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Case Report

Twin pregnancies discordant for digynic triploidy – A case series

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ABSTRACT

Objective: To analyse natural course and perinatal management in twin pregnancies discordant for digynic triploidy.
Case report: We present five cases of twins discordant for digynic triploidy. Pregnancy outcome was known for three of them. In one case, premature rupture of membranes occurred at 20 gestational weeks and both fetuses were miscarried. In two other pregnancies healthy co-twins were born at term after the triploid fetuses demise at 28 and 37 weeks. No maternal complications were observed.
Conclusion: Twin pregnancies discordant for triploidy poses a challenge for perinatal management. Expectant management should be considered in digynic triploid cases.

Introduction

Triploidy results from an extra haploid chromosome set of paternal (diandric) or maternal (digynic) origin. Majority of triploid conceptuses is miscarried at early developmental stage and its prevalence at 12–16 gestational weeks is only around 0.02% [1].

Regardless of parental origin, triploidy is lethal [2]. Twin pregnancy with triploidy in one of the fetuses poses a challenge for perinatal management due to risk of compromising the unaffected twin. To the best of our knowledge, only three cases of twins discordant for phenotypically digynic triploidy have been reported to date [3–5].

We describe sonographic features and pregnancy outcome in twin pregnancies discordant for digynic triploidy diagnosed in our Department over 22 years.

Case presentation

Between 1997 and 2018 there were five dichorionic twin pregnancies discordant for triploidy evaluated at our institution – all of digynic phenotype according to the classification proposed by McFadden and Kalousek [6]. Ultrasound features, karyotypes and pregnancy outcome are presented in Table 1. All twin pregnancies were tested due to sonographic abnormalities in one of the fetuses. The mean maternal age was 30.7 years. The mean gestational age at referral was 15.8 gestational weeks. Two cases were diagnosed in the first trimester. The mean gestational age at invasive testing was 17.4 weeks. In all affected fetuses triploidy was confirmed by routine karyotyping of amniotic cells. All co-twins had normal karyotypes (see Table 1).

In all cases there was asymmetrical fetal growth restriction (FGR) of the affected fetus (100%) and an evident discordance in growth between healthy fetus and affected co-twin (crown-rump length [CRL] in the first trimester or estimated fetal growth [EFW] after the first trimester) (see Table 1; Fig. 1).

Structural abnormalities were present in three out of five affected twins (60%), however in the remaining two fetuses a detailed structural survey was impossible due to severe FGR. Detected abnormalities are listed in Table 1 and included ventriculomegaly (n = 2), heart defects (n = 2), micrognathia (n = 1) and spleen agenesis (n = 1).

In four cases (80%) there was oligohydramnios in the affected fetus, which was detected at a mean of 21 gestational weeks [range: 11–37 weeks].

The outcome was known for three pregnancies (60%). In one case the pregnancy ended in miscarriage after a premature rupture of membranes of triploid fetus at 20 weeks. In the two remaining cases expectant management with strict ultrasound surveillance every 2–4 weeks was undertaken. In both cases the pregnancy

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Table 1
Characteristics of twin pregnancies discordant for digynic triploidy diagnosed in the Ultrasound Unit of the Department of Gynecologic Oncology and Obstetrics between 1997 and 2018.

No	Gestational age at the detection of fetal defects/at invasive testing	Karyotypes of affected fetus and a healthy co-twin	Asymmetrical FGR in the affected fetus (g. a.)	Growth discrepancy between the affected and healthy co-twin[a]	Oligohydramnios in the affected fetus (g. a. at diagnosis)	Structural defects in the affected fetus	Pregnancy outcome
1	11 weeks/16 weeks	69,XXY 46,XX	Yes (11 weeks)	CRL 18/52 mm (34.6%)	Yes (11 weeks)	Detailed anatomy scan impossible due to severe FGR	PPROM Miscarriage of both fetuses at 20 weeks
2	13 weeks/16 weeks	69,XXX 46,XX	Yes (13 weeks)	CRL 16/61 mm (26.2%)	Yes (18 weeks)	Ventriculomegaly Heart defect	IUFD of triploid fetus at 28 weeks FGR of normal fetus Labour induction at 37 weeks Female live birth 2330 g/45 cm
3	17 weeks/17 weeks	69,XXX 46,XX	Yes (17 weeks)	EFW 103/222 g (46.4%)	No (fetus lost to follow up at 17 gestational weeks)	Ventriculomegaly Trapezoid-shaped head	Unknown
4	18 weeks/18 weeks	69,XXY 46,XX	Yes (18 weeks)	EFW 100/178 g (56.2%)	Yes (18 weeks)	Detailed anatomy scan impossible due to severe FGR	Unknown
5	20 weeks/20 weeks	69,XXX 46,XX	Yes (20 weeks)	EFW 173/351 g (49.3%)	Yes (37 weeks)	Micrognathia Atrioventricular septal defect Spleen agenesis	IUFD of triploid fetus at 37 weeks Caesarean section at 38 weeks Female live birth 3400 g/56 cm

g.a.- gestational age; FGR - fetal growth restriction; CRL – crown – rump length; EFW – estimated fetal weight; PPROM - preterm premature rupture of membranes; IUFD – intrauterine fetal demise.
[a] CRL or EFW of the healthy twin – CRL or EFW of the affected twin/CRL or EFW of the healthy twin (%).

