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论著

兄妹同患假肥大型肌营养不良症

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摘要 摘要：目的 探讨同患假肥大型肌营养不良症（DMD）兄妹的临床以及实验室检查特点。方法 对患者进行临床观察、血清酶、肌电图、心电图及心脏彩色超声检查、肌肉病理HE染色, 免疫组织化学染色检测肌肉组织抗肌萎缩蛋白、utrophin的表达, 用多重连接探针扩增法对抗肌萎缩蛋白基因1～79号外显子进行缺失和/或重复突变检测, 利用抗肌萎缩蛋白基因的CA短串联重复序列（STR）对该家系进行STR-PCR连锁分析。结果 兄妹二人符合DMD诊断, 具有典型的DMD临床表现, 肌酸激酶、肌酸激酶同工酶、乳酸脱氢酶、羟丁酸脱氢酶和谷草转氨酶的水平均显著高于正常值, 肌电图呈肌源性损害, 肌肉HE染色符合DMD, 男患者的抗肌萎缩蛋白表达阴性, 女患者的少量肌纤维仍可见不连续膜阳性, 两患者抗肌萎缩蛋白基因的1~79号外显子未见缺失和重复突变, 女患者与男患者携带相同的母源性X染色体。结论 携带DMD致病基因的女性携带者可以具有临床以及实验室的典型表现, 应加强对携带者的全面检查。

关键词 假肥大型肌营养不良症 女性 家系 抗肌萎缩蛋白 多重连接探针扩增法

分类号

Sibling Brother and Sister both with Duchenne Muscular Dystrophy

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Abstract ABSTRACT:Objective To investigate the clinical and lab features of sibling brother and sister both with Duchenne muscular dystrophy (DMD). Methods We conducted comprehensive clinical and lab investigations including the test of serum enzymes, electromyography (EMG),electrocardiography, color Doppler echocardiography, HE staining of skeletal muscles, immunohistochemical study of dystrophin and utrophin, multiple ligation probe amplification（MLPA）on exon 1-79 of dystrophin gene, and short tandem repeat-poly-merase chain reaction of CA repeats located in dystrophin gene. Results These two patients were confirmed to suffer from DMD. They were characterized by typical features of DMD including typical clinical manifestations, increased serum enzymes, EMG presenting myogenic impairment, HE staining presentation belonging to DMD, negative dystrophin in brother, and inconstantly positive on the sarcolemma of sister. Furthermore, no deletion or duplication was found in the 1-79 exons of dystrophin gene. The suffering brother and sister carried the same maternal X chromosome. Conclusions Carriers of DMD gene show typical clinical and laboratory manifestations of DMD. Comprehensive examinations should be performed for such carriers.

Key words Duchenne muscular dystrophy female family dystrophin multiple ligation probe amplification

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