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:	81479 x3 – 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
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	Results with 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
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 60-72 hours of draw.
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 January 2017 (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. 1/11/17: Updated CPT coding.