

## 14例闭经患者的细胞遗传研究

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收稿日期 修回日期 网络版发布日期 接受日期

摘要 运用数种细胞遗传技术研究了14例闭经患者的病因,发现11例(占78.6%)具有异常核型。包括45,X(3例); 45,X/46,X,i(Xq)(2例); 45,X/46,XX/47,XXX(1例); 45,X”46,X, idic(X)(1例); 46,X, idic(X)(1例); 46,X, del(X)(q24)(1例); 45,X/46,X, del(X)(p11.3)(1例)和46,XY(1例)。其中6例(54.5%)涉及X染色体结构异常。对闭经患者的细胞遗传病因、核型与表型之间的可能关系以及失活X染色体的迟复制区段进行了讨论。

关键词

分类号

## Cytogenetic Studies on 14 Cases wi Amenorrhea

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### Abstract

Using several cytogenetic technics,the etiology of 14 cases with amenorrhea was studied.The abonormal karyotypes were found in 11 cases (78.6% cases studied),inc luding 45,X (3 cases);45,X/46,X,i (Xq) (2 cases);45,X/46,XX/47,XXX (1 case);45,X /46,X,idic (X) (1 case);46,X,idic (X) (1 case);46,X del (X) (q24) (1 case);45,X/ 46,X,idic (X) (p 11.3) (1 case) and 46,XY (1 case).54.5% of these cases involve the structural rearrangement of X chromosome.The cytogenetic mechanism of the pa tients with amenorrhea,the possible relationship between karyotype and phenotype ,and the pattern of the replicate region in inactivated X chromosomes were discu ssed.

### Key words

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