

Familial dysalbuminaemic hyperthyroxinaemia (FDH) OMIM: 103600
Genes: albumin (ALB) Loci: 4q11-q13 OMIM: 103600

SERVICE: mutation analysis of the albumin gene (exon 7 only)

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

REFERRALS: **Clinical Geneticists, Endocrinologists and Chemical pathologists**
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	£100
Presymptomatic:	2 weeks	£145

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis for ALB common mutations: p.Arg242His and p.Arg242Pro
- Majority of FDH cases are due to common ALB mutations: p.Arg242His and p.Arg242Pro

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:	Familial Dysalbuminemic Hyperthyroxinemia (FDH);
Name of Gene:	Albumin (ALB)

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 Endocrinologist	<input type="checkbox"/>
Consultant Chemical Pathologist	<input type="checkbox"/>

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

Criteria	Tick if this patient meets criteria
1. Family history of FDH	<input type="checkbox"/>
2. Raised serum fT4 or fT3 with inappropriately non-suppressed serum TSH AND	<input type="checkbox"/>
3. Attempts to exclude assay interference as a cause of the abnormal thyroid function tests should be made where possible	<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