

**Congenital non-goitrous hypothyroidism type 1 (CHNG1) OMIM: 275200**  
**Gene: TSHR Locus: 14q31 OMIM: 603372**

**SERVICE:** mutation and dosage analysis of the thyroid-stimulation hormone receptor (TSHR) gene

**TESTING:** **Diagnostic\*:** clinically affected patients  
**Presymptomatic:** patients at risk of developing CHNG1  
**Carrier:** relatives of clinically affected patients (known mutation)  
\*samples will only be accepted with a completed 'testing criteria' form (see attached)

**REFERRALS:** **Clinical Geneticists, Endocrinologists and 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)

<b>Diagnostic:</b>	<b>8 weeks</b>	<b>£415 (sequencing + dosage)</b>
<b>Carrier:</b>	<b>2 weeks</b>	<b>£145</b>

## TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-10 and splice site boundaries of the TSHR gene
- Multiplex ligation dependent probe amplification analysis of exons 1-10 of the TSHR 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, 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](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

Name of disease(s):	<b>Congenital nongoitrous hypothyroidism type 1 (CHNG1)</b>
Name of gene(s):	<b>Thyroid-stimulating hormone receptor (TSHR)</b>

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
<b>Consultant Paediatrician</b>	<input type="checkbox"/>
<b>Consultant Clinical Geneticist</b>	<input type="checkbox"/>
<b>Consultant Endocrinologist</b>	<input type="checkbox"/>

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

Criteria (all required)	Tick if this patient meets criteria
<ul style="list-style-type: none"> <li>• <b>Clinically euthyroid children with increased serum TSH, but normal free thyroxine concentrations OR</b></li> <li>• <b>Infants identified with congenital hypothyroidism by newborn screening who respond poorly to thyroxine treatment</b></li> </ul>	<input type="checkbox"/>
<ul style="list-style-type: none"> <li>• <b>Patients with persistently increased serum TSH concentrations in whom autoimmune thyroid disease has been excluded AND</b></li> <li>• <b>who have a family history of increased serum TSH in first degree relatives</b></li> </ul>	<input type="checkbox"/>

Recommend excluding TSH assay interference prior to genotyping in patients without a strong family history. Please contact lab for further details.

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