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视网膜色素变性中GUCA1B、GNGT1和RGS9基因突变筛查

Analysis of GUCA1B,GNGT1 and RGS9 Genes in Patients with Retinitis Pigmentosa

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摘要　为寻找视网膜色素变性的致病基因，从120个家系收集视网膜色素变性先证者，制备基因组DNA。应用PCR－异源双链-SSCP法，分析GUCA1B基因4个外显子、GNGT1基因编码区和RGS9基因视网膜特异性转录区，寻找基因变异。序列分析确定突变。结果表明，31人的GUCA1B基因外显子1存在T/C多态。所有先证者中均未检测到GUCA1B、GNGT1和RGS9基因突变。认为本组病例未发现GUCA1B、GNGT1和RGS9基因的突变。

Abstract:To screen possible disease-causing mutations in the GUCA1B gene, GNGT1 gene, and the alternative-splicing region of RGS9 gene in 120 probands with retinitis pigmentosa, genomic DNA was collected from 120 probands with retinitis pigmentosa out of 120 families. The coding sequences of the GUCA1B and GNGT1 genes and the alternative splicing region of the RGS9 gene were analyzed by using PCR-heteroduplex-SSCP method. Mutation was confirmed by DNA sequencing. A T/C polymorphism was identified in exon 1 of the GUCA1B gene in 31 of the 120 probands. Heteroduplex-SSCP analysis of the GUCA1B and GNGT1 coding regions and RGS9 alternative splicing region showed no mutations in 120 patients with retinitis pigmentosa. We found no evidence that mutation in GUCA1B, GNGT1, or RGS9 gene is a cause of retinitis pigmentosa.

关键词　视网膜色素变性　GUCA1B　GNGT1　RGS9　突变　多态　序列　Key words　retinitis pigmentosa　GUCA1B　GNGT1　RGS9　mutation　polymorphism　sequence

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