

Supplementary Table 1. Review of literature of patients with PARK-*PINK1* analyzed in the MDSgene database

S.NO	STUDY	TYPE OF STUDY	COUNTRY	N No.	MOTOR SYMPTOMS	NON-MOTOR SYMPTOMS	REPORTED MUTATIONS
1	Sharma, 2024 ¹	Family study	India	1	T, B, R, Dystonia	Anxiety Depression Cognitive impairment Constipation Insomnia	p.Phe385Ser: HOM
2	Caritativo,2023 ²	Family study	Philippines	5	B, T, R	-	p.Leu347Pro: HOM
3	Bakhit,2023 ³	Mixed	Sudan	13	B, R, T, postural instability	Anxiety/ depression/ cognitive impairment	p.Gln533Profs*29: HOM p.Arg483Serfs*7: HOM p.Gln456*: HOM
4	Soudyab, 2022 ⁴	Mixed	Iran	4	-	-	p.Asn223Lysfs*12: HOM p.Gly408Alafs*6: HOM c.1124-2A>T: HOM p.Gly163Valfs*22: HOM
5	Morales-Briceno, 2022 ⁵	Case report	Australia	4	T, B, R, Dystonia, motor fluctuations	Anxiety and depression	p.Leu347Pro: HOM
6	Kukkle,2022 ⁶	Mutational analysis	India	1	T, B, R	-	c.959+1G>A: HOM
7	Patel,2021 ⁷	Mutational analysis	New Zealand	5	T, B, R, Foot dystonia	Anxiety, Hallucinations, RBD, panic attacks	p.Leu347Pro: HOM
8	Milanowski,2021 ⁸	Mutational analysis	Poland	3	B, R, T	-	p.Ile368Asn: HOM p.Ala168Pro: HOM p.Gln456*: HOM
9	Li, 2021 ⁹	Case report	China	1	T, B, R	-	p.Arg492* + c.1488+1G>A: CH
10	Hayashida,2021 ¹⁰	Mutational analysis	Japan	3	T,B,R, Postural instability, hyperreflexia	Urinary incontinence Hyposmia	p.Tyr29Argfs*71: HOM p.Glu113Lysfs*45: HOM p.Gln115Thrfs*27: HOM
11	Smaili,2021 ¹¹	Mutational analysis	Morocco	3	B, T, R, postural instability	-	p.Gln456*: HOM
12	Tan,2020 ¹²	Mutational analysis	Malaysia	5	T, B, R, motor fluctuations	Cognitive impairment RBD Constipation Visual / auditory hallucinations	p.Leu347Pro: HOM
13	Chen,2020 ¹³	Mutational analysis	China	1	-	-	p.Asn521Thrfs*40: HOM
14	Li,2020 ¹⁴	Mutational analysis	China	1	-	-	p.Leu485_Arg497del+ p.Arg492*: CH.
15	Tan, 2019 ¹⁵	Mutational analysis	UK	2	B,R	-	c.(?_94)_(387+1_388-1)del+ p.Trp90*: CH

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							c.(959+1_960-1)_(1123+1_1124-1)del: HOM
16	Kishore, 2018 ¹⁶	Family study	India	2	T,B,R, Motor fluctuations	Depression	p.Phe385Ser p.Tyr404*
17	Wang et al, 2018 ¹⁷	Family study	China	1	B,R	Insomnia, urinary urgency	p.Lys520Argfs*3: HOM
18	Taghavi, 2018 ¹⁸	Mutational analysis	Iran	6	T, B, R, motor fluctuations	Hyposmia Autonomic involvement RBD Cognitive decline	p.Arg246*HOM
19	Russ, 2018 ¹⁹	Mixed(Generation of iPSC)	Sweden	1	-	-	p.Gln456*:HOM
20	Schormair, 2018 ²⁰	Mutational analysis	Czech Republic and Germany	1	T, R, B, Dysarthria, Dyskinesias	D,A	c.1474C>T;p.Arg492Ter: HOM
21	Gustavsson,2017 ²¹	Mutational analysis	Norway	4	T, early motor fluctuations	-	c.(776+1_777-1)_(959+1_960-1)dup+ p.Ser73Leu+ p.Arg152Trp :CH c.(776+1_777-1)_(1123+1_1124-1)del: HOM p.His360Arg + c.(776+1_777-1)_(1123+1_1124-1)del: CH p.Leu77Pro: HOM
22	Zech, 2017 ²²	Mutational analysis	Germany	1	Foot dystonia, B, R	-	p.Ala339Thrn+ p.Leu249Val: CH
23	Spataro, 2017 ²³	Mutational analysis	Spain	1	-	-	p.Trp90*:HOM
24	Bouhouche,2017 ²⁴	Mutational analysis	Morocco	2	B,R,T, Dyskinesias	Psychosis Sleep disorder Cognitive impairment Autonomic involvement	p.Gln456*: HOM p.Leu539Phe: HOM
25	Norman, 2017 ²⁵	Family study	Morocco	1	B,R,T, Lower limb hyperreflexia	Nil	p.Ala217Asp: HOM
26	Halder, 2015 ²⁶	Mutational analysis	India	1	R,T	Postural instability	p.Gln267*: HOM
27	Al-Mubarak,2015 ²⁷	Mutational analysis	Saudi Arabia	3	B, R, T, Dystonia	Depression	p.Thr313Met:HOM p.Gly409Arg:HOM
28	Steele, 2015 ²⁸	Mutational analysis	Guam	3	B, R, T, Head bobbing	-	p.Leu347Pro:HOM
29	Siuda,2015 ²⁹	Family study	Poland	2	B, R, T, Foot Dystonia	Autonomic dysfunction Sleep fragmentation	p.Gln456*:HOM

