

Frontotemporal dementia with Parkinsonism (FTDP-17)

OMIM 600274

Genes: MAPT; PGRN

Locus: 17q21.1

OMIM: 157140; 138945

SERVICE: mutation analysis of hotspot exons of the MAPT (microtubule associated protein tau) gene
mutation analysis of the PGRN (progranulin) gene

TESTING: Diagnostic: clinically affected patients
Presymptomatic: at risk relatives of clinically affected patients (known mutation)

*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: from Consultant Clinical Geneticists and Consultant Neurologists only

TARGET REPORTING TIME AND COSTS

(Non NHS patients are subject to 20% surcharge and payment must be agreed prior to testing)

Diagnostic:	8 weeks	£432 (sequence MAPT hotspot exons) £599 (sequence PGRN)
Presymptomatic:	2 weeks	£195 (sequence one exon)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1, 9-13 and splice site boundaries of the MAPT gene
- PCR and fluorescent sequence analysis of exons 0 (untranslated), 1-12 and splice site boundaries of the PGRN gene

SAMPLE REQUIREMENTS

- 5ml blood in EDTA or 50ul DNA
- All patient samples must be labelled with **name, date of birth and Hospital/NHS number**

Samples should be accompanied by a FULLY completed request card (available from the laboratory). Please include details of test, family history, patient address & postcode, GP, referring clinician and unit/hospital.

CONSENT

It is the responsibility of the referring clinician to ensure consent has been obtained for:

- testing and storage
- the use of the sample and the information generated from it to be shared with members of the patients family and their health professionals

After testing, part of this sample might be used anonymously for the development of new tests and to monitor the quality of laboratory results.

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UKGTN 'Testing criteria' template

Patient name:

Patient address:

Name of referrer:

Title/Position:

Department/Hospital:

Name of Disease/test:

Frontotemporal dementia with Parkinsonism-17

Referrals only will be accepted from one of the following:

(Please indicate with a tick which category refers to the referrer).

Referrer	Tick if this refers to you.
Consultant Clinical Geneticist	
Consultant Neurologist	

Minimum criteria required for testing to be appropriate as stated in the Gene Dossier:

Criteria	Tick if this patient meets criteria
Frontotemporal dementia and/or parkinsonism	
Frontal and/or temporal atrophy on neuroimaging	
Family history of dementia and/or parkinsonism in two or more first degree relatives consistent with an autosomal dominant mode of inheritance	
Neuropathology showing intraneuronal deposition of hyperphosphorylated tau protein demonstrated by phosphorylation-dependent anti-tau antibodies. Tau pathology consists of abundant neurofibrillary tangles in the hippocampus, cortical regions, and subcortical nuclei in some individuals and pretangle tau deposits in neurons and glial cells in most individuals.	

If the sample does not fulfil these criteria and you still feel that testing should be performed please contact the molecular genetics laboratory