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MOLECULAR GENETIC STUDIES OF NEURODEGENERATIVE DISEASE

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Introduction Large scale genetic studies such as genome-wide association studies (GWAS) in Parkinson's disease (PD) have revealed genetic susceptibility factors and continue to offer new insights both into the genetics of sporadic disease and its pathogenesis, with the potential for identification of an at-risk population and novel therapeutic targets. However, the methodology importantly requires larger data sets for replication of novel findings.

Aim To develop an independent UK-based PD DNA bank to act as a national and international resource for large-scale genetic studies.

Methods The study was initiated as part of an MRC-funded fellowship in 2009 and was adopted onto the NIHR portfolio in 2010. Patients were included if they had a clinical diagnosis of PD. Section 30 (Mental Capacity Act 2005) approval was also obtained to facilitate recruitment of those with diminished capacity. Patients were excluded if they had previously donated to another national DNA collection, such as PD GEN. Spouse/carer controls were also recruited. 37 Centres from across the UK took part in the study. DNA was extracted and stored at Royal Devon and Exeter Hospital, Devon.

Results To date over 1370 patients and 530 controls have been recruited to the study. The population characteristics of patients are: 61.1% male, 98.1% white, 18.6% positive family history,

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mean age 70.6 yrs (range 40–98), mean disease duration 5.6 yrs (range 0–49).

Conclusions We have established an independent PD DNA Bank which will have reached its target of 1500 patient samples by April 2013. This sample bank will be similar in size and composition to other national collections such as PD GEN and will act as a valuable resource locally, nationally and for the global PD research community. Its establishment within 3 years is a testimony to the efficient working of the clinical research networks within the UK.



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