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						Lower limb paraesthesia	
30	Monroy-Jaramillo, 2014 ³⁰	Mutational analysis	Mexico	9	B,R,T, Dyskinesias	Autonomic involvement Sleep disorder Cognitive decline	c.(_94)_(387+1_388-1)del: HOM c.(776+1_777-1)_(1123+1_1124-1)del + c.(387+1_388-1)_(675+1_676-1)del: CH c.(959+1_960-1)_(1123+1_1124-1)dup + c.(387+1_388-1)_(675+1_676-1)dup: CH p.Tyr444Metfs*39: HOM p.Cys92Alafs*15: HOM
31	Darvish, 2013 ³¹	Mutational analysis	Iran	7	B,R,T	Cognitive decline	c.(387+1_388-1)_(675+1_676-1)del + c.(387+1_388-1)_(959+1_960-1)del: CH c.(776+1_777-1)_(959+1_960-1)del :HOM c.(959+1_960-1)_(1123+1_1124-1)del: HOM
32	Koziorowski, 2013 ³²	Mutational analysis	Poland	1	R, B	-	p.Ile368Asn: HOM
33	Kilarski, 2012 ³³	Mutational Analysis	UK	1	T, R, B,	Urinary urgency, excessive sweating	p.Leu347Pro: HOM
34	Lohmann, 2012 ³⁴	Mixed	Turkey	3	-	-	p.Leu31*: HOM p.Pro416Leu: HOM
35	Scornaienchi, 2012 ³⁵	Mutational analysis	Italy	3	-	-	p.Gln126Pro: HOM p.Trp437*: HOM p.Gln456*: HOM
36	Keyser, 2010 ³⁶	Mutational analysis	South Africa	3	-	-	p.Tyr258*: HOM
37	Samaranch,2010 ³⁷	Family study	Spain	6	T, B, R, Postural instability	Anxiety	p.Leu485_Arg497del: HOM p.Leu485_Arg497del+ p.Glu417_Arg497del: CH
38	Cazeneuve,2009 ³⁸	Family study	Sudan	3	T, B, R, Hyperreflexia Foot dystonia	-	deletions of PINK1 exons 4-8 + 88bp insertion in DDOST intron 2: HOM
39	Camargos,2009 ³⁹	Mutational analysis	Brazil	1	T	Anxiety	c.(1251+1_1252-1)_(1488+1_1489-1)del: HOM
40	Myhre,2008 ⁴⁰	Family study	Jordan	4	-	-	p.Pro416Arg:HOM p.Ser419Pro:HOM
41	Guo,2008 ⁴¹	Mixed	China	4	T,B,R, Hyperreflexia,	-	p.Thr313Met:HOM p.Arg492*:HOM
42	Choi, 2008 ⁴²	Mutational analysis	Korea	1	B,R,T	-	p.Asn367Ser + p.Asn521Ilefs*2:CH

