Client: Example Client ABC123
123 Test Drive
Salt Lake City, UT 84108
UNITED STATES

Physician: Doctor, Example

Patient: Patient, Example

DOB	5/25/2015
Sex:	Female
Patient Identifiers:	01234567890ABCD, 012345
Visit Number (FIN):	01234567890ABCD
Collection Date:	01/01/2017 12:34

Tuberous Sclerosis Complex Panel, Sequencing and Deletion/Duplication, Fetal

ARUP test code 3002096

Maternal Contamination Study Fetal Spec	Fetal Cells
Maternal Contam Study, Maternal Spec	Whole Blood For quality assurance purposes, ARUP Laboratories will confirm the above result at no charge following delivery. Order Confirmation of Fetal Testing and include a copy of the original fetal report (or the mother's name and date of birth) with the test submission. Please contact an ARUP genetic counselor at (800) 242-2787 extension 2141 prior to specimen submission.
Tuberous Sclerosis Fetal Specimen	Cultured CVS
Tuberous Sclerosis Fetal Interp	Positive BACKGROUND INFORMATION: Tuberous Sclerosis Complex Panel, Sequencing and Deletion/Duplication, Fetal CHARACTERISTICS: Tuberous sclerosis complex (TSC) is a multisystem, genetic disorder causing numerous benign tumors as well as intellectual and developmental disabilities. Tumors can occur in the skin, brain, kidneys, and other organs, and can lead to significant health complications and may be life threatening. PREVALENCE: 1 in 6,000 individuals CAUSE: Pathogenic germline variants in TSC1 and TSC2 INHERITANCE: Autosomal dominant; approximately 66 percent are de novo PENETRANCE: Complete penetrance with variable expressivity CLINICAL SENSITIVITY: 95 percent GENES TESTED: TSC1, TSC2 METHODOLOGY: Targeted capture of all coding exons and exon-intron junctions of the targeted genes, followed by massively parallel sequencing. Sanger sequencing was performed as necessary to fill in regions of low coverage and confirm reported variants. A custom tiled comparative genomic hybridization array (aCGH) was used to detect large deletions or duplications in the targeted genes. Human genome build 19 (Hg 19) was used for data analysis. ANALYTICAL SENSITIVITY/SPECIFICITY: The analytical sensitivity of this test is approximately 99 percent for single nucleotide variants (SNVs) and greater than 93 percent for insertions / duplications / deletions from 1-10 base pairs in size. Variants greater than 10 base pairs may be detected, but the analytical sensitivity may be reduced. LIMITATIONS: A negative result does not exclude a diagnosis of TSC. This test only detects variants within the coding regions and intron-exon boundaries of the targeted genes. Regulatory

H=High, L=Low, *=Abnormal, C=Critical

region variants and deep intronic variants will not be identified and breakpoints of large deletions / duplications will not be determined. Single exon deletions / duplications or deletions / duplications less than 1kb may not be detected. Deletions / duplications / insertions of any size may not be detected by massive parallel sequencing. Diagnostic errors can occur due to rare sequence variations. In some cases, variants may not be identified due to technical limitations in the presence of pseudogenes, repetitive, or homologous regions. This assay may not detect low-level mosaic or somatic variants associated with disease. Interpretation of this test result may be impacted if this patient has had an allogeneic stem cell transplantation. Non-coding transcripts were not analyzed. Single exon deletions / duplications will not be called for the following exons: TSC2 (NM_000548) 17,29,41

This test was developed and its performance characteristics determined by ARUP Laboratories. It has not been cleared or approved by the US Food and Drug Administration. This test was performed in a CLIA certified laboratory and is intended for clinical purposes.

Counseling and informed consent are recommended for genetic testing. Consent forms are available online.

VERIFIED/REPORTED DATES

Procedure	Accession	Collected	Received	Verified/Reported
Maternal Contamination Study Fetal Spec	20-351-104150	12/16/2020 9:28:00 AM	12/16/2020 9:31:12 AM	12/16/2020 9:34:00 AM
Maternal Contam Study, Maternal Spec	20-351-104150	12/16/2020 9:28:00 AM	12/16/2020 9:31:12 AM	12/16/2020 9:34:00 AM
Tuberous Sclerosis Fetal Specimen	20-351-104150	12/16/2020 9:28:00 AM	12/16/2020 9:31:12 AM	12/16/2020 9:34:00 AM
Tuberous Sclerosis Fetal Interp	20-351-104150	12/16/2020 9:28:00 AM	12/16/2020 9:31:12 AM	12/16/2020 9:34:00 AM

END OF CHART

H=High, L=Low, *=Abnormal, C=Critical