 14 days results in conjoined twin development (fissure theory) (2,3,4). The etiology of CT is obscure and none of race, consanguinity, hereditary, birth order, maternal age, acute and chronic diseases during pregnancy, or maternal exposure has revealed any association with CT (3,5). Conjoined twinning has been reported in twine and triplet pregnancies following assisted reproductive techniques such as intravaginal fertilization and intracytoplasmic sperm injection (6,7). Conjoined twins are classified according to the most prominent site of attachment, plus “pagus”, a term that means “fixed” in Greek (2). Cephalothoracopagus is the rarest subtype of CTs and occurs once in every 58 sets of conjoined twins or once in every three million births (8,9); it refers to twins with fused head and thorax but separate pelves and limbs. If the single fused head has two faces each looking in opposite directions, the twins are referred to as Cephalothoracopagus janiceps twins (8,10). The term janiceps has its origin in Janus, the two-faced Roman god. When the two faces are asymmetrical and only one normal face can be seen along one side of an incomplete face (simple naris, cyclopic eyes, synotic ears, a proboscis, or two small eyes in a single palpebral fissure), it is called cephalothoracopagus
janiceps monosymmetros (10). Here we present such a case.

Case Report

A 33-year-old G2P1 pregnant woman, who had conceived spontaneously and had an unremarkable medical history with no history of drug, alcohol or cigarette smoking, underwent her early pregnancy ultrasound. The scan revealed one fetal pole, one yolk sac, and a normal fetal heart rate (FHR). The crown-rump length (CRL) was 12 mm equal to 7 weeks 3 days of pregnancy. She was scanned next time for aneuploidy screening ultrasound at 11 weeks, 4 days of the last menstrual period (LMP). The gestational age (GA) based on the CRL was equal to 12 weeks, 1 day of pregnancy. The thickness of the nuchal translucency (NT) was reported as 4 mm. The radiologist had reported possible technical errors in NT measurement and inability in nasal bone (NB) visualizing due to unfavorable fetal position. A hyperechoic herniated mass was also visualized at the level of the anterior abdominal wall and Gastroschisis was suspected. The patient was referred to a tertiary health center, (Yas Hospital, Tehran, Iran), for further evaluation. The next ultrasound exam was performed at 13-14 weeks of pregnancy. Unexpectedly, two alive fetuses were reported fused in head, thorax, and abdomen. The ultrasound diagnosis of cephalothoracopagus CTs was made. Because of the poor prognosis, counseling was provided for parents and elective medical pregnancy termination by misoprostol was carried out. The expelled twines were fused at the cranium, thorax, and abdomen to the umbilicus. Twines had a shared placenta and umbilical cord. The faces were dissimilar and on opposite sides of the head, 90 degrees to the separate spines. There was one normal face with two eyes, one complete nose, and one mouth, and one set of low-set ears (Figure 1A), and the opposing face had incomplete facial features as one cyclopic eye and irregular fused nares and mouth orifices (Figure 1B). The arms and legs of each of the twins were of similar length and development and followed the orientation of the spine (Figure 1C). The twins’ sex was not obvious but more likely were females.

Figure 1. A. Normal face with two eyes, one complete nose and one mouth, and one set of low set ears; B. Incomplete facial features as one cyclopic eye and irregular fused nares and mouth orifice; C. The arms and legs of each of the twins were of similar length and development and followed the orientation of the spine

Figure 2. A. Longitudinal section of twins, mimicking singleton pregnancy at 11 weeks 6 days of pregnancy; B. Fused brains at 13-14 weeks of pregnancy; C. Two separate beating heart at 13-14 weeks of pregnancy; D. Longitudinal section of twins at 13-14 weeks of pregnancy.
Discussion

The worldwide use of the ultrasound in prenatal screening for Down syndrome provides the diagnosis of conjoined twinning in early pregnancy. Since all CTs are monochorionic monoamnioniotic, when only one yolk sac is seen alongside two embryos in very early pregnancy, the index of suspicion should be raised. Two of first-trimester sonographic indications for possible conjoined twinning are: 1- bifid appearance of a sole fetal pole in the first trimester seen before 10 weeks of gestation, and 2- visualization of two hearts or two stomachs (3). From 8 weeks of gestation, fetal activity increases progressively, which facilitates the differential diagnosis between conjoined and non-conjoined monoamnioniotic twins (1). The first trimester (11-13w+6days) ultrasound is the best method for early diagnosis of CTs (3).

The earliest prenatal diagnosis of cephalothoracopagus twins was reported by Lam et al. 1998 (11). The diagnosis was made by vaginal ultrasound examination at 8 weeks' gestation from the findings of a single fetal pole with an irregular body outline, a disproportionally large head, and two separate cardiac pulsations and was confirmed after rescanning at 11 weeks (11). While early diagnosis of conjoined twins is feasible, it is not always easy in cephalothoracopagus twinning due to extensive conjoining; a single fused head and thorax, and limited fetal movement as was seen in our case.

Due to the monoamnioncity of the conjoined twins, a single yolk sac alongside a single CRL and normal fetal heart were observed at 7-8 weeks of gestation in our case; mimicking a singleton pregnancy and substantial missing of the second fetal heart (Figure 2A). When the second ultrasound scan was performed at 11 weeks 4 days of LMP in our case for NT measurement, the operator reported a fetus with normal fetal heart, increased NT, non-visualized NB, and herniated mass at the ventral abdominal wall, but yet to notice the second fetal heart and fused brains. Following the detection of the aforementioned anomalies, a thorough evaluation of the fetal thorax and cranium via scanning the entire uterus both in a longitudinal and transverse approach. A high level of concerns may raise for conjoined cephalothoracopagus twinning in case of finding a single fetal pole with an irregular body outline and a disproportionally large head in the presence of two separate fetal hearts in early pregnancy. Two fused heads with two brains and two sets of lower and upper extremities do confirm the diagnosis.

Conclusion

In Cephalothoracopagus twining a single yolk sac alongside a single CRL and normal fetal heart at 7-10 weeks of gestation may mimic a singleton pregnancy and lead to missing the second fetal heart and fused brains without applying a standardized scanning of the entire uterus both in a longitudinal and transverse approach. A high level of concerns may raise for conjoined cephalothoracopagus twinning in case of finding a single fetal pole with an irregular body outline and a disproportionally large head in the presence of two separate fetal hearts in early pregnancy. Two fused heads with two brains and two sets of lower and upper extremities do confirm the diagnosis.

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Conflict of Interest

Authors declared no conflict of interests.

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