

**Congenital Insensitivity to pain,  
Congenital Erythermalgia (excess pain), Paroxysmal Pain disorder (excess pain)**  
**OMIM: 224300, 133020, 16740**  
**Gene: SCN9A Loci: 2q24 OMIM: 603415**

**SERVICE:** mutation analysis of the SCN9A 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

(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>£625 (sequence)</b>
<b>Carrier:</b>	<b>2 weeks</b>	<b>£145 (sequence single exon)</b>

### TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-26 and splice site boundaries

### 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  
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Addenbrooke's Hospital  
Cambridge CB2 0QQ  
Tel: +44 (0) 1223 348866  
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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**

## UKGTN testing criteria

*UK Genetic Testing Network*

<b>Name of disease(s):</b>	<b>Congenital Indifference to Pain Congenital Erythermalgia (excess pain) Paroxysmal Pain disorder (excess pain)</b>
<b>Name of gene(s):</b>	<b>SCN9A</b>

<b>Patient name:</b>	<b>Date of birth:</b>
<b>Patient postcode:</b>	<b>NHS number:</b>
<b>Name of referrer:</b>	
<b>Title/Position:</b>	
<b>Department/Hospital:</b>	
<b>Contact email/telephone number:</b>	

**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 Clinical Geneticist</b>	
<b>Paediatric neurologist</b>	
<b>Neurologist</b>	

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

Criteria	Tick if this patient meets criteria
<b>No pain ever felt OR Excess paroxysmal severe pain AND</b>	
<b>Normal cognition</b>	
<b>Normal neurological examination</b>	

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