

**Factor XIII deficiency**      **OMIM: 613225**  
**Gene: F13A1**      **Locus: 6p25-p24**      **OMIM: 134570**

**SERVICE:**      **Mutation analysis of the *F13A1* gene**

**TESTING:**      **Diagnostic\*:**      **clinically affected patients or FXIII level below reference range**  
**Carrier:**      **relatives of affected patients (known *F13A1* mutation)**  
\*samples will only be accepted with a completed 'testing criteria' form (see attached)

**REFERRALS:**      **Consultant Haematologist, Haemophilia Nurse and Clinical Geneticist 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)

<b>Diagnostic:</b>	<b>8 weeks</b>	<b>£415 (sequence)</b>
<b>Familial mutation (s):</b>	<b>2 weeks</b>	<b>£145-£175</b>
<b>Prenatal:</b>	<b>3 days</b>	<b>£350 (including maternal contamination studies)</b>

## TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 2-15 and splice site boundaries of the *F13A1* gene

## 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 requested, the FXIII levels of the patient, 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.

## 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  
Level 6, Addenbrooke's Treatment Centre  
Addenbrooke's Hospital  
Cambridge CB2 0QQ  
Tel: +44 (0) 1223 348866  
Fax: +44 (0) 1223 348870  
Email: [becky.treacy@addenbrookes.nhs.uk](mailto:becky.treacy@addenbrookes.nhs.uk)

Website: [www.cuh.org.uk/genetics-labs](http://www.cuh.org.uk/genetics-labs)



**Accredited Medical Laboratory**  
**Reference No: 1275**

**Cambridge testing criteria**

UK Genetic Testing Network

Name of disease(s): **Factor XIII deficiency**Name of gene(s): **F13A1**

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.
<b>Consultant Haematologist</b>	
<b>Haemophilia Nurses</b>	
<b>Clinical Geneticist</b>	

**Minimum criteria required for testing to be appropriate:**

Criteria	Tick if this patient meets criteria
<b>Low FXIII levels (e.g. less than approx 0.5 IU/dl) on two or more separate occasions OR</b> (Please provide FXIII levels with referral)	
<b>Obligate Carrier e.g. parent of child with severe (homozygous/compound heterozygous) FXIII deficiency</b> (please provide details of familial mutation if known) OR	
<b>At risk family members (e.g. FH of FXIII deficiency) with known mutation</b> (Please provide details of familial mutation )	

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