Mitochondrial endophenotypes of PD

https://neurodegenerationresearch.eu/survey/mitochondrial-endophenotypes-of-pd/

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Contact information of lead PI Country

Luxembourg

Title of project or programme

Mitochondrial endophenotypes of PD

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FNR

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Total duration of award in years

3

Keywords

Research Abstract

Parkinson's disease (PD) is caused by genetic and environmental risk factors. Mitochondrial dysfunction is thought to be a crucial pathogenetic pathway in a subgroup of patients. We hypothesize that we will identify this subgroup by discriminating a postulated 'mitochondrial endophenotype' – based on existing genomic and transcriptomic data. First-level-models will be generated from transcriptome data from patients with parkin and PINK1 mutations and corresponding animal and cellular models. These models will be used as prior information for model building in larger genomic and transcriptomic data sets, allowing to sub-classify cohorts according to predominant pathogenic pathways. As first-stage validation, mitochondrial function will be tested in patient biomaterials with predicted mitochondrial phenotypes. Results of this validation will improve model generation and will be confirmed in further patient cohorts and animal and cellular models, including patient-derived induced pluripotent stem cells

(iPSCs).Pathway-specific biomarkers including imaging biomarkers will then be developed, and a proof-of-concept clinical trial will be performed in stratified sub-cohorts.

Further information available at:

https://www.fnr.lu/projects/mitochondrial-endophenotypes-of-pd-2/

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