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Case Report

Prenatal diagnosis and genetic counseling of mosaicism for chromosome t (7; 14) with a favorable outcome

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Summary

We report a case of prenatal diagnosis of mosaicism for chromosome t (7; 14) with a favorable fetal outcome. Similar chromosomal abnormalities have been observed in patients with hematologic malignancy. Chromosomal microarray analysis (CMA) revealed no genomic imbalance, prenatal ultrasound examination revealed no intrauterine growth restriction (IUGR) or dysmorphisms in this fetus. Therefore, combination of karyotype analysis, CMA, genetic counseling and prenatal ultrasound will prove a more specific risk evaluation for chromosomal translocation and mosaicism.

Key words: Karyotype analysis; CMA; Chromosome translocation; Mosaic; Hematologic malignancy; Prenatal diagnosis.

Introduction

There have been previously reported cases of the major chromosome structural aberration t (7; 14) in cells cultured in vitro [1]. Similar chromosomal abnormalities have been observed in patients with hematologic malignancy. Conventional karyotyping can identify chromosome translocation and mosaicism. Chromosomal microarray analysis (CMA) allows for the detection of microdeletions and microduplications that are over 1 kb. Prenatal ultrasound allows for detection of any fetal deformities. Therefore, the combination of karyotype analysis, CMA, genetic counseling and prenatal ultrasound, will prove a more specific risk evaluation for chromosomal translocation and mosaicism. Here, we report prenatal diagnosis and genetic counseling of mosaicism for chromosome t (7; 14) with well pregnancy outcome.

Case report

A thirty-nine-year-old woman (gravida 3, para 1) underwent amniocentesis at 20 weeks of gestation because of the maternal age. She had delivered one healthy child. She had a 42 years old husband, and they had no family history of congenital anomalies. Karyotype analysis of cultured amniocyte showed a karyotype of 46, XY,t(7;14)(q35;q13)[4]/46,XY[26]. The woman's karyotype was 46,XX, and her husband's karyotype was 46,XY. Chromosomal microarray analysis (CMA) on uncultured amniocytes showed no genomic imbalance. Periumbilical blood sampling (PUBS) was performed at 30 weeks of gestation. PUBS showed a karyotype of 46,XY,t(7;14)(q35;q13)[3]/46,XY[97] in 100 cultured lymphocytes (Figure 1). Simultaneous CMA analysis using uncultured cord blood lymphocytes showed no pathogenic microduplication or microdeletion. Prenatal ultrasound revealed no intrauterine growth restriction (IUGR) or dysmorphisms in the fetus. After genetic counseling, the woman and her husband decided to continue the gestation.

At 39 weeks of gestation, a 3500 g male baby was delivered naturally. The baby had normal physical findings at birth. At delivery, karyotype analysis of 100 peripheral blood lymphocytes showed 46, XY. At one year of age, the boy was normal; chromosome karyotype was 46, XY.

Because of the high homology of chromosome 7q and 14q, a frequent structural aberration in the case of rare chromosome aberration in cells cultured in vitro is t (7; 14) [2], and it is difficult to determine whether the translocation derives from laboratory culture or the fetus is a real mosaic.

The term "mosaic" indicates that some cells contain the normal chromosome, whereas others have the derivative chromosome t(7; 14). This case reveals that the mosaic percentage may change after long-term cultures in cord blood and amniotic fluid [3].

Karyotype analysis on the cultured amniocytes showed 13.3% (4/30 cells) mosaicism for derivative chromosome t(7; 14). CMA analysis using uncultured amniocytes showed no pathogenic microduplication or microdeletion.

This suggests some methods using uncultured amniocytes or cord blood lymphocytes, such as CMA analysis or FISH (fluorescence in situ hybridization) can be considered an operative technology for rapid and accurate confirmation of the presence of microduplication and microdeletion [4, 5].

