



## Metaphenomic annotation of clinicopathological Parkinson's disorders

**Reproducible analysis code for "Refining the Diagnostic Accuracy of Parkinsonian Disorders using Metaphenomic Annotation of the Clinicopathological Literature"**

The diagnostic precision of Parkinsonian disorders is not accurate enough. Even in expert clinics up to one in five diagnoses are incorrect. Gold standard diagnosis is post-mortem confirmation of the underlying proteinopathy; however, many clinicopathological studies focus on either a single disease or frame analyses in one temporal direction that may underestimate the true extent of mis- and missed diagnoses.

This work identified 125 published clinicopathological studies between 1992 – 2022, extracted phenotype information for ~9200 post-mortem cases, and curated the data in a standardized machine-readable format. This was used to develop a probabilistic model to quantify diagnostic likelihood based on clinical observations across all Parkinsonian disorders.

This software and data will reproduce all of the original analysis, contains frameworks for reading/curating Human Phenotyping Ontology data, and implementing the multi-observation, multi-condition probabilistic framework

### Category

Software/Dataset

### Authors

Christian Lambert