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研究报告

在两个X连锁显性腓骨肌萎缩症家系中发现同一GJB1基因突变Glu208Lys

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摘要

在两个X连锁显性腓骨肌萎缩症(Charcot-Marie-Tooth disease, CMT) 家系中进行了*GJB1*基因的突变分析。提取基因组DNA, PCR(polymerase chain reaction)反应扩增GJB1基因编码序列, 进行单链构象多态性(single strand conformational polymorphism, SSCP)分析, 对有差异SSCP带型的PCR产物进行测序, 结果在两家系中发现同一*GJB1*基因c.622G→A (Glu208Lys)突变。所发现的突变位点在国内尚未报道。

关键词 腓骨肌萎缩症 X-连锁 GJB1基因 突变

分类号

The same mutation Glu208Lys in the GJB1 gene was detected in 2 families with X-linked Charcot-Marie-Tooth disease

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Abstract

<P>Mutation of <EM>GJB1</EM> gene was investigated in two families with X-linked Charcot-Marie-Tooth disease. Genomic DNA from venous blood samples was prepared. The coding sequence of the <EM>GJB1</EM> gene was amplified from genomic DNA. PCR products were analyzed by single strand conformational poly-morphism (SSCP) method. The PCR product having an abnormal pattern was sequenced to detect the mu-tation. It was found that the samples of all patients and one little girl with normal phenotype showed an abnormal SSCP band, but not detected in the other unaffected members in the first large family. In the second small family, an abnormal SSCP band was found in all the patients, but not detected in the unaf-fected member. The result of DNA sequencing demonstrated that both families had a same mutation of 622G→A, which resulted in a substitution of Glu208Lys. This mutation has not been reported previously in China.</P>

Key words Charcot-Marie-Tooth disease X-linked GJB1 gene mutation

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