

Early Onset Familial Alzheimer Disease **OMIM: 104300**
Gene: APP **Locus: 21q21** **OMIM: 104760**

SERVICE: **mutation analysis of exons 16 and 17 of the APP gene**

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

REFERRALS: **from Clinical Geneticists and Consultant Neurologists only**
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	£175 (sequence + dosage)
Presymptomatic:	2 weeks	£145

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 16 and 17 and splice site boundaries
- Multiplex ligation dependent probe amplification analysis of exons 1, 2, 3, 4, 6, 7, 9, 12, 14, 16 and 18

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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Cambridge CB2 0QQ
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Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

Name of disease(s):	Early onset Familial Alzheimer disease (EOFAD)
Name of gene(s):	APP

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
Adult early onset slowly progressive dementia (<65 years at presentation), PS1, FTDP-17 & PGRN negative AND	<input type="checkbox"/>
Absence of other causes of dementia AND	<input type="checkbox"/>
Cerebral cortical atrophy by neuroimaging studies AND	<input type="checkbox"/>
More than one family member with early onset AD in which the age of onset is below 60 years	<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