

## Genetic Testing for Parkinsons Disease

### Contact Details

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### Samples Required

2-4ml blood in EDTA  
If sending DNA, please indicate the concentration

All samples must be accompanied by a FULLY completed request form. Please include details of testing required, family history and referring clinician. Failure to provide this information will result in a delay in testing.

Urgent testing **MUST** be arranged with the laboratory in advance.

### Consent and DNA Storage

It is the responsibility of the referring clinician to ensure that appropriate consent for genetic testing has been obtained.

Unless otherwise requested, following testing, all DNA samples will be stored in the laboratory.

Stored DNA samples may be used anonymously for quality assurance purposes and for developing new tests for the disorder in question. Unless otherwise requested, DNA samples will be tested and reported when new tests are developed.

### Background

Parkinson's disease (PD) is characterized by tremor, muscle rigidity and bradykinesia. Psychiatric manifestations are common and include depression and visual hallucinations. Dementia eventually occurs in at least 20% of cases. PD is classified by age of clinical symptoms; *Juvenile* PD before age 20, *Early-Onset* before age 45 and individuals with symptoms after 65 years of age are classed as *Late-Onset* PD. Although most cases are multi-factorial, PD can be inherited in an autosomal dominant or autosomal recessive manner. To date, 6 genes have been implicated in inherited PD, with mutations in the LRRK2 and PARK2 genes responsible for the majority of cases of dominant and recessive cases respectively.

### Genetic testing offered

For affected individuals:

- Complete screen (12 exons) of PARK2 gene
- Screen of the p.Gly2019Ser mutation in LRRK2

Pre-symptomatic testing for the familial mutation in at-risk relatives

### Accepted Referrers

- Consultant who is a sub-specialist in Movement Disorders
- Consultant Clinical Genetics

### Appropriate Referrals

To refine testing and aid interpretation, please provide the following information:

#### For PARK2 testing

- |  |        |
|--|--------|
| ○ PD onset before age 40 with or without a family history                                  | Yes/No |
| ○ PD onset before age 45 with prominent dystonia at onset with or without a family history | Yes/No |
| ○ PD onset before age 50 with positive family history or consanguineous marriage           | Yes/No |

#### For LRRK2 p.Gly2019Ser testing

- |   |        |
|---|--------|
| ○ Patient with any age of onset who has dominant family history of PD   | Yes/No |
| ○ Patient with a positive family history of PD regardless of inheritance pattern with onset before age 50 and who is PARK2 negative | Yes/No |

### Predictive testing

Referrals will only be accepted for patients who have been counseled in a Genetics Clinic. Referrals for predictive testing must include details of the familial mutation and/or the name and d.o.b of the index case.

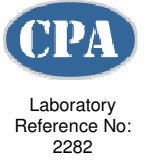
### Reporting time and cost

<u>Testing</u>	<u>Reporting time (working days)</u>	<u>Cost</u>
Sequence analysis of PARK2	40	£312
Deletion testing by MLPA	20	£143
Screen for LRRK2 p.Gly2019Ser	20	£130
Predictive testing for familial mutation	10	£130

**NB: Referrals from Scotland do not incur a charge**



EAST OF SCOTLAND REGIONAL GENETIC SERVICE



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THE FOLLOWING CLINICAL INFORMATION MUST BE PROVIDED FOR EACH SAMPLE:

Age of onset of symptoms: \_\_\_\_ years

Clinical Features (including any imaging results)

Dystonia present ? - Yes/No

Intact smell ? - Yes/No

Cognitive deficits ? - Yes/No

Family History (if none then please state the age of both parents)

SIGNATURE OF PHYSICIAN

I confirm that I have explained the nature of the genetic test to the patient and the implications of any result that is obtained:

..... DATE .....

SIGNATURE OF PATIENT

I confirm that I have understood the reasons for the genetic test above and give my consent for genetic analysis:

..... DATE .....