

Title: *LRRK2*-Related Parkinson Disease *GeneReview* Tables 4 and 5

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Note: The following information is provided by the authors and has not been reviewed by *GeneReviews* staff.

Table 4. Nonsynonymous Allelic Variants of *LRRK2*

Exon	RS#	cDNA	Amino Acid	Domain
1		c.28G>A	p.E10K	N-terminal
1	rs2256408	c.149G>A	p.H50R	N-terminal
2	rs72546335	c.155C>T	p.S52F	N-terminal
4	rs33995463	c.356T>C	p.L119P	N-terminal
6	ss197562451	c.632C>T	p.A211V	N-terminal
6	rs56108242	c.683G>C	p.C228S	N-terminal
6		c.693 T>C	p.S231P	N-terminal
7	rs28365216	c.713A>T	p.N238I	N-terminal
8		c.858 C>G	p.L286V	N-terminal
9	ss159695484	c.1000G>A	p.E334K	N-terminal
9	rs72546336	c.1088A>G	p.N363S	N-terminal
9	ss197562460	c.1096G>A	p.V366M	N-terminal
10		c.1134 C>T	p.L378F	N-terminal
11		c.1290 G>A	p.V430I	N-terminal
11	rs34594498	c.1256C>T	p.A419V	N-terminal
13		c.1517G>A	p.R506Q	N-terminal
14	rs35328937	c.1561A>G	p.R521G	N-terminal
14	ss159695485	c.1630 A>G	p.K544E	N-terminal
14	rs7308720	c.1653C>G	p.N551K	N-terminal
16		c.1847A>G	p.K616R	N-terminal
16		c.1828 T>C	p.I610T	N-terminal
17	ss142460315	c.1987T>C	p.S663P	N-terminal
18		c.2134A>G	p.M712V	ANK
18		c.2147C>T	p.A716V	ANK
18	rs10878307	c.2167A>G	p.I723V	ANK
19	rs34410987	c.2264C>T	p.P755L	ANK
19	rs35173587	c.2378G>T	p.R793M	ANK
19	rs72546337	c.2428A>G	p.I810V	ANK

Exon	RS#	cDNA	Amino Acid	Domain
19		c.2316 C>T	p.R772*	ANK
20		c.2611A>G	p.K871E	
21	rs58559150	c.2769G>C	p.Q923H	
21		c.2789A>G	p.Q930R	
22	rs17519916	c.2830G>T	p.D944Y	
23	ss159695492	c.2918G>A	p.S973N	
23	ss197562463	c.3018A>G	p.I1006M	LRR
24		c.3189 C>T	p.L1063F	
24	ss197562466	c.3200G>A	p.R1067Q	LRR
24	ss159695493	c.3287C>G	p.S1096C	LRR
24	ss142460318	c.3333G>T	p.Q1111H	LRR
25	rs34805604	c.3364A>G	p.I1122V	LRR
25	ss159695486	c.3451G>A	p.A1151T	LRR
25		c.3494T>C	p.L1165P	LRR
26		c.3574A>G	p.I1192V	LRR
27	rs72546324	c.3647A>G	p.H1216R	LRR
27	ss159695494	c.3683G>C	p.S1228T	LRR
27	rs60185966	c.3683G>T	p.S1228I	LRR
28	rs4640000	c.3784C>G	p.P1262A	LRR
29	ss159695488	c.3960A>	p.R1320S	
29	rs72546338	c.3974G>A	p.R1325Q	
29	rs17466213	c.4111A>G	p.I1371V	Roc
29	rs28365226	c.4125C>A	p.D1375E	Roc
30	rs7133914	c.4193G>A	p.R1398H	Roc
30	rs72546327	c.4229C>T	p.T1410M	Roc
30	ss197562470	c.4258G>A	p.D1420N	Roc
30		c.4314 A>G	p.I1438V	Roc
31		c.4324G>C	p.A1442P	Roc
31	ss159695489	c.4337C>T	p.L1446P	Roc
31		c.4348G>A	p.V1450I	Roc
31		c.4358 A>G	p.H1453R	Roc
31		c.4364delAT	p.D1455G	Roc
31		c.4402A>G	p.K1468E	Roc
31		c.4443 G>A	p.A1481T	Roc

