

Frontotemporal dementia with Parkinsonism (FTDP-17) OMIM: 600274
Gene: MAPT Locus: 17q21.1 OMIM: 157140

SERVICE: mutation and dosage analysis of the MAPT (microtubule associated protein tau) gene

TESTING: **Diagnostic*:** clinically affected patients
Presymptomatic#: patients at risk of developing FTDP (known mutation)
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Clinical Geneticists and Neurologists only**
#Presymptomatic testing is only accepted from a Consultant Clinical Geneticist
The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS

(Non UK National Health Service patients are subject to a surcharge. Payment must be agreed prior to testing – please include invoice form A)

Diagnostic:	8 weeks	£415 (sequencing + dosage)
Presymptomatic:	2 weeks	£145

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-13 (excluding exons 4a, 6 and 8 which are not usually transcribed in the six major brain isoforms) and splice site boundaries of the MAPT gene
 - Multiplex ligation dependent probe amplification analysis of all exons
- Between 25% and 40% of families with autosomal dominant frontotemporal dementia show mutations in MAPT

SAMPLE REQUIREMENTS

- 1-5ml blood in EDTA or 50ul DNA (concentration ~500ng/ul)
- 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
- **Samples and paperwork must include three unique and matching patient identifiers**

SHIPPING DETAILS

- DNA can be sent by first class post
- Blood must be appropriately packaged and preferably sent by courier to arrive as soon as possible.
- Do not freeze prior or during postage.

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.

CONTACT DETAILS

Genetics Laboratories, Box 143
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Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

Name of disease(s):	Frontotemporal dementia with Parkinsonism-17
Name of gene(s):	MAPT (microtubule associated protein tau)

Patient name:	Date of birth:
Patient postcode:	NHS number:
Name of referrer:	
Title/Position:	
Department/Hospital:	
Contact email/telephone number:	

Referrals will only 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	<input type="checkbox"/>
Consultant Neurologist	<input type="checkbox"/>

Minimum criteria required for testing to be appropriate:

Criteria	Tick if this patient meets criteria
Frontotemporal dementia and/or parkinsonism AND	<input type="checkbox"/>
Frontal and/or temporal atrophy on neuroimaging AND	<input type="checkbox"/>
Family history of dementia and/or parkinsonism in two or more first degree relatives consistent with an autosomal dominant mode of inheritance OR	<input type="checkbox"/>
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 OR	<input type="checkbox"/>
At risk family member with known mutation	<input type="checkbox"/>

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