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CASE REPORT

A Case of Chimeric Turner Syndrome with Normal Reproductive Function

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SUMMARY

Background: **Turner syndrome (TS) is a common sex chromosome disorder with the highest incidence among chromosomal abnormalities. Most of the patients showed short stature, small uterus, ovarian atrophy with a stringy shape, external genital dysplasia, primary amenorrhea, infertility, breast agenesis, and other symptoms which are important causes of female infertility.**
Methods: **Peripheral blood lymphocytes were cultured with 1,640 medium for 72 hours. The chromosome karyotypes were counted and analyzed after hypotonic operation, fixation, drop operation, and G-banding operation.**
Results: **The peripheral blood chromosome karyotype of the pregnant woman was 45,X,9qh+[25]/46,XX,9qh+[75]. The case was a patient with chimeric TS, and her chromosome 9 was polymorphic.**
Conclusions: **The clinical phenotype of patients with chimeric TS cannot be determined solely by chromosome karyotype. The influences of somatic mosaics and X chromosome inactivation and other factors on the clinical phenotype should be considered. This study enriched the theoretical basis for prenatal diagnosis and genetic counseling of chimeric TS.**
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KEYWORDS

turner syndrome, chimera, reproductive function

INTRODUCTION

Turner syndrome (TS), also known as congenital ovarian hypoplasia, is a female sex chromosome disorder caused by the complete or partial absence of an X chromosome in all or some of the body's cells, or other structural abnormalities of the X chromosome. The clinical characteristics of TS patients are growth disorders, abnormal reproductive system development, autoimmune system diseases or cardiovascular system abnormalities, and even the occurrences of tumors [1,2]. In different ethnic groups, the prevalence of TS is about 1/2,500 [3]. About 45 - 50% of patients with TS are 45,X haplotypes, and the rest are structural chromosomal abnormalities or chimerism. An individual with two or more karyotypes in the same body is called a chimera. This study described the karyotype and clini-

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cal manifestations of a patient with chimeric TS in order to provide assistance for prenatal diagnosis and genetic counseling of fetuses with chimeric TS.

SUBJECT AND METHODS

Clinical case

The patient was 33 years old and her height was 158 centimeters. She had her menarche at the age of 13. Her menstrual cycle was 35 days and menstrual period lasted 6 - 7 days. She had no abnormal clinical phenotype and abortion history. The chromosome of her husband's peripheral blood was normal. In March 2018, her daughter was born at full term and the girl's growth was normal. The present pregnancy is a healthy male fetus.

Research methods

Peripheral blood was collected in a heparin anticoagulant tube and inoculated into cell culture medium under sterile conditions. The cell culture bottle was placed in a 37℃ incubator for 72 hours, then 3 drops of colchicine were added and culture was continued for 2 hours before collecting cells. After hypotonic operation, fixation, drop operation, and G-banding operation, a Leica automatic analysis system was used to analyze the karyotypes of metaphase dividing cells. The karyotypes were described according to the International System for Human Cytogenetic Nomenclature (ISCN2020). Twenty metaphase dividing cells were counted and 5 karyotypes were analyzed in conventional specimens. But the numbers of karyotype counts and analyses were increased in chimeric specimens.

RESULTS

One hundred metaphase divisions of the patient were counted, in which 25 were X haplotypes. Chromosome 9 of the patient showed polymorphic variation. The patient's peripheral blood chromosome karyotype was 45,X,9qh+[25]/46,XX,9qh+[75]. She was a patient with chimeric TS.

DISCUSSION

Most of the patients with chimeric TS have the characteristics of TS, including growth retardation and dysplasia of the reproductive system and so on. The patients may suffer from uterine and ovarian hypoplasia or aplasia, and most of the symptoms will affect fertility leading to infertility [4]. The pathogenesis of 45,X/46,XX is not fully understood at present, and the 45,X cell line in the chromosome karyotype may be generated due to the instability of X chromosome with abnormal structure and replication errors during mitosis. This type of karyotype has normal cell lines, so the physical clinical characteristics of patients vary with the proportion of normal cell lines in the chromosomal karyotype [5]. However, the clinical phenotype cannot be determined solely by chromosome karyotype. The influence of somatic mosaics, X chromosome inactivation and other factors on the clinical phenotype should be considered [6,7]. For example, one study had shown that a TS patient with 25% X monomer chimerism had a clinical phenotype of hypoplasia of the primordium uterus, cord ovary, external genitalia and breast dysplasia, and short stature. However, the chimerism ratio of X monomer in this study was also 25% and the patient had no clinical phenotype of TS and even had spontaneous fertility. Through the comparison of the two patients, it is suggested that the clinical symptoms cannot be judged solely by chromosome karyotype in genetic counseling. It was reported that there were only 2 - 10% of women with TS, who most commonly were 45,X/46,XX TS, had spontaneous pregnancy ability. But these pregnant women had a higher risk of adverse pregnancy and spontaneous abortion [8,9]. Although the case in this study had spontaneous pregnancy and smooth delivery experience, the proper assessment and management of her complications should be strengthened. The effective application of a multidisciplinary approach and the promotion of appropriate medical education will help the patient to carry out the pregnancy and deliver smoothly.

Declaration of Interest:

All authors declare that they have no competing interests.

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