

Link to PD mutation database: <http://grenada.lumc.nl/LOVD2/TPI/>

Structure of database.

The gene homepage gives the possibility of switching and selecting one of the six genes. It gives general information about the gene included in the database: gene name, chromosomal location, number of sequence variants, searching possibilities among the variants. The homepage provides links to other resources such as OMIM, HGMD and Gene Tests.

LOVD Parkinson's disease Mutation Database
PARKIN (PARK2)

LOVD Gene homepage

General information	
Gene name	PARKIN
Gene symbol	PARK2
Chromosome location	6q25.2-q27
Database location	The Parkinson's Institute
Curator	The Parkinson's Institute
Date of creation	May 27, 2008
Last update	June 12, 2008
Version	PARK2 080612
Add sequence variant	Submit a sequence variant
First time submitters	Resource here
Total number of unique DNA variants reported	125
Total number of individuals with variant(s)	156
Total number of variants reported	157

Sequence variant tables

- [Unique sequence variants](#): Listing of all unique sequence variants in the PARK2 database, without patient data
- [Complete sequence variant listing](#): Listing of all sequence variants in the PARK2 database
- [Variants with unknown pathogenicity](#): Listing of all PARK2 variants reported to have no noticeable phenotypic effect (note: excluding variants of unknown effect)

Search the database

- By type of variant**: View sequence variant table after selecting one type of variant
- Simple search**: Query the database by selecting the most important variables (exam number, type of variant, disease phenotype)
- Advanced search**: Query the database by selecting a combination of variables
- Based on patient origin**: View all variants based on your patient origin search terms

Links to other resources

OMIM - Gene	602518
OMIM - Disease	PARK2
HGMD	PARK2
GeneTests.org	PARK2

By clicking on 'Unique sequence variants', you can get the listing of the sequence variants: DNA change, protein change described by standard nomenclature, exon, mutation type, heterozygosity and frequency.

LOVD - Variant listings

Exon	DNA change	Protein	Frequency	Heterozygosity	Mutation Type	DB-ID
01	ex1 dupl	-	0/252xref	-	duplication	PARKIN_00107
01	c.(?_103)_7+7dup (Reported 2 times)	-	-	-	duplication	PARKIN_00081
01	c.18A>T	-	-	-	substitution	PARKIN_00002
01	c.7+10>A	-	-	-	splice site mutation	PARKIN_00003
02	c.8-7_171+7del	-	-	-	deletion	PARKIN_00002
02	c.34G>C	p.G12R	-	het	missense	PARKIN_00112
02	c.43G>A	p.Val13Met	-	-	missense	PARKIN_00004
02	c.52G>A	p.D18N	-	-	missense	PARKIN_00134
02	c.95A>C	p.R32T	-	-	missense	PARKIN_00109
02	c.97C>T	p.Arg33X	-	-	missense	PARKIN_00005
02	c.98G>A	p.Arg33Gln	-	-	missense	PARKIN_00006
02	c.101delA (Reported 2 times)	p.G34delArgX10	-	-	frameshift	PARKIN_00008
02	c.104delGAG (Reported 2 times)	p.G36delArgX5	-	-	frameshift	PARKIN_00007
02	c.110C>T	p.Pro37Leu	-	-	missense	PARKIN_00009
02	c.125G>C	p.Arg42Pro	-	-	missense	PARKIN_00010
02	c.136G>C	p.Arg46Pro	-	-	missense	PARKIN_00011
02	c.154delA (Reported 2 times)	p.Asn52MetXc29	-	-	frameshift	PARKIN_00013
02	c.154_155delAT	p.Asn52X	-	-	nonsense	PARKIN_00012
02	c.156_157met	p.D53X	-	het	nonsense	PARKIN_00111
02	c.164C>T	p.T53I	2/72 EOPD, 0/91dbf	-	missense	PARKIN_00110
02	c.167T>A	p.Val56Glu	-	-	missense	PARKIN_00014
03	ex3 del	p.N58QxK39	4/72 EOPD, 0/81 dbf	het	frameshift	PARKIN_00114
03	c.172-7_412+7del (Reported 2 times)	-	-	-	deletion	PARKIN_00083
03	c.201_221metT	p.Trp74CysXx8	-	-	frameshift	PARKIN_00015
03	c.235G>T	p.Glu79X	-	-	nonsense	PARKIN_00016

By clicking on each variant, a new window opens with a full table of variant information and patient information, phenotype, and frequency data as well as the reference link to the original NCBI abstract.

LOVD - Variant listings

Patient data (#0000080)

Patient ID	80
Disease	EOPD
Reference	Kann et al 2002
Template	DNA
Technique	SEQ
Remarks	-
Remarks (non public)	-
# Reported	1
Submitter	Assign
Created by	The Parkinson's Institute
Date created	2008-06-12 20:38:45

Variant data

Allele	Unknown
Reported pathogenicity	Unknown
Concluded pathogenicity	Unknown
Exon	02
DNA change	c.110C>T
RNA change	-
Protein	p.Pro37Leu
Frequency	-
Heterozygosity	-
Mutation Type	missense
DB-ID	PARKIN_00009
Status	Non public
Created by	The Parkinson's Institute
Date created	2008-06-12 20:38:45