Exon	RS#	cDNA	Amino Acid	Domain
31	ss197562475	c.4448G>A	p.R1483Q	Roc
32	rs35507033	c.4541G>A	p.R1514Q	COR
32	rs33958906	c.4624C>T	p.P1542S	COR
32	rs17491187	c.4666C>A	p.L1556I	COR
33	rs721710	c.4793T>A	p.V1598E	COR
34		c.4838T>C	p.V1613A	COR
34	rs35303786	c.4937T>C	p.M1646T	COR
34	rs11564148	c.4939T>A	p.S1647T	COR
36	rs11564176	c.5173C>T	p.R1725STOP	COR
36		c.5174G>A	p.R1725Q	COR
36		c.5183G>T	p.R1728L	COR
36		c.5183G>A	p.R1728H	COR
36		c.5272 A>C	p.H1758P	COR
37	ss197562481	c.5385G>T	p.L1795F	COR
37		c.5467C>A	p.Q1823K	COR
38		c.5605A>G	p.M1869V	COR
38	rs35602796	c.5606T>C	p.M1869T	COR
38		c.5610G>T	p.L1870F	COR
38		c.5620G>T	p.E1874STOP	COR
39	ss159695497	c.5822G>A	p.R1941H	MAPKKK
41		c.6016T>C	p.Y2006H	MAPKKK
41	rs34015634	c.6035T>C	p.I2012T	MAPKKK
41	ss159695498	c.6091A>T	p.T2031S	MAPKKK
42	ss197562487	c.6187delCTCTA	p.L2063STOP	MAPKKK
42	rs33995883	c.6241A>G	p.N2081D	MAPKKK
43	rs12423862	c.6356C>T	p.P2119L	MAPKKK
44		c.6397 A>G	p.N2133S	
44		c.6415T>A	p.C2139S	
44		c.6422C>T	p.T2141M	
44		c.6428G>A	p.R2143H	WD40
44		c.D2175H	p.D2175H	WD40
44	rs35658131	c.6566A>G	p.Y2189C	WD40
46	rs12581902	c.6782A>T	p.N2261I	WD40
47		c.6928 C>T	p.T2310M	WD40

Exon	RS#	cDNA	Amino Acid	Domain
47		c.7008 A>G	p.I2336V	WD40
48	ss197562484	c.7067C>T	p.T2356I	WD40
48	ss159695487	c.7168G>A	p.V2390M	WD40
49	ss159695499	c.7183G>A	p.E2395K	WD40
49	ss197562492	c.7187insGT	p.T2397STOP	WD40
49	rs3761863	c.7190C>T	p.T2397M	WD40
49	rs60545352	c.7224G>A	p.M2408I	WD40
49		c.7269 A>T	p.T2423S	WD40
50		c.7397T>A	p.L2446H	WD40
50	rs55633591	c.7435A>G	p.N2479D	WD40
51		c.7468_deIC	p.E2490STOP	WD40

Variants of frequency >1% are highlighted in grey.

RS# (Reference SNP ID) and SS# (Submitter SNP ID), see Entrez SNP (<http://www.ncbi.nlm.nih.gov/snp>)

Table 5. Silent Allelic Variants of *LRRK2*

Exon	RS#	cDNA	Amino Acid	Domain
2	ss142460309	c.224G>A	p.A75A	N-terminal
4	rs41286468	c.364T>C	p.L122L	N-terminal
5	rs10878245	c.457T>C	p.L153L	N-terminal
5	rs35517158	c.546A>G	p.K182K	N-terminal
7	ss142460312	c.824C>T	p.H275H	N-terminal
8	rs17490713	c.867T>C	p.N289N	N-terminal
8	rs57355477	c.893T>C	p.A298A	N-terminal
8	rs41286466	c.936G>T	p.A312A	N-terminal
12	rs35847451	c.1383C>T	p.S461S	N-terminal
13	ss159695490	c.1464A>T	p.L488L	N-terminal
15	ss142460314	c.1647A>G	p.G558G	N-terminal
17	ss142460316	c.2022A>C	p.V674V	N-terminal
19	ss159695491	c.2481T>C	p.S827S	ANK
22	rs7966550	c.2857T>C	p.L953L	
23		c.3021C>T	p.S1007S	LRR
24	rs35808389	c.3342A>G	p.L1114L	LRR
30	rs11175964	c.4269G>A	p.K1423K	Roc
30	ss197562472	c.4290C>T	p.A1430A	Roc
31		c.4323C>T	p.R1441R	Roc
34	rs1427263	c.4872C>A	p.G1624G	COR
34	rs11176013	c.4911A>G	p.K1637K	COR
34	ss197562478	c.4959A>G	p.L1653L	COR
35	ss142460319	c.5163A>G	p.S1721S	COR
37	rs10878371	c.5457T>C	p.G1819G	COR
43	rs10878405	c.6324G>A	p.E2108E	MAPKKK
44	rs34869625	c.6510C>A	p.G2170G	WD40
48	rs33962975	c.7155A>G	p.G2385G	WD40

Variants of frequency >1% are highlighted in grey.

RS# is the Reference SNP ID, see Entrez SNP (<http://www.ncbi.nlm.nih.gov/snp>)