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CASE REPORT

Biamniotic Parasitic Conjoined Twins with Discordant Genotype

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Introduction

Conjoined twin pregnancies are uncommon. The prevalence of conjoined twins in the first trimester of pregnancy is estimated as 1:50,000, with an unexplained female predominance (3:1) [1,2]. It is thought that at 13-14 days gestational age an incomplete fission of the germinal disc causes the development of a conjoined twin pregnancy, obligatory being associated with a monochorionic monoamniotic placentation [3,4]. We present a rare case of monochorionic biamniotic conjoined twins with discordant genotype for sex chromosomes and postulate on its developmental origin.

Case Description

A 30-year-old primigravid woman was referred to our clinic at 12 weeks gestational age with a suspicion of an acardiac twin on a routine first trimester ultrasound scan. Expert abdominal and transvaginal two- and three-dimensional ultrasound examinations showed a monochorionic diamniotic (MCDA) omphalopagus conjoined twin (Figure 1A). Fetus 1 showed a normal intracranial anatomy, a non-covered defect of the abdominal wall with the liver out, ectopia cordis and signs of hydrops fetalis. Fetus 2 showed holoprosencephaly. The latter fetus also had a non-covered defect of the abdominal wall, absence of the heart and signs of hydrops. Only two limbs were visualized. Doppler ultrasound examination showed a single umbilical cord. Both fetuses were surrounded by a separate amniotic sac and two yolk sacs were identified. Examination with three-dimensional virtual reality confirmed these findings (Figure 1B). A provisional diagnosis of a monochorionic biamniotic conjoined twin of the heteropagus type was made.

A termination of pregnancy was performed at 13 weeks gestational age by application of misoprostol intravaginally. The fetuses showed to be traumatically separated at the time of the postmortem examination (Figure 1C). The external genitalia of fetus 1 showed to be female and the defect of the abdominal wall was confirmed, but only with the intestines extra-abdominally. Internal examination of fetus 1 showed a normal development of the internal organs compliant for the term of pregnancy, with normal localization of the heart. Fetus 2 had a proboscis, rudimentary upper limbs and a single lower limb with five digits. There was a defect of the abdominal wall. Internal organs could only be identified microscopically. Lungs, kidneys and adrenals were present. The heart could not be identified. Abdominal and pelvic internal organs could not be identified. The external genitalia were ambiguous. Both fetuses had a scoliosis with a prominent long and curved coccyx.

Rapid aneuploidy detection (RAD) of skin biopsies of both fetuses showed discordant sex chromosomes. Fetus 1 showed a normal female result, while fetus 2 showed a monosomy X, fitting the signs of hydrops fetalis on ultrasound examination. SNP array (Infinium_CytoSNP_850K v1.1BeadChip, Illumina, San Diego, USA) confirmed the RAD results, with no signs of mosaicism nor additional pathogenic copy number variants in both

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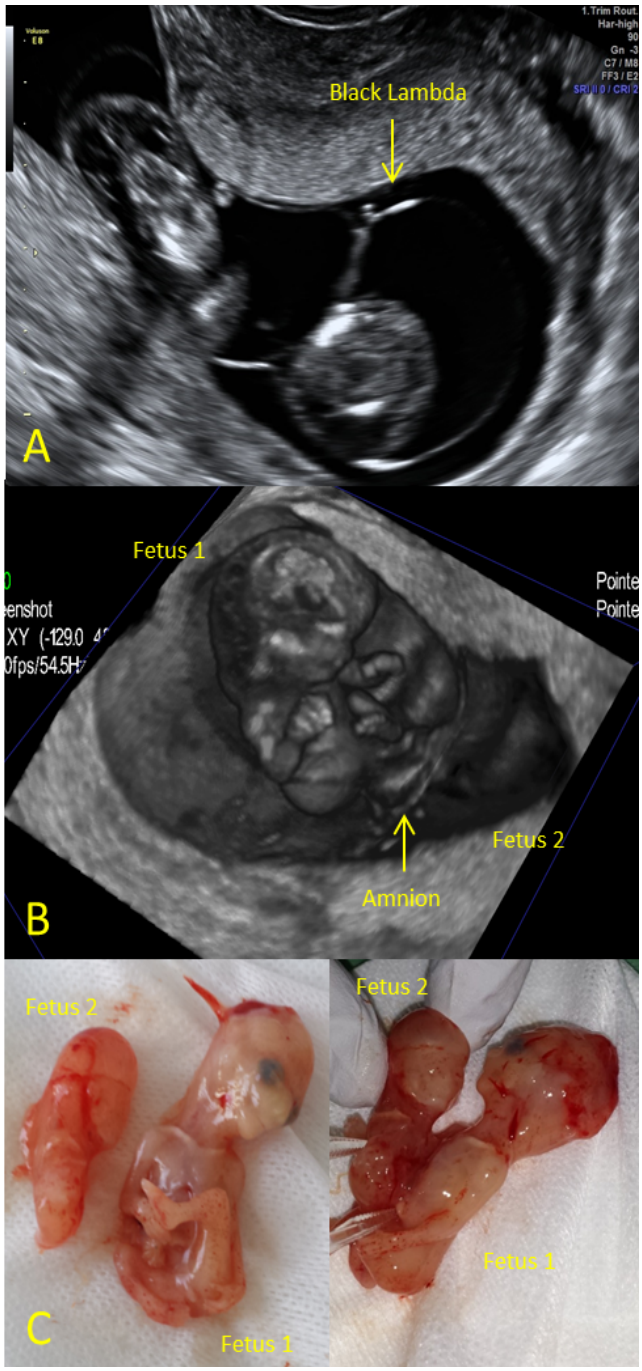

