Implicating Novel Genes in Familial Parkinsonism

https://neurodegenerationresearch.eu/survey/implicating-novel-genes-in-familial-parkinsonism/

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Country

Canada

Title of project or programme

Implicating Novel Genes in Familial Parkinsonism

Source of funding information

CIHR

Total sum awarded (Euro)

€ 629,688

Start date of award

01/10/2012

Total duration of award in years

5.0

The project/programme is most relevant to:

Parkinson's disease & PD-related disorders

Keywords
Research Abstract
Parkinson disease (PD) is the most common neurodegenerative movement disorder affecting ~1% of the population at 65 years, and increasing to 4-5% in 85-year-olds. PD affects over 100,000 Canadians and costs $2.5 billion annually, not considering the significant personal cost to patients and their families. The current treatments for PD do not address the causes of disease, but simply manage the symptoms. The development of preventative measures is

hampered by our understanding of its molecular causes. New sequencing technologies facilitate the identification of pathogenic mutations in novel genes in familial PD. The latest susceptibility gene (VPS35) for PD was discovered by applying these methods to a Swiss family in which multiple people were affected. The finding immediately improves an early and accurate diagnosis. Moreover, each gene/mutation helps implicate biological networks in disease, provides the targets and tools to model its pathogenesis, and enables therapeutic advance. This application is to apply similar genetic screening, sequencing and genotype approaches to implicate novel genes/mutations in the pathogenesis of PD. Gene mutation discovery will initially focus on 30 families that appear to have inherited PD down the family line. During the duration of the award, additional families and population resources will be referred by an extensive network of international clinical collaborators. We hypothesize further gene discoveries will illuminate a limited number of biological pathways in disease. The proteins encoded and network perturbed provide the rational and targets for novel treatments. More efficacious drugs, which provide symptomatic benefit by halting disease progression, may cure this common disease. We intend to improve the quality of life for Canadians living with PD, and reduce the future burden of this disease on society by providing an unequivocal genetic foundation upon which translational neuroscience may focus.

Lay Summary
Further information available at:

Types:
Investments > €500k

Member States:
Canada

Diseases:
Parkinson's disease & PD-related disorders

Years:
2016

Database Categories:
N/A

Database Tags:
N/A