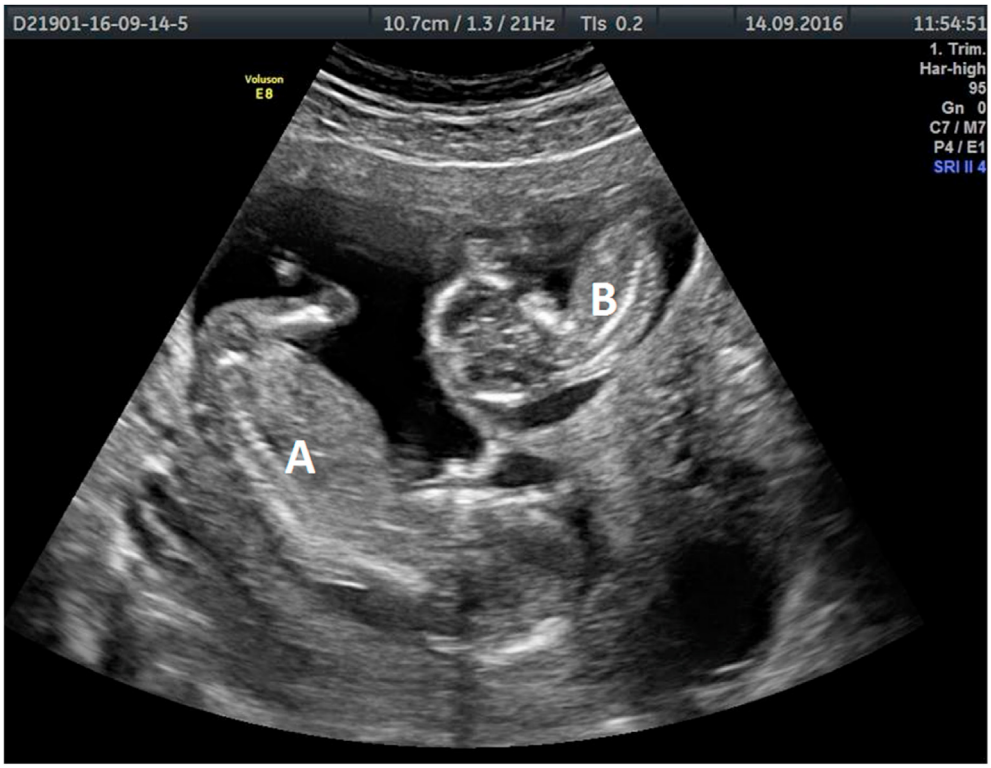

Fig. 1. Discrepancy in crown-rump length between co-twins discordant for digynic triploidy at 13 gestational weeks (A – normal fetus, B – triploid fetus).

ended in a live birth of the healthy co – twin at term after intrauterine demise of the affected fetus at 28 and 37 gestational weeks (see Table 1). In none of the cases maternal complications were observed.

Discussion

During 22 years there were five dichorionic twin pregnancies discordant for triploidy evaluated at our institution. Interestingly,

all of them had digynic phenotype. Triploidy may be associated with a variety of structural anomalies. However, none of them is specific. In singleton pregnancies the most frequent sonographic finding in digynic triploidy is a severe FGR [2]. Similarily in our cohort of twin pregnancies the most frequent sonographic finding in the affected fetus, present in all cases, was asymmetrical FGR, reported as early as in the first trimester of pregnancy. A discrepancy exceeding 15 mm in the crown-rump length between twins is a known marker for chromosomal defects in the smaller fetus [7]. Such a discrepancy was present in both fetuses evaluated in the first trimester in our study as well as in two digynic triploid twins reported in the literature [3,4].

According to the literature the risk of miscarriage in twins is similar for both chorionic villus sampling (CVS) and amniocentesis [8]. However, in our institution we prefer amniocentesis to CVS in twin pregnancies for several reasons. Firstly, amniotic cells derive from different fetal tissues and reflect genuine fetal karyotype. Secondly, amniocentesis limits the risk of cross – sampling of the co-twin. Finally, in digynic triploidy the placenta is usually very small [3], which may be associated with a higher rate of failed CVS.

Noteworthy, array comparative genome hybridization (aCGH), which is nowadays the gold standard in prenatal diagnosis also at our institution, does not detect triploidy. Therefore rapid molecular methods such as quantitative fluorescence-polymerase chain reaction (QF-PCR) or fluorescence in situ hybridization (FISH) should be considered in these cases [2].

Triploidy may have variable intrauterine presentation, but is considered lethal condition [2]. No live birth was observed in our case series, but one of the affected triploid fetuses survived in utero till 37 gestational weeks.

One pregnancy miscarried at 20 gestational weeks due to premature rupture of membranes. As the time between amniocentesis and miscarriage was long (four weeks), the relation between these two events is uncertain.

Selective termination in twin pregnancies is best performed before 18 gestational weeks, performed later it is associated with a higher risk of pregnancy loss and increased risk of preterm delivery [9]. We did not observe any maternal complications after the intrauterine demise of digynic triploid fetuses in our study group. Furthermore, in contrast to diandric cases, digynic triploidy is not associated with an increased risk for maternal complications, such as hypertension, preeclampsia, HELLP syndrome or hyperthyreosis [10]. Therefore, expectant management in these cases may be considered.

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Statement of ethics

All participants signed a written informed consent. Institutional Review Board approval was not required due to the retrospective nature of the study.

Declaration of competing interest

The authors have no conflicts of interest to declare.

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