
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 x1 – 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 (UAL test: KT2)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Daily, 24 hours (See [Special Instructions](#))

Turnaround Time: 10 working days

Special Instructions: 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.

Monday – Thursday collections are preferred. Samples collected on Friday before 1400 can be shipped for Saturday delivery with special arrangements. Friday after 1400, Saturday/Sunday and holiday collections, will be held in the lab and shipped on Monday, or next business day.

NOTE: Detailed and accurate completion of this document is necessary for reporting purposes. The Medical Genomics Laboratory issues its clinical reports based on the demographic data provided by the referring institution on the lab requisition form. It is the responsibility of the referring institution to provide accurate information. If an amended report is necessary due to

inaccurate or illegible documentation, additional reports will be drafted with charge.

Specimen

Specimen Type: Whole blood

Container: Lavender top (EDTA) tube

Draw Volume: 6 mL (Minimum: 3 mL) whole blood **must be** in EDTA (Lavender) tubes

Processed Volume: Same as Draw Volume

Collection: Routine blood collection, invert gently to mix

Special Processing: Lab Staff:

1. **Do Not** centrifuge. Send whole blood at room temperature.
2. **DO NOT SHIP ON ICE.**
3. Include completed forms and requisition.
4. Be sure the shipping air bill is marked "Priority", Domestic.
5. Specimens must be packaged to prevent breakage and absorbent material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers

Shipping:

Monday- Thursday, ship specimen as priority with proper forms, at ambient temperature via overnight courier.

Friday before 1400 specimens can be shipped at ambient temperatures for Saturday delivery. Call the University of Alabama Genomics lab (205-934-5562) for special instructions.

Friday after 1400, Saturday or Sunday and holidays specimens should be held in the lab at ambient temperatures and shipped ambient on Monday or the next business day (Monday-Thursday).

Note: Blood collections are stable for 1 week after collection.

Patient Preparation: None

Sample Rejection: Requests for Molecular Genetic testing will not be accepted for the following reasons: No label (patients full name and date of collection) on the specimens; No referring physician's or genetic counselor's names and addresses; No billing information; No Phenotypic checklist form; Mislabeled or unlabeled specimens; Incorrect specimen type; Specimen frozen; Mislabeled specimens

Interpretive

Reference Range: Interpretive report

Critical Values: N/A

Limitations: N/A

Methodology: Sequence analysis

References: [University of Alabama Medical Genomics Laboratory](#) March 2018
(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.
6/13/2018: Updated collection and shipping information.