Similar chromosomal abnormalities have been observed

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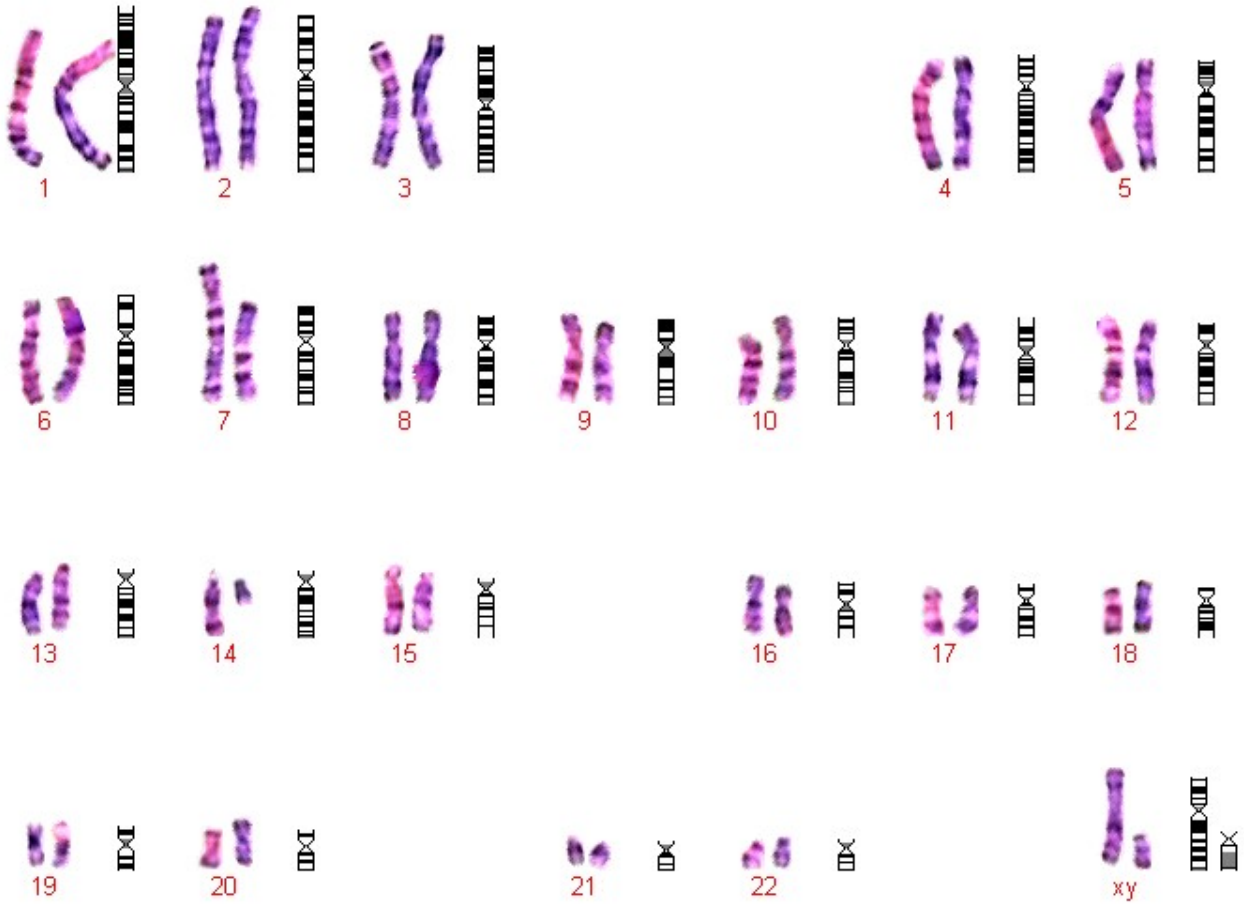

Figure 1. — The karyotype of 46,XY,t(7; 14)(q35; q13).

in patients with hematologic malignancy. One study identified the partner chromosome as a t(7; 14) (q35; q13)with the breakpoint upstream of EGFR [6]. This mutation leads to over-expression of the target oncogene, EGFR. Our patient was evaluated by the oncology team to rule out the possibility of hematologic malignancy.

This case provides an example of how a combination of karyotype analysis, CMA, genetic counseling and prenatal ultrasound examination can provide a more specific risk evaluation for chromosomal translocation and mosaicism.

Conflict of Interest

The authors declare no competing interests.

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