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脆性X综合征的基因诊断与产前诊断

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摘要　为了探讨简便、快速、准确、价廉的脆性X综合征的诊断方法，对6个智能低下家系进行了细胞遗传学检查，以及PCR直接扩增FMR1 5′端(CGG)$_{n<\sub>}$重复序列、RT-PCR扩增FMR1基因的cDNA序列的分子遗传学检查。A家系先证者脆性X染色体高表达（35/273），分子遗传学检查证实为脆性X综合征全突变患者；B家系先证者及其母亲无脆性X染色体表达，分子遗传学检查证实为非脆性X综合征患者；C家系的男性胎儿脆性X染色体表达（5/93），先证者及其母亲未发现脆性X染色体，分子遗传学检查证实男性胎儿为脆性X综合征全突变患者，其母亲为前突变携带者，哥哥为嵌合体患者；D家系先证者脆性X染色体高表达17%，其姐姐脆性X染色体5%，分子遗传学检查证实先证者为脆性X综合征全突变患者，其姐姐为嵌合体患者；E家系先证者及其母亲，F家系先证者发现可疑脆性X染色体，分子遗传学检查证实为非脆性X综合征家系。结论： PCR直接扩增FMR1基因(CGG)$_{n<\sub>}$重复序列联合RT-PCR扩增FMR1基因cDNA 序列简便、快速、价廉。可用于脆性X综合征的筛查、诊断及产前诊断，有推广应用价值。

关键词　脆性X综合征　FMR1基因　PCR　RT-PCR　基因诊断　产前诊断

分类号

Genetic Diagnosis and Prenatal Genetic Diagnosis of Fragile X Syndrome

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Abstract

In order to obtain a simple,fast,accurate and low-cost diagnosis method of fragile X syndrome,cytogenetic tests and molecular genetic tests were carried out with direct amplification of (CGG)<sub>n<\sub> repeat sequence in 5[']terminal of FMR1 gene by PCR and the cDNA sequence of FMR1 by RT-PCR from six mental retardation pedigrees.The proband of pedigree A with highexpression of fragile X chromosome(35/273) was detected to be a full mutation patient of fragile X syndrome by the molecular genetic test.There is no expression of fragile X chromosome in the proband and his mother of pedigree B,which was futher confirmed as a non-fragile X pedigree by the molecular genetic test.A male foetus of the pedigree C has fragile X chromosome(5/93),but the proband and his mother has no fragile X chromosome.By further detection using molecular genetic test,the male foetus is a full mutation patient of fragile X syndrome,his mother is a permutated carrier,and his brother is a mosaic patient.The proband of pedigree D has high expression of fragile X chromosome(17%),his sister also has expression of fragile X chromosome(5%).By further detection with molecular genetic test,the proband is a full mutation patient of fragile X syndrome,and his sister is a mosaic patient.The probands of pedigrees E and F of the mother were found with suspicions fragile X chromosome,being confirmed as the non-fragile X pedigrees by the molecular genetic test.The conclusion is that the analysis test with direct amplification of 5['](CGG)<sub>n<\sub> repeat sequence and cDNA sequence in FMR1 gene is simple,fast,low-cost and can be applied in screening,diagnosis and prenatal diagnosis of fragile X syndrome.

Key words fragile X syndrome　FMR1 gene　PCR　RT-PCR　genetic diagnosis　prenatal diagnosis

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