

Supplementary Table 1. Summary of previous reports with *FBXO7* mutation

	Shojaee et al. ¹	Di Fonzo et al. ²	Paisán-Ruiz et al. ³		Yalcin-Cakmakli et al. ⁴	Gunduz et al. ⁵	Lohmann et al. ⁶	Conedera et al. ⁷	Wei et al. ⁸	Jin et al. ⁹	Lorenzo-Betancor et al. ¹⁰	Yoo et al. ¹¹	Correa-Vela M ¹²	Wang et al. ¹³	This study	Total	Frequency (%)	
Reported year	2008	2009	2010		2014	2014	2015	2016	2018	2020	2020	2020	2020	2021	2022			
Family no.	1	1	2	1	2	1	1	1	1	1	1	1	1	1	1	1	18	
No. of cases	10	2	2	3	1	2	2	2	1	1	2	3	1	1	2	1	36	
Zygosity	Homozygote	Homozygote	Compound heterozygote	Homozygote	Homozygote	Homozygote	Homozygote	Homozygote	Compound heterozygote	Compound heterozygote	Compound heterozygote	Compound heterozygote	Homozygote	Compound heterozygote	Compound heterozygote			
Mutation																		
Mutation 1	R378G	R498X	IVS7+1G/T	R498X	R498X	R498X	R498X	L34R	R498X	N51S	W134X	G39R	Q388X	S123X	R345P	S356RfsTer56		
Mutation 2	R378G	R498X	T22M	R498X	R498X	R498X	R498X	L34R	R498X	E470X	IVS5-1G>A	L280fs	R27H	S123X	R345P	R27H		
Country of origin	Iran	Italy	Netherlands	Pakistan	Turkey	Turkey	Turkey	Turkey	Turkey	China	China	Yemen	Korea	Morocco	China	Korea		
Clinical findings																		
Sex	F 2, M 8	F, M	F, M	F 3, M 1	F, M	F, M	F, M	F, M	F	M	M 2	F 2, M	M	F	F, M	M		
AAO		10, 13	18, 19	17, 24, 22	17	14, 10	13, 17	52, 41	17	16	6 mo, 6 mo	21, 27, 30	28	2	30, 31	42		
Parkinsonism																		
Rigidity	3	2	2	3	1	2	2	2	1	1	2	3	0	1	2	1	28	77.8
Bradykinesia	3	2	2	3	1	2	2	2	1	1	NA	3	1	1	2	1	27	79.4
Rest tremor	0	2	1	0	0	1	1	2	1	1	NA	1	1	NA	1	0	12	36.4
Postural instability	3	2	2	3	1	2	2	2	1	0	NA	2	0	NA	2	1	23	69.7
Dystonia	NA	2	0	1	0	0	1	0	0	0	NA	2	0	NA	1	0	7	30.4
Pyramidal signs																		
Babinski signs	10	2	2	3	0	0	1	NA	0	1	NA	0	0	NA	1	0	20	64.5
Spasticity	10	2	0	NA	NA	0	NA	NA	NA	NA	2	0	0	NA	2	0	16	69.6
Hyperactive DTR	10	2	2	3	0	0	1	0	0	1	NA	0	0	NA	1	0	20	60.6
Cognitive decline	0	0	0	3	0	2	1	0	1	1	2	2	0	1	1	1	15	41.7
Other signs		Dysarthria, dysphagia, slow saccades, urinary/fecal incontinence, reduced upgaze	Slow saccades, dysphagia, reduced upgaze	Apraxia of eye opening, supranuclear gaze palsy, slow saccades, swallowing difficulty		Perioral myoclonus, tongue protrusion, psychosis	Chorea, tic, tachpemia, ICD	pRBD, hallucination	Slow saccades			Saccadic hypometria, cogwheel pursuit, hyposmia	Acute ataxia, kinetic tremor, upgaze limitation, absence epilepsy, paraplegia, dysphagia, optic neuropathy		Seizure, abnormal behavior			
Treatment																		
Levodopa response	Responsive 1/ no trial 2/NA 7	2	2	3	1	2	2	2	1	1	NA	Responsive 2/ no trial 1	1	0	2	0	19	88.0
Motor fluctuation	NA	2	2	NA	0	NA	NA	NA	1	0	NA	1	NA	NA	NA	0	6	31.6
Dyskinesia	NA	2	2	3	1	2	NA	NA	0	0	NA	1	NA	NA	NA	0	11	57.9
Complications	NA	Behavioral disturbances 2	Behavioral disturbances 2	Mood disorder 1	Psychosis, agitation	Psychosis, agitation, ICD	ICD	Hallucination	ICD, aggression, disinhibition	Manic behavior	NA	Aggressive behavior	Delusion, ICD		Gastrointestinal symptoms	Aggressive behavior, ICD, somnolence		
Imaging																		
MRI	Normal	Normal	Normal	General atrophy 2	Normal			Temporal atrophy	Mild general atrophy	Normal	Normal	Normal	Iron accumulation both pallidum	Moderate hippocampal and extensive cortical atrophy	Normal			
PET/SPECT			CIT SPECT no uptake	CIT PET no uptake									CIT PET no uptake	DaTSCAN normal	CIT PET no uptake	4		

NA, not available; AAO, age at onset; DTR, deep tendon reflex; pRBD, probable rapid eye movement sleep disorder; ICD, impulse control disorder; MRI, magnetic resonance imaging; PET, positron emission tomography; SPECT, single-photon emission computed tomography; CIT, N-(3-fluoropropyl)-2 β -carbon ethoxy-3 β -(4-iodophenyl) nortropane.

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