FAMILIAL PERICENTRIC INVERSION INCIDENTALLY DETECTED AT PRENATAL DIAGNOSIS

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Summary A case of familial heterozygous pericentric inversion of chromosome 1 [inv(1)(p13q23)] is presented. The inversion was incidentally detected in a fetus whose mother received prenatal chromosomal diagnosis due to her age (40 years old), and thereafter the same inversion was detected in the father whose phenotype was normal. No abnormalities were found in the phenotype of the newborn carrier. Semen analysis of the father revealed normal findings. The couple had no history of spontaneous abortion.

Key Words pericentric inversion, chromosome 1, prenatal diagnosis, semen analysis

Pericentric inversion of chromosome 1 is an extremely rare chromosomal rearrangement of which only 16 cases have been reported. Herein, we report a new case of a Japanese family having heterozygous pericentric inversion of chromosome 1, the breakpoints of which were p13 and q23. This is the first case in which pericentric inversion of chromosome 1 was prenatally diagnosed.

Case Report

A woman visited our clinic at Tohoku University Hospital for prenatal chromosomal diagnosis of her fetus because of her advanced age (40 years old). It was her first pregnancy. Phenotypes of her and her husband were normal.

Amniocentesis was performed at the 16th gestational week and the amniocytes were cultured in Chang’s media under 5% CO₂ in air. The karyotype revealed the fetus to be a heterozygote with an pericentric inversion of chromosome 1 [46, XY,inv(1)(p13q23)] (Fig. 1). Based on the result, chromosomal examinations of the parents were consequently performed. Fetal blood cell karyotyping was carried out to deny any artifacts. Leukocytes of those samples were cultured in RPMI.
Fig. 1. High resolution banding of fetal chromosome 1. The inverted chromosome is shown on the right. The inverted segment is almost 20% of the chromosome length. Idiograms are shown to indicate the breakpoints, p13q23.

1640 containing phytohemagglutinin under 5% CO2 in air. The parental karyotypes revealed the father to be a carrier of the same inversion. Based on the facts that ultrasonography revealed no major malformation in the fetus, that the inversion was familial, and that the inverted segment was small (less than 20%), we concluded that the inversion had no adverse effect on the phenotype of the fetus. Therefore, the parents decided to continue the pregnancy.

The mother spent an uneventful pregnancy until the 39th gestational week, but ultrasonographic examination revealed that amniotic fluid to be high-echoic at the time of the onset of labor. The biophysical condition of the fetus was normal. Amniotic fluid obtained at amniotomy seemed to be chyle and its color was white. Microscopical observation revealed that the fluid contained many squamous cells, which probably originated in the vernix caseosa. Despite the abnormal amniotic fluid, she gave birth to a healthy male newborn after spontaneous labor in the 39th gestational week. The birth weight was 3,856 g, the body length 53 cm, the head circumference 35.5 cm, and the chest circumference 31 cm. The newborn had neither asphyxia nor malformations. Macroscopic observation of the placenta and the fetal membrane revealed no abnormal findings. The newborn was also normal in posture, appetite, and bowel movement, and results of the neurological examination and the congenital metabolic error screening were normal. The newborn grew without any clinical problems thereafter.

The father’s parents, who had already died, had had no history of repeated spontaneous abortions. The father’s four brothers are phenotypically normal and their spouses have not suffered from habitual abortions. Moreover, results