**Part 1 – Clinical Description**

1. **\*\***Primary Clinical Diagnosis (check one):1

Parkinson’s disease: [ ] Present [ ] Absent \*\*Age at symptom onset:

Progressive supranuclear palsy: [ ] Present [ ] Absent \*\*Age at symptom onset:

Dementia with Lewy Bodies: [ ] Present [ ] Absent \*\*Age at symptom onset:

Parkinson’s disease dementia: [ ] Present [ ] Absent \*\*Age at symptom onset:

Multiple system atrophy: [ ] Present [ ] Absent \*\*Age at symptom onset:

Other (specify): [ ] Present [ ] Absent \*\*Age at symptom onset:

1. \*\*Signs Supportive of PD Diagnosis:1

Asymmetric onset: [ ] Present [ ] Absent [ ] N/A

Bradykinesia: [ ] Present [ ] Absent [ ] N/A

Resting Tremor: [ ] Present [ ] Absent [ ] N/A

Postural Instability: [ ] Present [ ] Absent [ ] N/A

Rigidity: [ ] Present [ ] Absent [ ] N/A

Gait difficulties: [ ] Present [ ] Absent [ ] N/A

Levo-dopa induced dyskinesia: [ ] Present [ ] Absent [ ] N/A

Olfactory loss: [ ] Present [ ] Absent [ ] N/A

Cardiac sympathetic denervation: [ ] Present [ ] Absent [ ] N/A

REM-sleep behavior disorder: [ ] Present [ ] Absent [ ] N/A

1. \*\*Response to anti-parkinsonism therapy:1

[ ]  Tried and responsive

[ ]  Inadequate dose

[ ]  Not tried/not given

[ ]  Tested and unresponsive

1Question and permissible values from [Coriell Institute for Medical Research](https://www.coriell.org/1/About-Us/Legal-Notice) used and modified with permission.

**Part II – Genetics Summary**

1. \*\*Was genetic testing performed? [ ]  Yes [ ]  No

IF YES, please answer questions below:

1. \*\*What year was the genetic testing performed?
2. \*\*Indicate the source(s) of the genetic test results: (Choose all that apply)

[ ]  Neurologist

[ ]  Physician

[ ]  Genetic counselor

[ ]  Medical records

[ ]  Other, specify:

1. \*\*Was the patient informed of the test results? [ ]  Yes [ ]  No
	1. \*\*If YES, who informed them of the results?

[ ]  Genetic Counselor

[ ]  Neurologist

[ ]  Self (results from Direct-to-Consumer test)

1. \*\*Known Variant/s in subject’s DNA:1 [ ]  Present [ ]  Absent [ ]  Unknown
2. If present or absent, describe:1

1Question and permissible values from [Coriell Institute for Medical Research](https://www.coriell.org/1/About-Us/Legal-Notice) used and modified with permission.

1. \*\*Has the participant had a sample drawn for DNA banking? [ ]  Yes [ ]  No [ ]  Unknown
	1. If YES:
		1. \*\*Specify the type of sample drawn:

[ ]  Blood draw [ ]  Buccal smear (cheek swab) [ ]  Saliva [ ]  Other, specify:

* + 1. \*\*Specify the study for which the sample was initially taken:
		2. \*\*Specify where the sample is banked, if known:
		3. \*\*Did the participant sign a consent form at the time the sample was taken?

[ ]  Yes [ ]  No [ ]  Unknown

* + 1. \*\*Does the consent form for this sample allow for sharing of the sample? **[ ]** Yes **[ ]** No
		2. \*\*Has the participant given a sample of blood to a repository? **[ ]** Yes **[ ]** No

\*\*If YES, name of repository:

1. \*\*Has the participant registered for brain donation? **[ ]** Yes **[ ]** No
	1. \*\*If YES, name of repository:
2. \*\*Is a brain available from a family member? **[ ]** Yes **[ ]** No
3. If YES:
	* 1. \*\*Name of repository:
		2. \*\*Sample ID:
		3. \*\*Repository contact:
		4. \*\*Type of tissue collected:

**Part III – Study Description**

1. Study type(s) (Please check all that apply):2

[ ]  Longitudinal

[ ]  Case-control

[ ]  Case set

[ ]  Control set

[ ]  Parent-offspring trios

[ ]  Cohort

[ ]  Clinical trial

[ ]  Other, specify:

1. Is this study related to a pre-existing registered dbGaP study? 2 [ ] Y [ ]  N
2. If YES, please provide the phs accession number and/or title of the study: 2
3. Is aggregate-level data appropriate for General Research Use? 2 [ ] Y [ ]  N
4. Please check all data types expected for this study: 2
	1. General

