

Tuberous sclerosis (TSC)

OMIM 191100

Genes: 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)

REFERRALS: from Consultant Physicians 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 | £1200 (sequencing + MLPA of both genes) |
| Presymptomatic: | 2 weeks | £195 (sequence one exon) |
| | | £157 (dosage analysis) |
| Prenatal: | 3 days | £555 |

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

Sequencing analysis detects all reported TSC1 mutations and 85% TSC2 mutations. The remainder of TSC2 cases are due to large rearrangements of the TSC2 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 postcode:

Name of referrer:

Title/Position:

Name of Disease/test:

Tuberous Sclerosis

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 | |
|--|----------------------------|
| 1. One or more of the following criteria: | Please tick feature |
| • 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 Cambridge Regional Genetics Laboratory (01223 348866) to discuss testing of the sample.