CASE REPORTS

A case of 46, XY/47, XYY mosaicism

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Abstract

In some clinical features, the failure of the couple to reproduce may be related to the chromosome abnormality of the patient. This report was based on one example about the topic.

Key Words: Chromosome, Chimera, Abnormal gene

1 Medical record

1.1 General information

A male patient, 39 years old, married, 179 cm, 77 kg, was well-developed with no abnormalities in phenotype and intelligence. Because his wife had not been conceiving a child naturally since they got married 10 years ago, she was given assisted reproduction in other hospital: for the first time, the embryo implanted by means of intracytoplasmic sperm injection did not survive; at the second time, his wife received IVF-ET with the survival rate of 50% (2 living embryo of 4 human embryos), but she got abortion accidentally after 40 days. The patient's wife was healthy with no abnormalities shown in the physical examination. Both husband and wife denied the exposure to radioactive and toxic substances. Pedigree study was as follows: the patient's parents showed normal phenotype, had a son and a daughter; the patient's sister showed normal intelligence and phenotype, had a son with normal intelligence.

1.2 Laboratory examinations

Laboratory examinations for the patient were as follows: routine semen analysis in 2013: sperm survival rate 46% (normal reference range $\geq$ 50%), grade A sperm 11.1% (normal reference range $\geq$ 25%), grade A + grade B sperm 33.4% (normal reference range $\geq$ 50%). Other test items showed no abnormalities. After treatment (treatment condition unknown), the results of routine semen analysis in 2014 were as follows: sperm survival rate 63%, grade A sperm 23.2%, grade A + grade B sperm 66.7%.

1.3 Cytogenetic examinations

Cytogenetic examinations were as follows: the couples were both taken peripheral blood to make a routine preparation of chromosomes for G-banding analysis with 100 metaphase cells counted. 8 cells were given karyotype analysis, the results of which were confirmed by C-banding analysis (see Figure 1), the patient's karyotype was 46, XY, Yqh + [93]/47, XYY, Yqh + [7] (see Figure 2), his wife's karyotype was normal.

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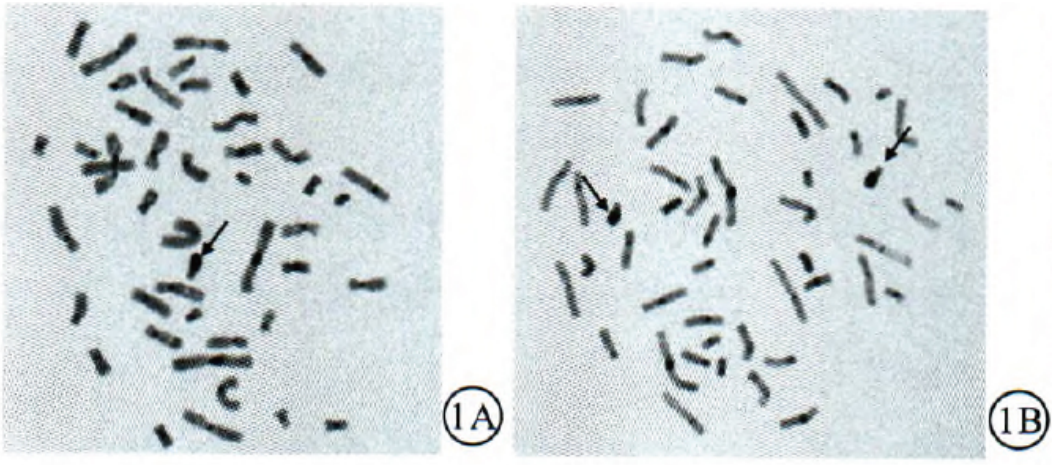

Figure 1: Patient's C-banding Karyotype; 1A: the arrow shows the Y chromosome; 1B: the arrow shows 2 Y chromosome

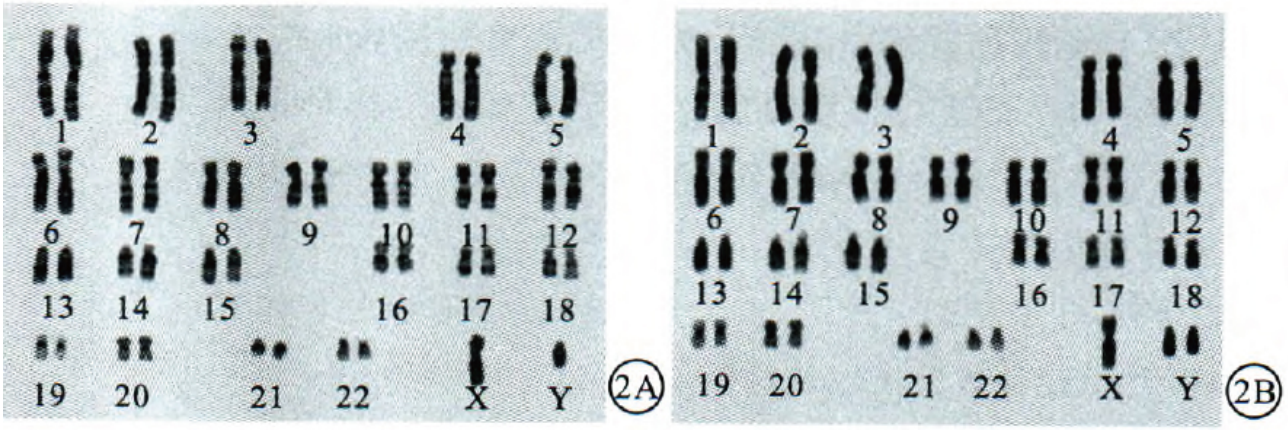

Figure 2: Patient's G-banding Karyotype; 2A: 46, XY; 2B: 47, XYY

2 Discussion

In the mosaic numerical abnormalities of Y chromosomes, it is rare to see 47, XYY mosaicism in a low proportion. It is probable that Y chromosomes in part of cells in the embryonic cleavage stage, do not segregate and form cell lines of 47, XYY and 45, X. In the development of embryo, cell lines of 45, X are dead and then 46, XY/47, XYY mosaicism is formed. Different proportions of tissue mosaicism may exist due to histological differentiation. The patient was of great stature, with asthenospermia as well as part of clinical features of 47, XYY syndrome. The reason why the couple failed to give birth may be related to chromosome abnormalities shown in this patient.

Conflicts of Interest Disclosure

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

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