Figure 1: Biamniotic parasitic conjoined twins A) Ultrasound examination: Two amniotic sacs surrounding the conjoined twins, with the occurrence of a 'black lambda', which indicates the pregnancy to be monochorionic diamniotic (MCDA); B) Screenshot of three-dimensional virtual reality: Showing two amniotic sacs; C) Photo of the postmortem examination of the traumatically separated twins.

biopsies. Zygosity testing (AmpFlSTR® Identifiler® PCR Amplification Kit, Applied Biosystems, Life Technologies, Carlsbad, USA) confirmed monozygosity. Parental karyotypes were normal.

Conclusion

An early diagnosis of conjoined twins with 2D- and 3D-ultrasound scans can be made in the first trimester [5,6]. The incomplete fission theory describes the development of conjoined twins and monochorionic and monoamniotic membranes are considered obligatory. However, rare cases of MCDA conjoined twins were reported before [1,3,7,8], not matching this etiology. Alternative etiologies were published. Destephano, et al. [3], described a theory based on the incomplete fission theory. He hypothesized that initially one embryonic disc is situated in the amniotic cavity, followed by an abnormal gastrulation process, resulting in the formation of two primitive streaks instead of one in the embryonic disc. In between the two primitive streaks dimpling and folding of the amniotic sac causes the formation of two separate amniotic cavities and both embryos remain connected omphalopagus [3,9]. As an alternative for the incomplete fission theory a fusion theory was formulated by Spencer, et al. [10]. The latter theory is based on the idea that two separate embryonic discs exist in two different amniotic sacs from early on. These two embryonic discs fuse during development forming the diamniotic conjoined twin [10,11]. The non-mosaic chromosomal discordance would indicate an early separation in the postzygotic stage and supports the fusion theory in our case. Discordant karyotypes in monozygotic twins have been described before [12-15]. Gilbert, et al. and Gou, et al. described cases of monozygotic twins discordant for Turner syndrome and reviewed the known cases with this phenomenon, which is very rare and the prevalence remains unknown [12,13]. Rock, et al. described a case of monochorionic twin pregnancy, discordant anomalies an with a deletion on chromosome 7 in one of the fetuses, likely resulted from a postzygotic mitotic error [14]. Tachon, et al. described monozygotic twins with mosaic 47, XXY/46XX, but with different phenotype and discordant proportions for the 47, XXY and 46, XX cell lines, believed to be a postzygotic origin of the mosaicism [15]. The possible mechanisms of mitotic non-disjunction resulting in heterokaryotypia in monozygotic twins require more evidence. The occurrence of heteropagus parasitic conjoined twins, with signs of dizygosity, based on DNA-extraction, due to fusion was described before by Logrono, et al. [16]. However, these findings could be artefactual or do not indicate dizygosity, but a divergence due to postzygotic mitotic mutations, as described by Machin in a letter to the editor [17]. The occurrence of a biamniotic conjoined twin pregnancy in combination with a discordant genotype was never published so far. This occurrence supports the fusion theory as an etiologic explanation for conjoined twins.

Conflict of Interest

The authors report no conflict of interest.

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