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论著

多重连接依赖式探针扩增和变性高效液相色谱法检测Duchenne型肌营养不良症患者DMD基因的缺失/重复突变

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摘要 摘要：目的 比较多重连接依赖式探针扩增法（MLPA）和变性高效液相色谱法(DHPLC)检测Duchenne型肌营养不良症（DMD）患者DMD基因缺失/重复突变的效果。方法 选择2004年10月~2005年10月在我院确诊的22位无关DMD男性患者,采用MLPA法对经DHPLC技术检测过的患者的DMD基因的缺失/重复突变进行突变筛查,同时对先证者的23位女性亲属进行基因的缺失/重复突变检测。结果 DHPLC技术和MLPA法均检测出11位先证者具有DMD基因缺失突变,3位先证者具有DMD重复突变;MLPA法除能更精确地检测出上述突变外,还检测出DHPLC法未检测出的两位患者的DMD基因存在缺失突变。16个家系中18位可能的女性携带者中,12位经检测为缺失/重复突变携带者。两种方法均未检测到6位先证者及其女性亲属DMD基因具有缺失/重复突变。结论 与DHPLC法和传统的多重PCR方法相比,MLPA法检测DMD基因的缺失/重复突变位置更为准确。MLPA法可用于检测先证者及家系中女性携带者DMD基因的缺失/重复突变。

关键词 Duchenne肌营养不良症 多重连接依赖式探针扩增法 变性高效液相色谱法 DMD基因缺失/重复突变

分类号

Identification of Deletion/duplication Mutations in DMD Gene by Multiple Ligation Probe Amplification and Denaturing High-performance Liquid Chromatography

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Abstract ABSTRACT:Objective To compare the effectiveness of using multiple ligation probe amplification (MLPA) and denaturing high-performance liquid chromatography (DHPLC) in screening the exon deletions and duplications of the DMD gene. Methods MLPA technique was applied to detect exon deletions and duplications previously confirmed by denaturing high-performance liquid chromatography (DHPLC). Results From October 2004 to October 2005, 22 unrelated DMD probands and their possible female relatives with clinical diagnosis with dystrophinopathy at our hospital entered this study. Both DHPLC and MPLA detected DMD gene depltions in 11 probands and DMD duplications in 3 probands. MLPA detected deletions and duplications in 2 probands, which were not detected by DHPLC. MLPA also successfully identified the carriage status of the potential female carriers of the probands. Conclusion Compared with DHPLC and traditional PCR techniques, MLPA is a superior tool to analyze the deletions and duplications in affected males as well as in the identification of the carriage status of potential females carriers.

Key words Duchenne muscular dystrophy multiple ligation probe amplification denaturing high

performance liquid chromatography deletion/duplication mutation

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