

Table 1a. Clinical characteristics of patients with V180I genetic Creutzfeldt-Jakob disease

Patient	Sex	Age		c129	Stage	MC	Brain Atrophy	DWI Hiperintensity		BGA	IINED	IIED	PSWC
		Onset	Exam					Cortex	BG				
1	F	90	90	MM	1	N	Mild	Bi (R<L)	N	10–11	Lt T	N	N
			91		3	Y	Moderate	Bi (R<L)	Bi (R<L)	poor	Gen	N	N
2	F	77	80	MM	2	N	Mild	Bi (R>L)	N	8–9	Rt F	N	N
3	M	70	71	MV	2	Y	Moderate	Bi (R<L)	Bi (R<L)	11–12	Multi	N	N
4	F	68	69	MM	2	Y	Mild	Bi (R<L)	N	9–10	Multi	N	N
5	F	81	81	MM	2	Y	Mild	Bi (R>L)	Bi (R>L)	9–10	Gen	N	N
6	F	80	81	MM	2	N	Mild	Bi	Bi	10–11	Gen	N	N
7	M	70	71	MM	2	Y	Mild	Bi	N	11–12	N	N	N
8	F	80	80	MM	2	Y	Mild	Bi (R<L)	N	11–12	Lt Hemi	N	N
9	M	78	79	MM	2	Y	Mild	Bi (R<L)	N	9–10	Lt T	N	N
			80		3	Y	Severe	Bi	Bi	6–7	Bi CPO	N	N
10	F	85	85	MM	2	N	Mild	Bi (R>L)	N	8–9	Bi F (R>L)	N	N
			85		3	Y	Severe	Bi (R>L)	N	poor	Gen	N	N

Table 1b. Clinical characteristics of patients with M232R genetic Creutzfeldt-Jakob disease

Patient	Sex	Age		c129	Stage	MC	Brain Atrophy	DWI Hyperintensity		BGA	IINED	IIED	PSWC
		Onset	Exam					Cortex	BG				
1	M	71	72	MM	1	N	Mild	Bi (R>L)	Bi	9–10	Rt F	N	N
			72		2	Y	Moderate	Bi	Bi	poor	Bi F	Lt T	N
			72		3	Y	Moderate	Bi	Bi	poor	Bi FCP	N	N
2	M	66	66	MM	1	N	Mild	Lt	N	9–10	Lt F	N	N
			67		3	Y	Moderate	Bi (R<L)	Bi (R<L)	4–5	Gen	Lt hemi	N
3	F	42	42	MM	3	Y	Mild	Bi	Bi	poor	Gen	Gen	Y
4	M	71	74	MV	2	Y	Mild	Lt	N	9–10	Lt F	N	N
			75		3	Y	Moderate	Bi (R<L)	Bi (R<L)	poor	Lt hemi	Lt hemi	N
5	F	85	86	MM	2	N	Moderate	Bi (R<L)	N	8–9	Bi P (R<L)	Bi P (R<L)	N
			92		3	N	Moderate	Bi (R<L)	N	poor	Multi	Lt F	N
6	M	64	64	MM	2	Y	Mild	Bi (R<L)	Bi (R<L)	12–13	Bi F (R<L)	Lt hemi	N
			64		3	Y	Mild	Bi (R<L)	Bi (R<L)	poor	Lt hemi	Lt hemi	Y

c129: codon 129, MC: myoclonus, DWI: diffusion-weighted imaging, BG: basal ganglia, BGA: background activity,

IINED: interictal nonepileptiform discharges, IIED: interictal epileptiform discharges, MM: methionine-methionine, MV: methionine-valine,

Bi: bilateral, Gen: generalized, F: frontal, T: temporal, P: parietal, Hemi: hemispheric, Multi: multi focal, CPO: centro-parieto-occipital