

Parkinson’s Disease Version 2.0 NINDS CDE Project Genetics Subgroup Summary

The Genetics Subgroup focused on gathering standardized key variables related to information on the genetic etiology of participants with Parkinson’s disease/parkinsonism. The focus was on variables related to family history, sample processing, and genetic testing results while also capturing some information on variables related to phenotype, in both the clinical and research setting, with the goal of enhancing data quality for use across studies and over time.

The Parkinson’s Disease (PD) v1.0 CDE recommendations did not include a genetics CRF. The subgroup reviewed existing NINDS CDEs used in other disorder recommendations, and the Coriell Institute of Medical Research and dbGaP CRFs to develop the Parkinson’s Disease Genetics CRF. The subgroup was also the primary reviewers responsible for updating the Family History CRF and secondary reviewers for the Medical History of Parkinson’s Disease CRF that the General and Motor Subgroup recommended.

The Parkinson’s Disease Genetics CRF includes six parts: clinical description, genetics summary, study description, genotype platform information, variant/mutation analysis, and the DNA elements table that gathers information on the Parkinsonism mendelian genes.

In the Family History CRF, the pedigree information was expanded upon by including more relative types, collecting the number of healthy family members and with psychiatric disorder, and the type of neurological disorder using code list options. Standardized methods were used to collect phenotypic data elements so atypical presentations along the neurodegenerative disease spectrum could be captured.

The recommendations focus mainly on PD and are not meant to address patients with prodromal Parkinson’s, as that is a clinical consideration.

Summary of Recommendations

CRF Name	Subdomain	Classification
Parkinson’s Disease Genetics	Laboratory Tests and Biospecimens/Biomarkers	Supplemental – Highly Recommended; Supplemental
Family History	General Health History	Core; Supplemental
Medical History of Parkinson’s Disease	History of Disease/Injury Event	Core; Supplemental

The Genetics Subgroup used established standards in the Genetics field within PD to develop their recommendations. The subgroup reviewed and used data elements from Coriell and dbGaP to help develop a CRF template. The Coriell form used was the Parkinsonism Clinical Data Elements Form. The dbGaP forms used were the Extramural Institutional Certification Form and the Data Submission Form for NIH Funded Studies. The subgroup also looked at Genetics CRFs that were developed for other disorders through the NINDS CDE Project. These CRFs covered genomic analysis, mutation analysis, genetic testing, clinical diagnostics, and a DNA elements table with the focus on genes.

The subgroup members did not encounter any unique concerns regarding the collection PD data. The Parkinson’s mendelian genes in the Genetics CRF do not cover related dementias or parkinsonisms. PD diagnoses can evolve, and more comprehensive screenings could be required to show these other variants. The Genetics CRF does not address these related issues.