

Factor XI (FXI) deficiency

Gene: F11

Locus: 4q35

OMIM: 612416

OMIM: 264900

SERVICE: mutation analysis of the F11 gene

TESTING: **Diagnostic*:** clinically affected patients or FXI level below reference range
Predictive: relatives of affected patients (known F11 mutation)
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: from Consultant Haematologist, Haemophilia Nurses and Haemophilia Centre
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	£415 (sequencing)
Familial mutation (s):	2 weeks	£145-£175

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 2-15 and splice site boundaries of the F11 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, FXI levels, 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
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

Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

Cambridge testing criteria

Name of disease(s): **Factor XI (FXI) Deficiency**

Name of gene(s): **F11**

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 Haematologist	
Specialist Haemophilia Nurse	
Clinical Geneticist	

Minimum criteria required for testing to be appropriate:

Criteria	Tick if this patient meets criteria
Low FXI levels (e.g. less than approx 0.7 IU/dl) on two or more separate occasions OR (Please provide FXI levels with referral)	
Obligate Carrier e.g. parent of child with severe (homozygous/compound heterozygous) FXI deficiency (please provide details of familial mutation if known) OR	
At risk family members (e.g. FH of FXI deficiency) with known mutation (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