

Incontinentia pigmenti type II (IP)

OMIM 308300

Gene: NEMO (NF-KB Essential Modulator)

Locus: Xq28

OMIM: 300248

SERVICE: common deletion of the NEMO gene only

TESTING: Diagnostic: clinically affected females (usually lethal in males)
Presymptomatic: female patients at risk of developing IP (familial NEMO deletion)
Prenatal: females at risk of having an affected child (familial NEMO deletion)

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

REFERRALS: from Consultant Clinical Geneticists and Consultant Paediatricians only

TARGET REPORTING TIME AND COSTS

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

Diagnostic:	8 weeks	£133
Presymptomatic:	2 weeks	£133
Prenatal:	2 weeks	£555

TECHNICAL INFORMATION

- PCR across the common deletion of exons 4-10, using NEMO and deltaNEMO pseudogene specific primers

80% of IP patients have a deletion of exons 4 to 10 of the NEMO gene

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:

Incontinentia pigmenti type II, Bloch-Sulzberger Syndrome

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 Paediatric Neurologist	

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

Criteria	Tick if this patient meets criteria
1. One or other of following dermatological signs in a female <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	
2. One or other of the following <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	
3. Male case satisfying the above with an XXY karyotype	

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