

Hereditary Fructose Intolerance (HFI)

OMIM 229600

Gene: Aldolase B

Locus: 9q22.3

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SERVICE: mutation analysis of the aldolase B gene

TESTING: Diagnostic*: clinically affected patients
Carrier: relatives of clinically affected patients (known ALDOB mutation)
Prenatal: at risk of having an affected child (known ALDOB mutation)

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

REFERRALS: from Consultant Gastroenterologists and Consultant Paediatricians

TARGET REPORTING TIME AND COSTS

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

Diagnostic:	8 weeks	£267	(3 common mutations only)
		£533	(sequencing exons 2-9)
Carrier:	2 weeks	£195	(sequence one exon)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis for common mutations: p.Ala150Pro, p.Ala175Asp, p.Asn335Lys
- PCR and fluorescent sequence analysis of exons 2-9 and splice-site boundaries of the ALDOB gene

Approximately 85% of mutations in Europe are due to common mutations: p.Ala150Pro, p.Ala175Asp, p.Asn335Lys

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:

Hereditary Fructose Intolerance;

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 Paediatricians	
Consultant Gastroenterologist	
Metabolic Medicine Consultant	

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

Criteria	Tick if this patient meets criteria
Family history of fructose intolerance	
Symptoms of fructose intolerance	

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