

Dysthyrotropinaemic hyperthyroxinaemia:

Thyroid Hormone Resistance (THR); Familial Dysalbuminemic Hyperthyroxinemia (FDH);
OMIM 229600; 103600

Genes: TRbeta; albumin (ALB); Loci: 3p24.3; 4q11-q13; OMIM: 190160; 103600

SERVICE: mutation analysis of the TRbeta gene
mutation analysis of the albumin gene (exons 3 and 7 only)

TESTING: Diagnostic*: clinically affected patients
Presymptomatic: relatives of clinically affected patients (known TRbeta/ALB mutation)

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

REFERRALS: from Consultant Clinical Geneticists and Consultant Endocrinologists

TARGET REPORTING TIME AND COSTS

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

Diagnostic:	8 weeks	£75 (TRbeta)
		£50 (ALB)
Presymptomatic:	2 weeks	£75 (TRbeta)
		£50 (ALB)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 7-10 and splice site boundaries of the TRbeta gene
- PCR and fluorescent sequence analysis for ALB common mutations: p.Leu90Pro, p.Arg242His, p.Arg242Pro

All reported mutations of the TRbeta have been detected in the hormone-binding domain (exons 7, 8, 9 and 10)
Majority of FDH cases are due to common ALB mutations: p.Leu90Pro, p.Arg242His and p.Arg242Pro

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:

Dysthyrotophinaemic hyperthyroxinaemia

Thyroid Hormone Resistance (THR);

Dysalbuminemic Hyperthyroxinemia (FDH);

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 Endocrinologist	
Consultant Chemical Pathologist	

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

Criteria	Tick if this patient meets criteria
Family history of thyroid hormone resistance or FDH	
Raised serum fT4 or fT3 with inappropriately non-suppressed serum TSH	
Attempts to exclude assay interference as a cause of the abnormal thyroid function tests should be made where possible	

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