

Supplemental Table: Clinical, genetic and imaging findings of 30 FXTAS cases with functional neuroimaging reported in the literature.

	Clinical findings		CGG repeats	MCP sign	SPECT	
	Symptoms	L-Dopa response			DaTSCAN	IBZM
Zuhlke et al; 1 female (J Neurol 2004)	T, A, N, P	-	84	N	Abnormal	Abnormal
Ceravolo et al; 4 males (Neurology 2005)	T, A, P	-	105	Y	Normal	-
	T, P	-	73	N	Normal	-
	A, C, AutD, P	N	85	N	Normal	-
	T, P	N	97	-	Normal	-
Kamm et al; 1 male (Brain 2005)	T, P, A, AutD	N	61	N	-	Abnormal
Scaglione et al; 5 Males (J Neurol 2008)	T, P, G, A	-	103	Y	Normal	-
	T, P, G, A	-	80	Y	-	-
	T, P, A	N	91	Y	Abnormal	Abnormal
	T, P, G, A	N	97	Y	Abnormal	-
	T, P, A	-	95	Y	Abnormal	-

Apartis et al; 4 females, 18 males* (Neurology 2012)	A (91%) T (86%) P (64%) C (55%)	N (3/3)	102	64% (14/22)	47% (7/15) abnormal	-
Hall et al; 2 females, 1 male (Parkinsonism Relat Disord 2010)	T, P T, P, C T, P	Y Y Y**	41 51 60	N N N	Normal Normal Normal	- - -
Healy et al (case 1) (Mov Disord 2009)	T, P, N, A	N	72	Y	Abnormal	Abnormal
& De Pablo- Fernandez et al (case 2) 2 males	T, P, A, G	N	100	N	Abnormal	-
Total:		Y 23% (3/13)		Y 57% (21/37)	Abnormal 45% (13/29)	Abnormal 100% (4/4)

* Only 15 patients with functional neuroimaging available

** responded to pramipexole

Abbreviations: A, ataxia; AutD, autonomic dysfunction; C, cognitive decline; DaTSCAN, striatal dopamine transporter SPECT; G, vertical gaze palsy; IBZM, striatal postsynaptic D2 receptor SPECT; MCP, middle cerebellar peduncle; N, no; P, parkinsonism; T, tremor; Y, yes.