SHORT COMMUNICATION

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Turner Syndrome With Pseudodicentric Y Chromosome Mosaicism

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The objective was to compare the impact of gonadal cell line upon the phenotype of a Turner syndrome patient with mosaic karyotypes. A 10-year-old female presented with typical Turner syndrome. Chromosomal analysis of lymphocytes revealed 45,X (16%)/46,X,pseudodicentric Y (p ter→q12::q12→p ter) (84%). Karyotype of the gonads revealed 45,X (85%)/46,X,pseudodicentric Y (p ter→q12::q12→p ter) (15%). Discrepancy of the individual cell lines between the lymphocytes and the tissue might exist. The phenotype of patients with mosaicism is related with the individual proportions of gonadal cell lines.

KEY WORDS: Fluorescence in situ hybridization; mosaicism; pseudodicentric Y chromosome; Turner syndrome.

INTRODUCTION

Sex-chromosome mosaicism is frequent in Turner's syndrome. About 50% of patients have Turner's syndrome combined with abnormal structure of X or Y chromosome (1). Pseudodicentric Y chromosome is the result of a single break in the pericentric region followed by chromatid duplication and the joining of chromatids from one side of the break (2). Although numerous cases with pseudodicentric Y chromosome mosaicism have been reported (3–5), scanty reports about the gonad karyotype have been reported. The karyotype of gonad tissue is not yet performed routinely. In this report, we first demonstrated the difference of individual cell lines between the lymphocytes and the gonads. We hypothesized that the female phenotype may be due to the prominent 45,X in gonads.

CASE

A 10-year-old girl presented with thin (25 Kg, 5th–10th percentile) and short stature (116 cm, <3rd percentile), broad chest, widely spaced and hypoplastic nipples, prominent anomalous auricles, slight cubitus valgus, hyperconvex nails, retarded bone age (8-year-old), normal motor and mental development, and female genitalia. She was born after an uncomplicated term pregnancy. Chromosomal analysis of lymphocytes revealed a mosaic karyotype with 45,X (16%)/46,X,pseudodicentric Y (p ter→q12::q12→p ter) (84%) (Fig. 1) and ish (DXZ1 × 1, DYZ1 × 1, DYZ3 × 2, DYZ4 × 2, DYZ5 × 2, DAZ × 2) de novo after fluorescence in situ hybridization (FISH) analysis (Fig. 2). Turner syndrome with a pseudodicentric Y chromosome mosaicism was impressed.

Laparoscopic exploration revealed an infantile uterus and bilateral streak gonads. Salpingo-oophorectomy was performed. Pathology of the gonads revealed fibrous ovarian stroma without evidence of gonadoblastoma. Karyotyping of the gonads revealed 45,X (85%)/46,X,pseudodicentric Y (p ter→q12::q12→p ter) (15%).

DISCUSSION

The 45,X/46,XY mosaicism presents many phenotypes, including normal male to phenotypic female (6,7). Although numerous cases with pseudodicentric Y chromosome mosaicism have been reported, their biological basis of sex development and differentiation remains unclear (4). Sexual development depends on the structure of the pseudodicentric Y chromosome and the level and distribution of mosaicism present. It has been proposed that the 45,X cell line possess more apparent influence upon the phenotype than does the Y chromosome (3).

SRY gene (sex-determining region in Yp chromosome) is essential for testicular development (5). If Yp chromosome has the breakpoint, the SRY gene may be impaired. The altered structure, position, and function of SRY may interrupt virilization. Papenhauser *et al.* (8) described a boy with mosaic 45,X/46,XY and asymmetrical leg growth. They detected the higher proportions of 45,X and 46,XY cell lines in the short leg and the normal leg, respectively. It suggested that the proportion of a cell line might determine its development.

In this report, we observed the discrepancy of karyotypes between lymphocytes and gonadal cells in a

Fig. 1. Karyotyping of the 46,X,pseudodicentric Y (p ter→q12::q12→p ter).

Fig. 2. Fluorescence in situ hybridization (FISH) analysis of the pseudodicentric Y chromosome (DXZ1 × 1, DYZ1 × 1, DYZ3 × 2, DYZ4 × 2, DYZ5 × 2, DAZ × 2).

woman with mosaic Turner's syndrome. The breakpoint in Yq chromosome preserved the intact SRY. Therefore, the lack of virilization may be due to her higher proportion of gonadal 45,X cell line.

In conclusion, the discrepancy in the individual cell lines between the lymphocytes and the tissue existed and deserved further survey. The phenotype of patients with mosaicism is related with the individual proportions of gonadal cell lines.

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