

## PD GEN Study

PD GEN is a DNA databank for people with Parkinson's participating in the large randomised controlled trials PD MED, PD SURG, PD REHAB & PD COMM.

PD GEN is still open to recruitment.

### Design

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PD GEN is a DNA databank for patients with Parkinson's, from the large randomised controlled trials PD MED, PD SURG, PD REHAB & PD COMM.

### Aim of Study

To distribute genetics samples and their associated anonymised epidemiological and research data, from patients with Parkinson's, to approved researchers working in the field and to help understanding of the genetic factors involved in Parkinson's disease.

### Setting

In approximately 60 elderly care and neurology units (throughout the UK to reflect population diversity) that have recruited patients into one of the following studies: PD MED, PD SURG, PD REHAB & PD COMM.

### Target population

Patients with PD of any age that been recruited into PD MED, PD SURG, PD REHAB or PD COMM.

### What the samples are being used for so far

The first significant use of the PD GEN samples was when 600 of them were screened as part of the 2100 samples in the first published UK genome-wide association study (GWAS) in PD which confirmed the importance of genetic variants in SNCA (alpha-synuclein) and MAPT<sup>1</sup>. These data, along with GWAS data in a further 500 of the PD GEN samples, were then included in a meta-analysis which led to the identification of a further five new loci associated with the risk of developing PD<sup>2,3</sup>.

Our current work with the UK PD Genetics Consortium involves further genetic analysis of genetic variants in immune regulatory genes, detailed genetic study of genes specifically targeted based on our previous large GWAS and studies of exome sequencing in the disease (funded by Parkinson's UK).

During the last 2 years we have also joined, as the only UK member, the international Genetic Epidemiology of Parkinson's Disease Consortium (GEO-PD), which aims to investigate both genetic and environmental influences in PD. This is particularly pertinent to our PD GEN collection, as we collect retrospective epidemiological data along with the DNA samples.

We have supplied DNA samples, genotypic and epidemiological data to several large scale studies with investigators from the US, Europe and Asia including studies of 'Alpha-synuclein reduced expression genotypes and survival in PD' (manuscript under review), large scale GWAS studies in PD using non-genetic factors as risk modifiers<sup>4</sup>, studies of rare coding variants in susceptibility to PD (work currently underway) and studies of the role of VPS35 mutations in PD (underway).

Recent work in house on the PD GEN samples has included studies of mendelian PD genes in early onset PD<sup>5</sup> and studies of the influence of variants in cell cycle regulation genes in the development of cognitive impairment in PD with Dr Z Nagy, following on from her work on the influence of these polymorphisms in the development of Alzheimer's disease. We are now seeking funding for large scale pharmacogenomic work on the PD GEN samples.

(see [publications \(/research/activity/mds/trials/bctu/trials/pd/pdgen/publications.aspx\)](#) page for references)