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MJD基因CAG不稳定性扩增与临床研究 Gene Mutation and Clinical Analysis in Machado-Joseph Disease

张宝荣, 黄鉴政, 夏家辉, 丁美萍, 朱永良, 应智林, 潘 乾, 汤熙翔, 姚梅琪 ZHANG Bao-rong1, HUANG Jian-zheng1, XIA Jia-hui2, DING Mei-ping1 ZHU Yong-liang1, YING Zhi-lin1, PAN Qian2, TANG Xi-xiang2, YAO Mei-qi1

1.姚梅琪浙江医科大学附属第二医院神经内科,杭州310009 2.湖南医科大学医学遗传学国家重点实验室,长沙410078 1.Department of Neurology,Second Hospital,Zhejiang Medical University,Hangzhou310009,China; 2.National Laboratory of Medical Genetics,Hunan Medical University,Changsha410078,China

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摘要　为了解Machado-Joseph病(MJD)基因突变及临床的神经电生理特点，对16个诊断为遗传性小脑性共济失调(SCA)家系的45例病人及30例家系的“正常”人作MJD基因突变分析，检出MJD基因的病人行肢体运动及感觉神经传导速度(MCV及SCV)、脑干诱发电位(BAEP)，视觉诱发电位(VEP)的检查。结果检出10个家系25例病人及1例症状前18岁女孩有MJD基因突变，CAG三核苷酸重复73～79次，异常等位基因片段长380～402bp，均为杂合子；正常人CAG三核苷酸重复18～40次，等位片段长200～270bp，电生理发现MJD的SCV减慢比MCV明显，而下肢的MCV、SCV又较上肢明显，BAEP、VEP均有不同程度的潜伏期延长或波的异常；MJD的父亲遗传早于母亲，进展也较块，临床以小脑性共济失调为突出症状，其次为构音障碍、突眼等，肌肉萎缩仅见于晚期病人；MRI示小脑萎缩较明显，脑干萎缩并不严重，未见明显的颈髓萎缩。

Abstract: To investigate the gene mutation of clinical and neuroelectrophysiological characteristics in Machado-Joseph disease(MJD). The gene mutation was detected in 45 patients diagnosed as spinocerebellar ataxia(SCA) and 30 “healthy relatives”. Brain stem evoked potentials (BAEP), visual evoked potentials(VEP) and motor conduction velocity (MCV) and sensory conduction velocity (SCV) were performed on MJD. Gene mutations were detected in 25 patients and a 18-year-old girl among 16 families. Trinucleotide repeats of CAG were 73～79. The fragments of abnormal alleles were 380～402bp, and all patients were heterozygous. The copy numbers of normal alleles were 18～40, fragments from 202～270bp. SCV reduction was much obvious compared to MCV, MCV and SCV in lower limb ?ere much more slow than that in upper's. BAEP, VEP were also delayed in latency. The anticipation in parental sex bias were much more obvioius than that in matental's. Cerebellar ataxia was most severe, the next were dysarthria and bulging eyes. Amytrophy was seen only in bed ridden patients. Cerebellar atrophy was more severe than brain stem, cord atrophy was n't observed in all MJD.

关键词　Machado-Joseph病 基因突变 临床电生理 Key words MJD Gene mutation Clinical electrophysiology

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