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43	Kumazawa,2008 ⁴³	Mutational analysis	Japan/ Multicentric (13 countries)	6	T,B,R, Dystonia, hyperreflexia	Autonomic dysfunction	p.Cys388Arg:HOM p.Thr313Met:HOM p.Trp437Arg:HOM p.Asp297Metfs*22:HOM
44	Moro,2008 ⁴⁴	Mutational analysis	Germany and Canada	1	B,R,T	-	p.Val170Gly:HOM
45	Savettieri,2008 ⁴⁵	Family study	Italy	2	T, B,R, off state dystonia	Cognitive impairment	p.Asp297Metfs*22:HOM
46	Gelmetti,2008 ⁴⁶	Case report	Italy	1	T, B, R, foot dystonia	Depression, Sleep fragmentation	p.Val317Ile+p.Ala244Gly:CH
47	Prestel,2008 ⁴⁷	Family study	Germany	2	B,R,T, Dyskinesias	-	p.Gln126Pro: HOM
48	Weng, 2007 ⁴⁸	Mutational analysis	Taiwan	2	T, dystonia	-	p.Gln239*+p.Arg492*:CH p.Arg492*: HOM
49	Marongiu, 2007 ⁴⁹	Family study	Italy	1	B,R,T, Dyskinesias	Psychosis	c.1252-2_1272del+c.(?-94)_(*820_?)del: CH
50	Doostzadeh,2007 ⁵⁰	Mutational analysis	USA	1	B, R, T, Foot Dystonia	Hyposmia	p.Leu347Pro:HOM
51	Chishti,2006 ⁵¹	Mixed	Saudi Arabia	2	T, B, R, motor fluctuations	-	p.Thr313Met:HOM
52	Leutenegger,2006 ⁵²	Family study	Sudan	5	B,R,T, Foot dystonia, early dyskinesias	-	p.Ala217Asp:HOM
53	Hedrich,2006 ⁵³	Family study	Germany	4	B,R, Motor fluctuations	Psychiatric disturbances, RLS	p.Gln456*:HOM
54	Criscuolo,2006 ⁵⁴	Mixed	Italian	1	B,R,T, Early wearing off	Depression	p.Trp437*:HOM
55	Zadikoff,2006 ⁵⁵	Case report	Canada	1	T, B, R, painful off dystonia, On state dyskinesias, blepharospasm	Anxiety	p.Gln456*:HOM
56	Tan, 2006 ⁵⁶	Mutational analysis	Singapore	2	T,R,B, Dyskinesias	RLS	p.Arg246*:HOM p.Tyr258*:HOM
57	Ibanez, 2006 ⁵⁷	Mutational analysis	Europe and North Africa	12	T,B,R, Lower limb hyperreflexia, Dyskinesias, Dystonia	A, D, Psychosis	p.Gln456*: HOM p.Gln456*+p.Cys125Gly: CH p.Glu240Lys:HOM p.Leu369Pro+p.Arg492*: CH p.Gly386Ala:HOM p.Gly409Val:HOM p.Lys520Argfs*3:HOM p.Cys549Trpfs*5+p.Lys24Glyfs*31:CH

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58	Bonifati, 2005 ⁵⁸	Mutational analysis	Italy	3	B,R,T, Lower limb hyperreflexia, Dystonia	D, A, Postural instability	p.Ala168Pro:HOM p.Trp437*:HOM P.Gln456*:HOM
59	Klein,2005 ⁵⁹	Mutational analysis	Italy	1	B, R, Foot Dystonia, hypereflexia	Depression and autonomic dysfunction	p.Gln534_Ser535insGln:HOM
60	Albanese,2005 ⁶⁰	Case report	Italy	1	B, R, Freezing of gait	-	p.Ala168Pro: HOM
61	Li, 2005 ⁶¹	Family study	Multicentre (9 countries)	3	T, B, R, Hyperreflexia	Depression Hallucinations	p.Cys388Arg:HOM c.1123+1247*881del:HOM
62	Rogaeva, 2004 ⁶²	Mutational screen	USA	2	T, B, R, Early dyskinesias	-	p.Glu240Lys + p.Leu489Pro: CH p.Leu347Pro:HOM
63	Hatano, 2004 ⁶³	Family study	Japan	10	T,B,R, Lower limb hyperreflexia, Dyskinesias	Postural instability Psychosis Sleep disorder Anxiety	p.Arg246*:HOM p.Arg492*+p.Gln239*: CHp.His271Gln:HOM p.Leu347Pro:HOM p.Glu417Gly:HOM
64	Rohe,2004 ⁶⁴	Family study	Italy	1	T, B, R, Foot dystonia	Anxiety	p.Asp525Valfs*38:HOM
65	Valente, 2004 ⁶⁵	Mutational screen	Italy	2	T, B, R	Urinary urgency Orthostatic hypotension	p.Arg464His+p.Cys92Phe : CHp.Ala168Pro: HOM
66	Valente, 2004 ⁶⁶	Family study	Italy	9	B,T,R, Dyskinesia	Postural instability Autonomic involvement Cognitive decline	p.Gly309Asp:HOM P.Trp437*:HOM

A: Anxiety; B: Bradykinesia; CH: Compound heterozygous; D: Depression; del: Deletion; dup: Duplication; HOM: Homozygous, R: Rigidity; RLS: Restless leg syndrome; T: Tremor; UK: United Kingdom; USA: United States of America.

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