

Incontinentia pigmenti type II (IP) **OMIM: 308300**
Gene: IKBKG (NEMO) **Locus: Xq28** **OMIM: 300248**

SERVICE: **common deletion and sequencing analysis of the IKBKG (NEMO) gene**

TESTING: **Diagnostic:** **clinically affected females (usually lethal in males)**
 Presymptomatic: **female patients at risk of developing IP**
 Prenatal: **females at risk of having an affected child (common deletion)**
 *samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Clinical Geneticists and Consultant Paediatricians 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)

Diagnostic:	2 weeks	£145 common deletion
	8 weeks	£295 sequencing
Carrier:	2 weeks	£145
Prenatal:	3 days	£350 (including maternal contamination studies)

TECHNICAL INFORMATION

- PCR across the common deletion of exons 4-10, using IKBKG (NEMO) and deltaNEMO pseudogene specific primers
- Sequencing analysis of exons 2-10 of IKBKG (NEMO) and the highly homologous deltaNEMO pseudogene
- RNA studies are under development to confirm mutations found by sequencing

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): **Incontinentia pigmenti type II, Bloch-Sulzberger Syndrome**

Name of gene(s): **NEMO (NF-KB Essential Modulator)**

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	
Consultant Paediatrician	

Minimum criteria required for testing to be appropriate:

Criteria	Tick if this patient meets criteria
<p>1. One or other of following dermatological signs in a female</p> <ul style="list-style-type: none">• Typical neonatal erythema and blistering +/- warty papules and plaques• Hyperpigmentation consistent with lines of Blaschko• Linear, atrophic hairless scarring in an adult with	
<p>2. One or other of the following</p> <ul style="list-style-type: none">• Incomplete dentition (hypo or anodontia) and malformed crowns• Nail dystrophy• Patchy alopecia• Retinal dysplasia• History of male miscarriages• X-linked family history of either dermatological features or signs listed above• Characteristic IP histology on skin biopsy e.g. eosinophil filled blisters	
<p>3. Male case satisfying the above with an XXY karyotype</p>	

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