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Case Report

Prenatal diagnosis of mosaicism for trisomy 7 in a single colony at amniocentesis in a pregnancy with a favorable outcome

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ABSTRACT

Objective: We present prenatal diagnosis of mosaicism for trisomy 7 in a single colony at amniocentesis with a favorable outcome.**Case report:** A 40-year-old woman underwent amniocentesis at 17 weeks of gestation because of advanced maternal age. Amniocentesis revealed a result of 47,XY,+7[1]/46,XY[26]. In 27 colonies of cultured amniocytes, all five cells in one colony had trisomy 7, while the rest 26 colonies had a normal karyotype. The parental karyotypes were normal. Repeat amniocentesis was performed at 19 weeks of gestation. Interphase fluorescence *in situ* hybridization (FISH) was applied on the uncultured amniocytes, and the result showed trisomy 7 signals in 4% (3/75 cells) of the uncultured amniocytes compared with 1.4% (1/70 cells) in the normal control. Uniparental disomy (UPD) 7 was excluded by polymorphic DNA marker analysis. The cultured amniocytes at repeat amniocentesis had a karyotype of 46,XY. Prenatal ultrasound findings were unremarkable. A healthy 3332-g male baby was delivered at 38 weeks of gestation. The karyotype of cord blood lymphocytes was 46,XY. The boy was phenotypically normal at age 8 months at follow-up. No trisomy 7 signal could be detected in the postnatal FISH analysis of the urinary cells.**Conclusion:** Mosaicism for trisomy 7 in a single colony at amniocentesis without UPD 7 can be associated with a favorable outcome.© 2019 Taiwan Association of Obstetrics & Gynecology. Publishing services by Elsevier B.V. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

Introduction

We previously reported prenatal diagnosis of mosaicism for trisomy 21 in a single colony [1], trisomy 5 in a single colony [2], trisomy 15 in a single colony [3] and mosaicism for trisomy 2 in a single colony [4] at amniocentesis in four different cases with favorable outcomes. Here, we present an additional case of

mosaicism for trisomy 7 in a single colony at amniocentesis with a favorable outcome.

Case report

A 40-year-old, gravida 3, para 2, woman underwent amniocentesis at 17 weeks of gestation because of advanced maternal age. Her husband was 41 years old. The couple had two healthy children, and there was no history of congenital malformations in the family. Amniocentesis revealed a karyotype of 47,XY,+7[1]/46,XY[26]. In 27 colonies of cultured amniocytes, all five cells in one colony had trisomy 7, while the rest 26 colonies had a normal karyotype. The parental karyotypes were normal. Repeat amniocentesis was

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performed at 19 weeks of gestation. Interphase fluorescence *in situ* hybridization (FISH) was applied on the uncultured amniocytes by using the bacterial artificial chromosome probe RP11-1133D5 [7p22.3; spectrum green, fluorescein isothiocyanate (FITC)] and RP11-3L24 (7q11.1; spectrum red, Texas Red). The result showed trisomy 7 signals in 4% (3/75 cells) of the uncultured amniocytes compared with 1.4% (1/70 cells) in the normal control. Polymorphic DNA marker analysis on the DNA extracted from the uncultured amniocytes and parental bloods excluded uniparental disomy (UPD) 7. The cultured amniocytes at repeat amniocentesis had a karyotype of 46,XY in 26/26 colonies. Prenatal ultrasound findings were unremarkable. The parents elected to continue the pregnancy, and a healthy 3332-g male baby was delivered at 38 weeks of gestation. The cord blood lymphocytes had a karyotype of 46,XY in 40/40 cells. The boy was phenotypically normal at age 8 months at follow-up. The interphase FISH analysis on all urinary cells revealed a normal result. No trisomy 7 signal could be detected in the 40/40 urinary cells.

Discussion

Patients with mosaic trisomy 7 may present variable features ranging from normal phenotype to facial dysmorphism, enamel dysplasia, sparse hair, hypomelanosis of Ito, pigmentary abnormalities, radial defects, Potter syndrome, Goldenhar syndrome and Blaschkolinear malformation syndrome [5–16].

The most peculiar aspect of prenatally detected mosaic trisomy 7 by amniocentesis is the possible association with maternal UPD 7 and Silver-Russell syndrome (SRS) (OMIM 180860), which is characterized by prenatal and postnatal growth retardation, a typical triangular face, relative macrocephaly, body asymmetry, fifth finger clinodactyly, micrognathia, a high arched palate, feeding difficulty and neuropsychological developmental delay [17–21]. In a review of 13 cases of mosaic trisomy 7 detected at amniocentesis, Chen et al. [14] found normal phenotype in nine cases and phenotypic abnormalities in four cases (4/13 = 31%) of which two cases were associated with maternal UPD 7 and SRS [17–19]. Flori et al. [18] suggested that patients with SRS and maternal UPD 7 may be resulted from an undetected low-level mosaic trisomy 7. Maternal complete isodisomy 7 can be the result of a post-zygotic mitotic segregation error, whereas maternal heterodisomy 7 can be the result of trisomic rescue after a meiotic non-disjunction event after the first meiotic cell division [22,23]. Prenatal sonographic features of SRS include intrauterine growth restriction, short stature and limb asymmetry [24–26]. Therefore, prenatal diagnosis of mosaic trisomy 7 at amniocentesis should include a careful ultrasound examination of fetal growth, short stature and limb asymmetry, and a molecular genetic investigation to exclude maternal UPD 7.

Mosaic trisomy 7 at amniocentesis has been associated with pseudomosaicism and cytogenetic discrepancy between cultured and uncultured amniocytes [14,15]. The present case shows that prenatal diagnosis of mosaicism for trisomy 7 in a single colony at amniocentesis without UPD 7 can be associated with a favorable outcome, and interphase FISH analysis on uncultured amniocytes is useful for rapid differential diagnosis of true mosaicism from pseudomosaicism at amniocentesis under such a circumstance.

Conflicts of interest

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

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