Tuberous Sclerosis Complex (*TSC1 & TSC2*) – OMIM 191100/613254

Background

Tuberous Sclerosis Complex (TSC) is a complex genetic condition caused by an alteration in either the *TSC1* (OMIM 605284) or *TSC2* (OMIM 191092) gene. People with TSC have growths (tubers or lesions) in different organs of the body (brain, heart, eyes, skin, kidneys, lungs) and may have epilepsy, learning disabilities, autism spectrum disorder and kidney problems. Symptoms vary from one person to another ranging from very few symptoms to being more severely affected. Diagnosis can occur at any time, from before birth to any time during adulthood, depending upon the symptoms present. TSC has a dominant inheritance pattern so there is a 50% chance of an affected individual having a child who also has the condition. TSC affects around 1 in 6000 people, although there are many undiagnosed cases due to the obscurity of the disease and the mildness of the symptoms in some cases.

Recommended Clinical Referral Criteria

- Family history of TSC
- Patients with a clinical diagnosis of TSC based on known symptoms

Molecular Analysis

Next Generation Sequencing (NGS): *TSC1* and *TSC2* genes are enriched using an Illumina TruSight Cancer assay and sequenced on an Illumina NextSeq. Any gaps will be filled with Sanger sequencing. **Dosage analysis:** Analysis of *TSC1* and *TSC2* genes by NGS or MLPA A variant is detected in 75-90% of cases with a clinical diagnosis of TS
Family follow-up: Testing for known familial pathogenic variants in *TSC1* and *TSC2* genes

Turnaround Times (TAT)

Prices available on request, please contact the laboratory using details below.

Test	TAT (calendar days)
Testing for known familial variant in *TSC1* and *TSC2*	3 – prenatal 28 - priority 42 - routine
Next generation sequencing and dosage analysis (*TSC1* and *TSC2*)	42

Contact Details

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Sample Requirements

Blood: 5ml in EDTA (1ml neonates/infants).
Please contact lab prior to sending a prenatal sample.

Please label samples with three identifiers and date of collection
All samples must be accompanied by request form
Consent for testing & DNA storage is assumed when request for test received

Links

Orphanet
http://www.orpha.net/
OMIM
http://www.omim.org/
Genetic Test Registry
http://www.ncbi.nlm.nih.gov/gtr/
Support
Tuberous Sclerosis Association UK www.tuberous-sclerosis.org