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Genetic counseling on amniocyte level II mosaicism

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A 36-year-old woman who was a junior college graduate and a housewife received amniocentesis because of her advanced maternal age. Giemsa banding revealed the karyotype of the amniocytes was 46, XY. More than two cells with 46, XY, t (1; 2) (p21; q22) in one colony, however, were found among the 15 colonies analyzed (Fig. 1). Mosaicism level II was diagnosed.

Genetic counseling was arranged because of the parents' concerns. The counseling process included information on chromosomes and karyotypes. Mosaicism is the coexistance of a normal and abnormal karyotype. Mosaicism detected prenatally is categorized into three levels. Level I is when a single abnormal cell is found in one colony, and is considered an artifact of the culture. If more than two cells with the same chromosome abnormality are found in one colony, or the same abnormality is found in more than two colonies from one culture, and the abnormality is not seen in colonies from other independent cultures, it is considered level II mosaicism (Fig. 1). These cases are usually found to be pseudomosaicism, and additional studies may be performed. Level III mosaicism is defined as the detection of two or more cells with the same chromosome abnormality distributed over two or more independent cultures. Those cases are likely to be true mosaicism.

According to the standard procedures in the cytogenetic laboratory, additional studies were not needed to eliminate the possibility of chromosomal abnormality in our case. The parents were counseled that major malformations occur in 3-4% of live births. Prenatal genetic counseling was also recommended for the next pregnancy.

The follow-up of this pregnancy was uneventful, and the baby was born at full term with a birth body weight of 2870 gm. The perinatal course was smooth and repeat blood chromosome testing revealed a karyotype of 46, XY (20 cells).

Further reading

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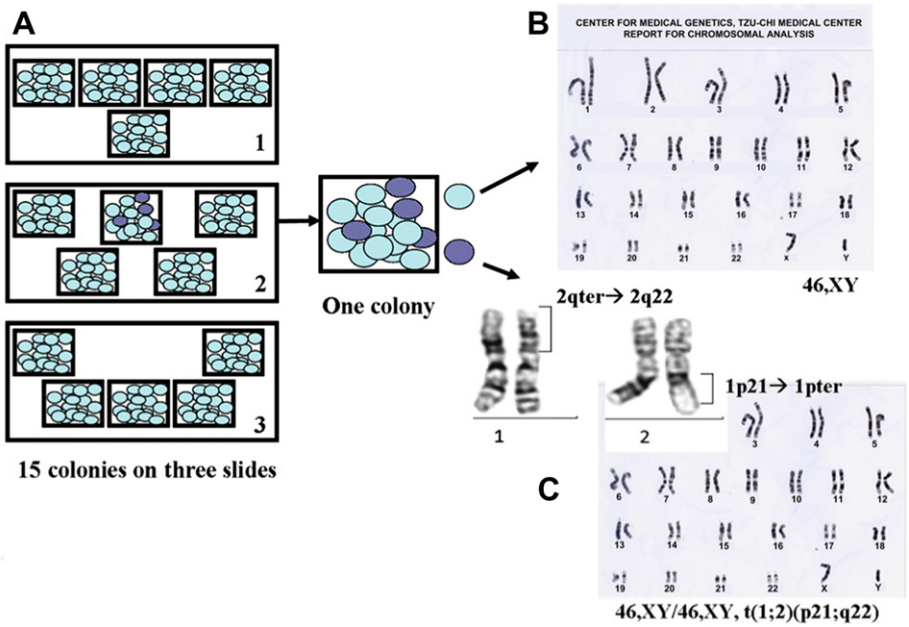

Fig. 1. (A) There are more than two cells with the same abnormality in one colony among the 15 colonies analyzed on culture slide number 2. (B) The karyotype of the green cells is 46, XY. (C) The karyotype of the lined cells is 46, XY/46, XY, t (1; 2) (p21; q22). No other abnormal cells can be seen in colonies from slide 1 and slide 3.