神经退行性疾病与遗传病专栏

脊髓小脑性共济失调3型/ Machado-Joseph病血清NSE与S100B浓度的测定

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摘要：

目的：探讨脊髓小脑性共济失调3型/ Machado-Joseph 病(SCA3/MJD)血清神经元特异性烯醇化酶(NSE)、S100B蛋白(S100B)作为神经元损伤/缺失和神经胶质增生的生化标志及其意义。方法：对102例SCA3/MJD患者和性别年龄与之相匹配的100例健康对照者进行血清NSE和S100B浓度的测定，分析其组间差异有无统计学意义及其与年龄、发病年龄、病程、CAG重复次数、国际协作共济失调评估量表（ICARS）评分、共济失调等级量表（SARA）评分的相关性。结果：SCA3/MJD患者组血清NSE和S100B浓度均较健康对照组有不同程度增高［(6.95±2.83) ng/mL与(4.83±1.70)ng/mL，P<0.05；(0.07±0.06) ng/mL与(0.05±0.02)ng/mL，P<0.05］。在SCA3/MJD患者组中，血清NSE浓度分别与年龄、病程、ICARS评分、SARA评分呈正相关；而血清S100B浓度与年龄、发病年龄、病程、ICARS评分、SARA评分无相关。CAG重复次数与不同年龄组的SCA3/MJD患者的血清NSE浓度、血清S100B浓度并无相关。结论：血清NSE可能作为的一种监测SCA3/MJD患者病程进展及评估病情严重程度的生化指标，而血清S100B仅可能作为一种显示SCA3/MJD患者脑损伤的潜在的生化指标。

关键词： 神经元特异性烯醇化酶　S100B蛋白　脊髓小脑性共济失调3型　Machado-Joseph 病　生化标志

Serum concentrations of NSE and S100B in spinocerebellar ataxia type 3/Machado-Joseph disease

Abstract:

ObjectiveTo determine the neuronal damage or loss and gliosis at the cellular level in spinocerebellar ataxia type 3/Machado-Joseph disease(SCA3/MJD), and evaluate the potential use of neuron-specific enolase (NSE) and protein S 100 B(S100B) serum concentrations as biochemical markers. MethodsSerum concentrations of NSE and S100B were measured in 102 SCA3/MJD patients and 100 healthy subjects matched by sex and age. The correlations between both markers and age, age of onset, disease duration, CAG repeat size, scores of international cooperative ataxia rating scale(ICARS), and scale for the assessment and rating of ataxia(SARA) were analyzed. ResultsCompared with the healthy controls, patients with SCA3/MJD had higher NSE serum concentrations ［(6.95±2.83)ng/mL vs (4.83±1.70) ng/mL, P<0.05］ and higher S100B serum concentrations ［(0.07±0.06) ng/mL vs (0.05±0.02) ng/mL, P<0.05］. In the SCA3/MJD patients group, NSE levels presented a positive correlation with age, disease duration, ICARS scores and SARA scores, whereas S100B levels did not correlate with age, age of onset, disease duration, ICARS scores and SARA scores. CAG repeat size did not correlate with the NSE levels and S100B levels in different age groups of SCA3/MJD patients. ConclusionSerum NSE might be a useful marker to monitor disease progression and represent the degree of severity of a certain disease. Elevated S100B serum concentrations in patients compared to healthy controls may suggest an application of this protein as a peripheral marker of brain impairment in SCA3/MJD.

Keywords: neuron-specific enolase (NSE)　protein S 100 B(S100B)　spinocerebellar ataxia type 3　Machado-Joseph disease　biochemical markers

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