

## MAYO CLINIC | Targeted Genes and Methodology | LABORATORIES | for Tuberous Sclerosis Gene Panel Targeted Genes and Methodology Details

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed. NGS and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript <sup>a</sup>
TCS1	NM_000368.5
TCS2	NM_000548.5

<sup>&</sup>lt;sup>a</sup> Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.