
Lab Dept: Anatomic Pathology

Test Name: TUBEROUS SCLEROSIS (TSC1/TSC2) COMPLEX SEQUENCING & DELETION/DUPLICATION

General Information

Lab Order Codes: TUBS

Synonyms: TSC Mutation Evaluation

CPT Codes: 81405 x1 – Molecular Pathology, Level 6
80406 x1 – Molecular Pathology, Level 7
81479 x1 – Molecular Pathology Unlisted

Test Includes: TSC1/TSC2 complex evaluation includes both sequencing and deletion/duplication.

Logistics

Test Indications: Tuberous Sclerosis Complex (TSC) is mainly caused by mutations in the TSC1 and TSC2 genes, which are tumor suppressors that are involved in cellular proliferation and act through multiple signaling pathways (TSC is inherited in an autosomal dominant manner with two-thirds of cases resulting from sporadic germline mutations while one-third of cases are inherited from an affected parent. It presents with complete penetrance, but has variable expressivity. Phenotypes of TSC types can be similar, but TSC2 mutations are reported to cause a more severe clinical presentation (Truncating mutations are found in the majority of TSC cases. Causative mutations reported to date include missense, splice site, small insertions and deletions, and large duplications and deletions.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: University of Alabama Medical Genomics Laboratory (UAL test: TSCP-NG)

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

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

Turnaround Time: Results with 25 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: Mislabeled or unlabeled specimens; frozen specimens

Interpretive

Reference Range: Interpretive report

Critical Values: N/A

Limitations: N/A

Methodology: DNA based sequencing and dosage (del/dup) analysis by MLPA.

References: [University of Alabama Medical Genomics Laboratory](#) June 2018
(800) 499-4363 Fax (205) 996-2929

Updates: 2/11/2013: CPT update
3/1/2013: Test moved from Boston University to Prevention Genetics, no longer includes MPLA, deletion/duplication. If needed, order separately.
4/16/2014: Test moved from Prevention Genetics to University of Alabama and now includes the TSC1/2 Complex.
1/11/2017: Updated CPT coding.
3/12/2018: CPT update.
6/13/2018: Updated collection and shipping information.