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Research Letter

First-trimester diagnosis of recurrent omphalocele associated with fetal trisomy 18 but without parental mosaicism

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During the first pregnancy, the patient involved in this study was a 32-year-old, primigravid woman who had been diagnosed with fetal omphalocele by prenatal ultrasound at 12 weeks of gestation (Fig. 1A). The patient's husband was 36 years old. The parents were healthy, and there was no family history of congenital malformations. The patient had not received assisted reproductive technology during this pregnancy. The first pregnancy was subsequently terminated, and a fetus was delivered with isolated omphalocele. Postnatal cytogenetic analysis of chorionic villi obtained by placental sampling revealed a karyotype of 47, XY + 18. Polymorphic DNA marker analysis by quantitative fluorescent polymerase chain reaction (QF-PCR) assays showed a maternal origin of the extra chromosome 18. The patient was pregnant again 1 year later, and a prenatal ultrasound at 12 weeks of gestation revealed recurrent isolated omphalocele (Fig. 1B). Amniocentesis was performed at 13 weeks of gestation, which revealed a karyotype of 47, XX + 18. The mother had a karyotype of 46, XX and the father had a karyotype of 46, XY. The second pregnancy was terminated at 14 weeks of gestation, and a fetus weighing 28 g was delivered with isolated omphalocele. QF-PCR analysis of the placental tissue revealed a maternal origin of the extra chromosome 18.

With the advent of prenatal ultrasound, the early detection of fetal omphalocele is feasible [1]. The peculiar aspect of the present case is the occurrence of recurrent isolated omphalocele associated

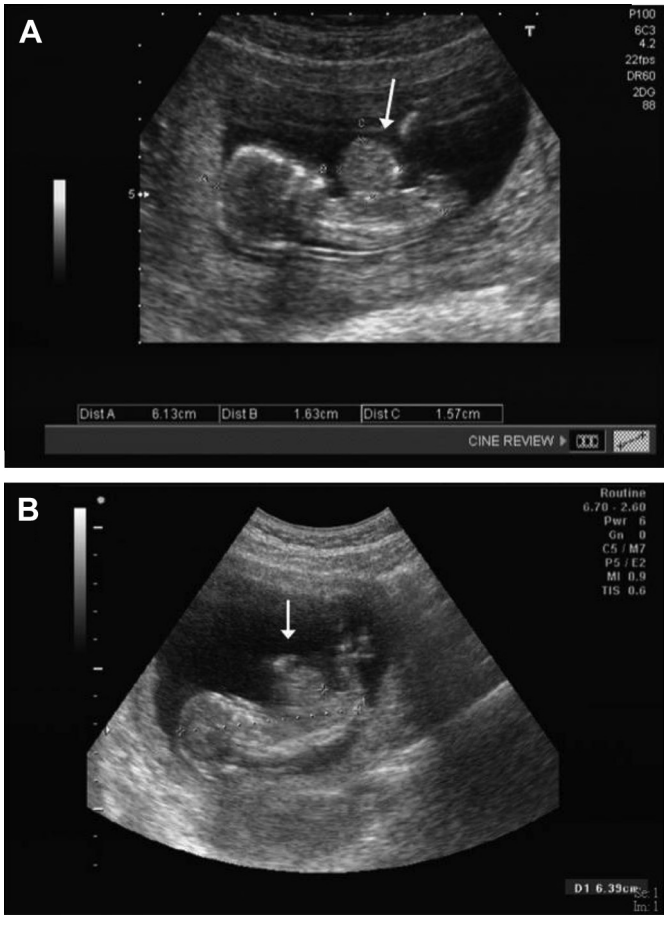

Figure 1. (A) Prenatal ultrasound at 12 weeks of gestation during the first pregnancy shows an isolated omphalocele (arrow); (B) prenatal ultrasound at 12 weeks of gestation during the second pregnancy shows an isolated omphalocele (arrow).

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with trisomy 18 of maternal origin in two consecutive pregnancies. Omphalocele is not uncommon in fetuses associated with trisomy 18. Snijders et al [2] found that omphalocele occurred in 31% of fetuses ($n = 137$) associated with trisomy 18. Chen [3] found that trisomy 18 occurred in 24.1% (277/1148) of fetuses with prenatally detected omphalocele. In a review of the first-trimester sonographic findings in trisomy 18, Sepulveda et al [4] found that the common structural anomalies detected by ultrasound are omphalocele (21%), abnormal posturing of the hands (6%), megacystis (4%), and abnormal four-chamber view of the heart (4%). The present case was associated with a maternal origin of the extra chromosome 18 in two consecutive pregnancies. Various studies have shown that in cases with trisomy 18, only 9% are of paternal origin, and 91% are of maternal origin of which 60% are caused by a meiosis II error, 30% are caused by a meiosis I error, and about 8% are caused by a mitotic error [5–7]. Chen et al [8] reported that 9.7% of cases with trisomy 18 are the result of paternal nondisjunction, whereas 90.3% of cases with trisomy 18 are the result of maternal nondisjunction. The live birth prevalence of trisomy 18 is estimated to be 1:6000 [9,10]. The recurrence risk of a family with a child with complete trisomy 18 is usually reported to be 1% [11]. Prenatal diagnosis of recurrent trisomy 18 in a family should alert the presence of parental mosaicism for trisomy 18 [12–16]. Less than 5% of cases with trisomy 18 are mosaic, and the individuals with mosaic trisomy 18 can be apparently phenotypically normal adults [13–16]. Therefore, prenatal detection of recurrent trisomy 18 should include a cytogenetic investigation of the parents to exclude parental mosaic trisomy 18. This case represents a very rare occurrence of recurrent trisomy 18 with the same structural abnormality without a parental mosaicism.

Conflicts of interest

The authors have no conflicts of interest relevant to this article.

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