[ ]  Individual Phenotype

[ ]  Individual Genotype

[ ]  Individual Sequencing

[ ]  Supporting Documents

[ ]  Metagenomic

[ ]  Proteomic/Metabolomic

[ ]  Images

* 1. Sample Types

[ ]  Germline

[ ]  Tumor/Normal

[ ]  DNA

[ ]  RNA

[ ]  Mitochondria

[ ]  Microbiome

[ ]  From Repository

* 1. Array Data

[ ]  SNP Array

[ ]  Expression Array

[ ]  Methylation Array

* 1. Genotypes

[ ]  Array derived genotypes

[ ]  CNV calls from microarray

[ ]  CNV calls derived from Sequencing

[ ]  Genotype calls derived from Sequence

[ ]  Somatic SNV (MAF)

[ ]  Array CGH CNVs

* 1. Sequencing

[ ]  Whole Genome

[ ]  Whole Exome

[ ]  Targeted Genome

[ ]  Targeted Exome

[ ]  Whole Transcriptome

[ ]  Targeted Transcriptome

[ ]  Epigenomic Marks

[ ]  Sanger

[ ]  16S rRNA

* 1. Analysis

[ ]  Association/Linkage results

[ ]  Array-derived expression

[ ]  Bulk RNA Seq derived expression

[ ]  Array-derived methylation

**Part IV – Genotype Platform Information**

1. Name and version: 2
2. Vendor: 2
3. # Probes: 2
4. URL: 2
5. Description (optional): 2

2Question and permissible values from dbGaP/database of Genotypes and Phenotypes/ National Center for Biotechnology Information, National Library of Medicine (NCBI/NLM)/<https://www.ncbi.nlm.nih.gov/gap> used and modified with permission.

**Part V: Variant/Mutation Analysis**

1. Lab name:
2. Date report issued:
3. Variant analysis results available on this participant: [ ]  Yes [ ]  No

 (IF NO, Stop completing form)

1. Variant analysis performed on the participant: [ ]  Yes [ ]  No
2. If NO, was variant analysis performed on a family member? [ ]  Yes [ ]  No
3. If NO, provide explanation:
4. Variant analysis results:
5. Variant(s) detected:

[ ]  Homozygous

[ ]  Compound Heterozygous

[ ]  Heterozygous

[ ]  Hemizygous

[ ]  Digenic (variants in more than one gene)

[ ]  No pathogenic variant detected

1. Are there additional variants in other genes of unknown significance? [ ]  Yes [ ]  No

If YES, indicate:

1. Are there additional genes sequenced with no variants detected? [ ]  Yes [ ]  No

If YES, indicate:

1. What type of testing was performed?

[ ]  mtDNA panel testing

1. What tissue?

[ ]  Blood

[ ]  Muscle

[ ]  Liver

[ ]  Other, please specify

1. What genes?

[ ]  mtDNA genome deletion/duplication analysis

1. What tissue?

[ ]  Blood

[ ]  Muscle

[ ]  Liver

[ ]  Other, please specify

1. What genes?

[ ]  Karyotype

1. What tissue?

[ ]  Blood

[ ]  Amnio

[ ]  Skin

[ ]  Other, please specify

1. Allele specific Information
2. Allele #1
3. Gene Name:
4. Variant Class:

[ ]  Reduced Number of Copies

[ ]  Increased Number of Copies

[ ]  Missense

[ ]  Nonsense

[ ]  Splice

[ ]  Pseudoexon

[ ]  Potential (variant of unknown significance)

[ ]  Subexonic Insertion/Deletion

[ ]  Other, specify:

1. For Exonic Deletions/ Duplications:
	* 1. Was the copy number directly tested for all exons?[ ]  Yes [ ]  No [ ]  Unknown
		2. Are the limits of the copy number completely defined? [ ]  Yes [ ]  No [ ]  Unknown
			1. First Exon affected:
			2. Last Exon affected:
			3. Whole gene affected? [ ]  Yes [ ]  No [ ]  Unknown
			4. Predicted reading frame: [ ]  In [ ]  Out [ ]  Unknown
			5. Are known gene promoters affected:[ ]  Yes [ ]  No [ ] Unknown
2. For Missense/nonsense variant or Pseudoexons:
	* 1. Was the entire coding region sequenced: [ ]  Yes [ ]  No
		2. Targeted variant analysis only: [ ]  Yes [ ]  No
			1. If YES, type of analysis: [ ]  Hot-spot [ ]  Known familial variant [ ]  Other, specify:
		3. Missense/nonsense variant location (choose one)
			1. Exon (Point Mutation):
			2. Intron:
			3. Other:
		4. Missense/nonsense variant subclass information:
			1. Insertion Deletion: [ ]  Insertion [ ]  Deletion [ ]  Insertion/Deletion
			2. Nonsense Type: [ ]  UAA [ ]  UAG [ ]  UGA [ ]  Not applicable
3. mRNA analysis
	* 1. mRNA analysis performed: [ ]  Yes [ ]  No [ ] Unknown
			1. If YES, were implications confirmed: [ ]  Yes [ ]  No
4. Variant Information (HGVS Nomenclature)
	* 1. cDNA: (if relevant, data to be entered by site)
		2. mRNA: (if relevant, data to be entered by site)
		3. Protein: (if relevant, data to be entered by site)
5. Allele Specific Information
6. Allele #2
7. Was a second disease allele identified? [ ]  Yes [ ]  No (Skip to question 30)
8. Is allele #2 identical to allele #1 (Homozygous only):[ ]  Yes (Skip to question 30) [ ]  No

