**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

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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.