

一例45, X/46,XY嵌合型Turner综合征的临床和细胞遗传学研究

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摘要 本文报道了一例45, X/46, XY嵌合型Turner综合征的病例, 并进行了外周血淋巴细胞、两侧性腺及皮肤成纤维细胞培养及其核型分析。对核型及表型的关系, 肿瘤的发生率以及45, X/46, XY嵌合体患者的性腺发育和分化, 进行了讨论。

关键词

分类号

Clinica and Cytogenetic Studies on a Turner's Syndrome with 45,X/46,XY Mosaicism

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Abstract

This paper report a case of Turner's syndrome with 45,X/46,XY mosaicism in peripheral blood lymphocyte. In order to prevent gonadal neoplasm, laparotomy was performed and both gonads were removed. Cultures of the skin and both gonads were carried out. The karyotype for skin was 45,X/46,XY, but for the gonads, 45,X. Pathological examination of the gonads showed ovarian interstitial tissue on the side and presence of ovarian follicles on the other side. No testicular tissues were noted. According to the clinical and cytogenetic investigation of our patient and those reported in the literature. There is the indicating that Y chromosome in the peripheral lymphocytes does not affect the differentiation of the testes. Differentiation of the gonads is directly related to the karyotype in the gonads itself.

Key words

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