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FamPD: Identification of new genes causing familial forms of PD

Project Description

Parkinson's disease (PD) is a common and severe neurodegenerative disease that leads to progressive motor impairment as well as autonomic and cognitive disturbances. In recent years, undoubted significant and rapid progress in our knowledge of the causes of PD has been made by in depth analysis of relatively rare forms of inherited PD that share many clinical and pathologic characteristics with the common sporadic form of the disease. These discoveries have provided a starting point and have inspired intense research work in many laboratories world-wide to unravel the molecular and cellular events that lead to the dysfunction and death of dopamine cells, with the aim to identify novel targets for causative or even preventive treatments. Despite this significant progress, it is assumed that many important genes that may cause or contribute to the development of PD still remain to be discovered.

This proposal seeks to take a coordinated approach to the genetic analysis of a large number of families with PD which have been identified by the three applying groups over several years, in whom no defects in the currently known genes has been detected. These families belong to different groups, including those with autosomal-dominant and autosomal-recessive inheritance. Accordingly, different approaches are suggested to identify the underlying genetic defects. Recent advances in the technologies have meant that performing genome wide searches for genetic variants in a large number of patients and families is now rapid, robust and feasible. This project is focused on new genetic discoveries. We anticipate that the finding of new genes will focus future research efforts into new pathways and ultimately provide the field with potential targets for therapeutic interventions.

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