

UKGTN Testing Criteria

Test name: Parkinson Disease 6 Gene Panel	
Approved name and symbol of disorder/condition(s): Parkinson Disease 6 Gene Panel - See Appendix 1	OMIM number(s): See Appendix 1
Approved name and symbol of gene(s): See Appendix 1	OMIM number(s): See Appendix 1

Patient name:	Date of birth:
Patient postcode:	NHS number:
Name of referrer:	
Title/Position:	Lab ID:

Referrals will only be accepted from one of the following:	
Referrer	Tick if this refers to you.
Consultant Neurologists	
Consultant Clinical Geneticists	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:	
Criteria	Tick if this patient meets criteria
1. Parkinson disease affected and Early onset (< 50 years) OR	
2. First degree relative affected at <50yrs and family history compatible with autosomal recessive inheritance OR	
3. late onset with family history	

Additional Information:

For panel tests: At risk family members where familial mutation is known do not require a full panel test but should be offered analysis of the known mutation

If the sample does not fulfil the clinical criteria or you are not one of the specified types of referrer and you still feel that testing should be performed please contact the laboratory to discuss testing of the sample.

Testing Criteria Appendix 1

Conditions in panel test

OMIM standard name of condition and symbol	OMIM number	HGNC standard name and symbol of the gene	HGNC number	OMIM number	References or locus specific database links
PARKINSON DISEASE 8, AUTOSOMAL DOMINANT; PARK8	607060	LEUCINE-RICH REPEAT KINASE 2; LRRK2	18618	609007	http://www.LOVD.nl/LRRK2 ; http://www.molgen.vib-ua.be/PDMutDB/
PARKINSON DISEASE 2, AUTOSOMAL RECESSIVE JUVENILE; PARK2	600116	PARKINSON PROTEIN 2, E3 UBIQUITIN PROTEIN LIGASE (PARKIN); PARK2	8607	602544	http://www.LOVD.nl/PARK2 ; http://www.molgen.vib-ua.be/PDMutDB/
PARKINSON DISEASE 7, AUTOSOMAL RECESSIVE EARLY-ONSET; PARK7	606324	PARKINSON PROTEIN 7; PARK7	16369	602533	http://www.LOVD.nl/PARK7 ; http://www.molgen.vib-ua.be/PDMutDB/
PARKINSON DISEASE 6, AUTOSOMAL RECESSIVE EARLY-ONSET; PARK6	605909	PTEN-INDUCED PUTATIVE KINASE 1; PINK1	14581	608309	http://www.LOVD.nl/PINK1 ; http://www.molgen.vib-ua.be/PDMutDB/
PARKINSON DISEASE 1, AUTOSOMAL DOMINANT; PARK1	168601	SYNUCLEIN, ALPHA (NON A4 COMPONENT OF AMYLOID PRECURSOR); SNCA	11138	163890	http://www.LOVD.nl/SNCA ; http://www.molgen.vib-ua.be/PDMutDB/
PARKINSON DISEASE 4, AUTOSOMAL DOMINANT; PARK4	605543	SYNUCLEIN, ALPHA (NON A4 COMPONENT OF AMYLOID PRECURSOR); SNCA	11138	163890	http://www.LOVD.nl/SNCA ; http://www.molgen.vib-ua.be/PDMutDB/
DEMENTIA, LEWY BODY; DLB	127750	SYNUCLEIN, ALPHA (NON A4 COMPONENT OF AMYLOID PRECURSOR); SNCA	11138	163890	http://www.LOVD.nl/SNCA ; http://www.molgen.vib-ua.be/PDMutDB/
PARKINSON DISEASE 17; PARK17	614203	VACUOLAR PROTEIN SORTING 35, YEAST, HOMOLOG OF; VPS35	13487	601501	15 families with pathogenic mutations described on OMIM 1 index case identified in house