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# 一例带有两条9号染色体次缢痕丢失的患者及其家系的细胞遗传学研究

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摘要 本文报道了一例两条9号染色体次缢痕缺失的女性病例，临床表现为严重智力低下和语言障碍。经外周血淋巴细胞染色体G、C显带分析，确定其核型为46，XX，del(9) del(9) (pter→q11::q13→qter)。又对其父母及妹妹进行了染色体分析，患者之父与两个妹妹的核型均正常，其母是46，XX，del(9) (pter→q11::q13→qter)核型的携带者。

关键词

分类号

# Studies on Cytogenetics of A Patient with two Chromosomes 9 Secondary Constricti on Deletion and Pedigree

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**Abstract**

This report describes a female case who is complete deletion of secondary constriction on two chromosomes 9.The major clinical phenomenon is severe mental retarda-tion.

The chromosome analysis of her peripheral blood lymphocyto using G banding and C banding techniques shows the karyotype of 46,XX,del (9),del (9) (pter→q11::q13→qter).

Further,the chromosome analysis of her parents and two young sisters are made.The karyotypes of her father and two young sisters were normal.While her mother's karyotype is 46,XX,del (9) (pter→q11::q13→qter).Her mother is a carrier of secondary constriction deletion on one chromosome 9.

The function of secondary constriction on chromosome 9 and relation between the function and the phenotype were discussed.The origin of aberrant chromosomes was analysed.

**Key words**

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