

Product datasheet for **RC402323**

Tuberin (TSC2) (NM_000548) Human Mutant ORF Clone

Product data:

Product Type:	Mutant ORF Clones
Product Name:	Tuberin (TSC2) (NM_000548) Human Mutant ORF Clone
Mutation Description:	E254X
Affected Codon#:	254
Affected NT#:	760
Nucleotide Mutation:	TSC2 Mutant (E254X), Myc-DDK-tagged ORF clone of Homo sapiens tuberous sclerosis 2 (TSC2), transcript variant 1 as transfection-ready DNA
Effect:	Tuberous sclerosis lymphangiomyomatosis
Symbol:	TSC2
Synonyms:	LAM; PPP1R160; TSC4
E. coli Selection:	Kanamycin (25 ug/mL)
Mammalian Cell Selection:	Neomycin
Vector:	pCMV6-Entry (PS100001)
Tag:	Myc-DDK
ACCN:	NM_000548
ORF Size:	759 bp
Restriction Sites:	SgfI-XhoI



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OTI Disclaimer:	The molecular sequence of this clone aligns with the gene accession number as a point of reference only. However, individual transcript sequences of the same gene can differ through naturally occurring variations (e.g. polymorphisms), each with its own valid existence. This clone is substantially in agreement with the reference, but a complete review of all prevailing variants is recommended prior to use. More info
OTI Annotation:	This clone was engineered to express the complete ORF with an expression tag. Expression varies depending on the nature of the gene.
Components:	The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).
RefSeq:	NP_000539
RefSeq Size:	759 bp
RefSeq ORF:	5424 bp
Locus ID:	7249
Cytogenetics:	16p13.3
Domains:	Rap_GAP, Tuberin
Protein Families:	Druggable Genome
Protein Pathways:	Insulin signaling pathway, mTOR signaling pathway, p53 signaling pathway
MW:	27.8 kDa
Gene Summary:	Mutations in this gene lead to tuberous sclerosis complex. Its gene product is believed to be a tumor suppressor and is able to stimulate specific GTPases. The protein associates with hamartin in a cytosolic complex, possibly acting as a chaperone for hamartin. Alternative splicing results in multiple transcript variants encoding different isoforms. [provided by RefSeq, Jul 2008]