



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

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