

Familial Dysautonomia (Hereditary Sensory and autonomic Neuropathy Type III)

OMIM: 223900

Gene: IKBKAP

Loci: 9q31

OMIM: 603722

SERVICE: mutation analysis of the IKBKAP gene

TESTING: **Diagnostic*:** clinically affected patients
Carrier: relatives of clinically affected patients (known mutation)
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Consultant Clinical Geneticists, Paediatric neurologists and neurologists only**
The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS

(Patients outside of the UK National Health Service are subject to 20% surcharge and payment must be agreed prior to testing)

Diagnostic:	2 weeks	£145 (test for common mutation only)
	8 weeks	£625 (sequence)
Carrier:	2 weeks	£145 (sequence single exon)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 2-37 and splice site boundaries
- c.2204+6T>C mutation in intron 20 is responsible for virtually all cases of FD in Ashkenazi Jews

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
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): **Familial Dysautonomia**

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

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	
Neurologist and Paediatric neurologist	

Minimum criteria required for testing to be appropriate:

Criteria	Tick if this patient meets criteria
If not of Ashkenazi Jewish descent, exclude alternative diagnosis first	
Autonomic features predominate and Axelrod's 1983 5 signs present	
Inheritance pattern compatible with recessive	

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