

Tuberous sclerosis complex (TSC) **OMIM: 191100**
Gene: TSC1; TSC2 **Locus: 9q34; 16p13.3** **OMIM: 605284; 191092**

SERVICE: **mutation and dosage analysis of the TSC1 and TSC2 genes**

TESTING: **Diagnostic*:** **clinically affected patients**
Presymptomatic: **patients at risk of developing TSC (known TSC1/TSC2 mutation)**
Prenatal: **at risk of having an affected child (known TSC1/TSC2 mutation)**
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Clinical Geneticist and Paediatric Neurologist**
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:	8 weeks	£850 (sequencing + dosage of both genes)
Presymptomatic:	2 weeks	£145
Prenatal:	3 days	£350 (including maternal contamination studies)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 3-23 and splice site boundaries of the TSC1 gene
 - PCR and fluorescent sequence analysis of exons 1-41 and splice site boundaries of the TSC2 gene
 - Multiplex ligation dependent probe amplification analysis of all exons
- Current literature indicates that approximately 80% of individuals with TSC would be expected to have a mutation identified by the methods that we have used (sequence analysis and MLPA)

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): **Tuberous sclerosis complex (TSC)**

Name of gene(s): **TSC1, TSC2**

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 Paediatric Neurologist	

Minimum criteria required for testing to be appropriate:

One or more of the following criteria	Tick if this patient meets criteria
• Family History of TS in 1 degree relative	
• Facial angiofibroma or forehead plaques	
• Periungual fibroma	
• Hypomelanotic maculars (>3)	
• Shagreen patch	
• Cortical tuber	
• Subependymal nodule	
• Subependymal giant cell astrocytoma	
• Multile retinal nodular hamartomas	
• Cardiac rhabdomyoma	
• Lymphangiomyomatosis	
• Renal angiomyolipoma	

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