‘孤発性Creutzfeldt−Jakob病’と診断されている症例の中に医原性Creutzfeldt−Jakob病症例が含まれている可能性についての検討

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「診断基準の策定・改訂」、「診療ガイドラインの策定・改訂」に関する研究

Comparison of the clinical features between CJD patients with and without history of neurosurgery

	CJD with history of neurosurgery	CJD without history of neurosurgery	p
Number of patients	27	1,128	
Sex, female (%)	63.0%	57.4%	n.s.
Age at onset (years), (range)	71.0 ± 8.8, (49 - 88)	68.7 ± 9.6, (30 – 91)	n.s.
Disease duration of CJD* (months), (range)	6.1 ± 7.8, (1 – 28)	6.7 ± 12.0, (0 – 171)	n.s.
Incubation period** (years), (range)	15.0 ± 9.1, (1 – 35)		
Polymorphism at codon 129 of prion protein gene			
Met/Met, % (n)	92.6% (25)	97.6% (1,101)	n.s.
Met/Val, % (n)	7.4% (2)	1.9% (22)	
Val/Val, % (n)	0% (0)	0.4% (5)	
Positive rate of PSWCs on EEG, % (n)	81.5% (22/27)	94.3% (1,057/1,121)	p=0.021
Positive rate of 14-3-3 protein in cerebrospinal fluid, % (n)	90.9% (20/22)	84.1% (675/803)	n.s.
Positive rate of tau in cerebrospinal fluid (cut off 1200 pg/ml), % (n)	92.8% (13/14)	88.7% (503/567)	n.s.

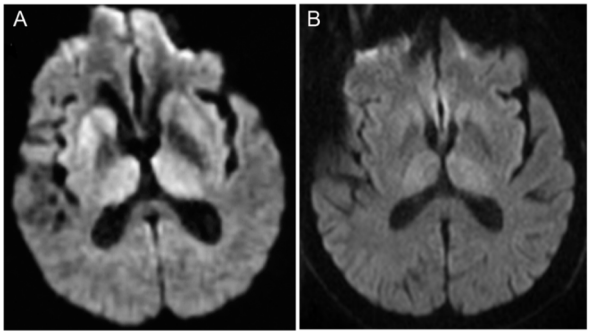
*Disease duration of CJD: duration between the onset of CJD and the appearance of the akinetic mutism or death in the patients who died without akintic mutism, **Incubation period: duration between neurosurgery and the onset of CJD.
CJD: Creutzfeldt-Jakob disease, PSWCs: periodic sharp-wave complexes, EEG: electroencephalogram, n.s.: not significant

Patients with history of neurosurgery who had no periodic sharp-wave complexes during whole disease duration of Creutzfeldt-Jakob disease.

Pt.	Age at onset	Sex	Initial symptom	Disease duration*, months	Codon 129 of PrP	Hyperintensity lesions on DWI	Pathological findings
1	75	Male	Dementia	11	Met/Met	CC, BG	ND
2	49	Female	Insomnia	28	Met/Met	ND	MM2-thalamic type sCJD
3	63	Female	Gait disturbance	19	Met/Met	BG, Th	ND
4	75	Female	Drowsiness, gait disturbance	6	Met/Met	BG, Th	CJD-MMiK
5	64	Female	Visual impairment	21	Met/Met	CC	MM2-cortical type sCJD

*Disease duration: duration between the onset of CJD and the appearance of the akinetic mutism or death in the patients who died without akinetic mutism.
Pt.: patient, *PrP*: prion protein gene, DWI: diffusion-weighted images, ND: not done, CC: cerebral cortex, BG: basal ganglia, Th: thalamus

Hyperintensity lesions in bilateral thalamus on DWI of the brain in 2 patients with history of neurosurgery who had no PSD .



A. Patient 4　　B. Patient 3

解　説

1. 孤発性CJDまたは分類不能のCJDと診断されている症例の中には、硬膜移植を伴わない脳外科手術歴があり、CJD-MMiKと同様の非典型的な臨床症候、病理所見、プロテアーゼ抵抗性PrPを呈する症例が存在する。
2. プリオン蛋白遺伝子コドン129多型がMMで頭部MRI DWIで両側視床に高信号を認めることがCJD-MMiKの診断マーカーとなる可能性がある。