If NO, repeat filling out allele specific information for Allele #2.

1. For Mitochondrial DNA variant:
	1. Quantitative analyses (Heteroplasmy assessment)

[ ]  Evaluation method

[ ]  Restriction PCR

[ ]  Deep sequencing

[ ]  Allele specific PCR

[ ]  qPCR (deletions, depletion)

[ ]  Southern blot

[ ]  Other

[ ]  Heteroplasmy level

[ ]  Blood

[ ]  Muscle

[ ]  Urinary sediment

[ ]  Buccal cells

[ ]  Other

1. **DNA Elements Table**

| **Gene (Test)** | **Test performed?** | **Variant shown?** | **Detection method, specify (Choose all that apply):** | **Pathogenic Certainty3** | **Comments** |
| --- | --- | --- | --- | --- | --- |
| *SNCA* | [ ]  Yes[ ]  No[ ]  Unknown | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |
| *LRRK2* | [ ]  Yes[ ]  No[ ]  Unknown | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |
| *GBA* | [ ]  Yes[ ]  No[ ]  Unknown | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |
| *PRKN* | [ ]  Yes[ ]  No[ ]  Unknown | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |
| *PINK1* | [ ]  Yes[ ]  No[ ]  Unknown | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |
| *DJ1* | [ ]  Yes[ ]  No[ ]  Unknown | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |
| *VPS35* | [ ]  Yes[ ]  No[ ]  Unknown | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |
| *CHCHD2* | [ ]  Yes[ ]  No[ ]  Unknown | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |
| Other, specify: | [ ]  Yes, specify test: [ ]  No  | [ ]  Yes, list and specify:Name of variant(s):[ ] Nucleotide change:[ ]  Amino acid change:[ ]  Copy number change:[ ]  No[ ]  Unknown | [ ]  Direct Sequencing (PCR)[ ]  Panel[ ]  Whole Genome Sequencing[ ]  Whole Exome Sequencing[ ]  Long Read Sequencing[ ]  TaqMan Assay[ ]  Sanger[ ]  Multiplex Ligation-dependent Probe Amplification (MLPA) [ ]  Fluorescent In-Situ Hybridization (FISH)[ ]  Other, Specify: | [ ]  Definitely Pathogenic[ ]  Probably Pathogenic[ ]  Possibly Pathogenic[ ]  Variant of Unknown Significance (VUS)[ ]  Benign[ ]  Other, Specify: |  |

General Instructions

This form contains data elements collecting information on the genetic etiology of participants with Parkinson’s disease/parkinsonism. The focus is on variables related to sample processing and genetic testing results while also capturing some information on variables related to phenotype in both the clinical and research setting.

Important note: None of the data elements included on this CRF Module are classified as Core (i.e., strongly recommended for all Parkinson’s disease clinical studies to collect). Some of the data elements are classified as Supplemental – Highly Recommended (i.e., essential information for specified conditions, study types, or designs), as indicated by asterisks below, and should be collected if genetics studies are performed.

\*\*Element is classified as Supplemental – Highly Recommended

The remaining data elements are Supplemental (i.e., non-Core) and should only be collected if the research team considers them appropriate for their study. Please see the Data Dictionary for element classifications.

Specific Instructions

Please see the Data Dictionary for definitions for each of the data elements included in this CRF Module.

References

1Question and permissible values from [Coriell Institute for Medical Research](https://www.coriell.org/1/About-Us/Legal-Notice) used and modified with permission.

2Question and permissible values from dbGaP/database of Genotypes and Phenotypes/ National Center for Biotechnology Information, National Library of Medicine (NCBI/NLM) <https://www.ncbi.nlm.nih.gov/gap> used and modified with permission.

3Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, Grody WW, Hegde M, Lyon E, Spector E, Voelkerding K, Rehm HL; ACMG Laboratory Quality Assurance Committee. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015 May;17(5):405-24.