MM2皮質型孤発性Creutzfeldt-Jakob病の臨床診断基準案の作成

研究分担者：金沢大学大学院脳老化・神経病態学（神経内科学） 浜口　毅

「診断基準の策定・改訂」

MM2皮質型孤発性Creutzfeldt-Jakob病の臨床診断基準案

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 prion protein gene), and Western blotting of prion protein (type 2)

B:

1. **progressive dementia**
2. **no mutation and methionine homozygote at codon 129 of prion protein gene**
3. **hyperintensity lesions confined to cerebral cortex on diffusion weighted image of brain magnetic resonance image**
4. **only 1 or less out of the following 4 clinical features within 6 months after onset: (1) myoclonus, (2) pyramidal or extrapyramidal sign, (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 routine investigation.

'Probable'	
Sensitivity: 90.0%	Nine of 10 patients with MM2-cortical sCJD can be diagnosed.
Specificity: 99.1%	Seven of 771 patients who did not have MM2-cortical type sCJD can be misdiagnosed as MM2-cortical type sCJD. None of 7 patients were prion diseases.

解　説

1. 新たなMM2皮質型孤発性Creutzfeldt-Jakob病（sCJD）の診断基準案を提案した。
2. この診断基準でのprobable MM2型皮質型sCJDの感度は88.9%で、特異度は99.1%であった。