

MM2型孤発性Creutzfeldt-Jakob病(sCJD)の 臨床的特徴とMM2皮質型sCJDの臨床診断基準案の提案

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MM2皮質型sCJDの新たな臨床診断基準案

A:

Confirmed with neuropathological (large confluent vacuoles) and immunohistochemical (perivacuolar prion protein deposits) analysis, genetic analysis of prion protein gene (no mutation and methionine homozygote at codon 129 of the prion protein gene), and Western blotting of the prion protein (type 2)

B:

- 1.progressive dementia**
- 2.no mutation and methionine homozygote at codon 129 of the prion protein gene**
- 3.hyperintensity lesions confined to the cerebral cortex on a diffusion weighted image of brain magnetic resonance image**
- 4.only 1 or none out of the following 4 clinical features within 6 months post-onset: (1) myoclonus, (2) pyramidal or extrapyramidal signs, (3) cerebellar ataxia or visual impairment, and (4) akinetic mutism**

Definite: A; Probable: B 1-4; Possible: B 1-3

'Probable' and 'possible' cases are in the absence of an alternative diagnosis from a routine investigation.

'Probable'

Sensitivity:
77.8%

Seven of 9 patients with MM2-cortical sCJD can be diagnosed.

Specificity:
98.5%

Thirteen of 847 patients who did not have MM2-cortical type sCJD can be misdiagnosed as MM2-cortical type sCJD. The 13 misdiagnosed cases consisted of 3 with MM1+2C, 1 with MM2T type sCJD, and 9 patients with no prion diseases.

解 説

- MM2皮質型sCJDの臨床診断基準案を作成した
- その診断基準案の「MM2皮質型sCJDほぼ確実例」を診断する感度は77.8%で特異度は98.5%であった
- MM2視床型については、神経症候や検査所見で特異的なものを同定できず、新たな臨床診断基準案を作成することは出来なかった