
Lab Dept: Anatomic Pathology

Test Name: TUBEROUS SCLEROSIS (TSC1) KNOWN MUTATION

General Information

Lab Order Codes: TSC1K

Synonyms: TSC Familial Mutation Evaluation (Known Mutation Analysis)

CPT Codes: 81403 – Known familial variant, not otherwise specified, for gene listed in Tier 1 or Tier 2, DNA sequence analysis, each variant exon

Test Includes: Detects a single sequence variation in TSC1 based on proband's mutation

Logistics

Test Indications: Any combination of seizures, developmental delay, skin lesions, cortical tubers and hamartomas in various parts of the body.

Family member testing to be used when the proband's mutation has been established by previous testing.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: University of Alabama Medical Genomics Laboratory

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours

Turnaround Time: Results in 2 – 5 weeks

Special Instructions: Restricted draw times, see Test availability. A completed [requisition form](#) and informed consent with a phenotypic checklist must accompany each sample. For questions regarding the forms, please call 1-800-499-4363.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 1 mL blood

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| Processed Volume: | Same as Draw Volume |
| Collection: | Routine venipuncture for blood specimens, invert gently to mix |
| Special Processing: | Lab Staff: Do Not centrifuge. Specimen must remain in original collection container. Ship specimen ASAP, with proper forms, at ambient temperature via overnight courier. Blood must reach the U of Alabama within 60 hours of draw. |
| Patient Preparation: | None |
| Sample Rejection: | Mislabeled or unlabeled specimens; frozen specimens |

Interpretive

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| Reference Range: | Interpretive report |
| Critical Values: | N/A |
| Limitations: | N/A |
| Methodology: | Sequence analysis |
| References: | University of Alabama Medical Genomics Laboratory April 2014 (800) 499-4363 Fax (205) 996-2929 |
| Updates: | 2/11/2013: CPT update 3/5/2013: Test moved from Boston University to Prevention Genetics 4/15/2014: Moved from Prevention Genetics to University of Alabama. |