

Product datasheet for **RC402339**

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:	K348X
Affected Codon#:	348
Affected NT#:	1042
Nucleotide Mutation:	TSC2 Mutant (K348X), Myc-DDK-tagged ORF clone of Homo sapiens tuberous sclerosis 2 (TSC2), transcript variant 1 as transfection-ready DNA
Effect:	Tuberous sclerosis
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:	1041 bp
Restriction Sites:	SgfI-XhoI



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ORF Nucleotide Sequence:

>RC402339 representing NM_000548
 Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC
 GCC**CGGATCGCC**

ATGGCCAAACCAACAAGCAAAGATTCAAGCTTGAAGGAGAAGTTAAGATTCTGTTGGGACTGGGAACAC
 CGAGGCCAAATCCCAGGTCTGCAGAGGGTAAACAGACGGAGTTTATCATCACCGGAAATACTGAGAGA
 ACTGAGCATGGAATGTGGCCTCAACAATCGCATCCGGATGATAGGGCAGATTTGTGAAGTCGAAAAACC
 AAGAAATTTGAAGAGCACGCAGTGAAGCACTCTGGAAGGCGGTGCGGGATCTGTTGCAGCCGGAGCGGC
 CGCTGGAGGCCCGGCACGCGGTCTGGCTCTGCTGAAGGCCATCGTGCAGGGCAGGGCAGCGTTTGGG
 GGTCTCAGAGCCCTCTCTTTAAGGTCATCAAGGATTACCTTCCAACGAAGACCTTACGAAAGGCTG
 GAGGTTTTCAAGGCCCTCACAGACAATGGGAGACATCACCTACTTGGAGGAAGAGCTGGCTGACTTTG
 TCCTGCAGTGGATGGATGTTGGCTTGTCTCGGAATTCCTTCTGGTGTGGTGAAGTGGTCAAATTCAA
 TAGCTGTTACCTCGACGAGTACATCGCAAGGATGGTTCAGATGATCTGTCTGCTGTGCGTCCGGACCGCG
 TCCTCTGTGGACATAGAGGTCTCCCTGCAGGTCTGGACGCCGTGGTCTGCTACAACCTGCCTGCCGGCTG
 AGAGCCTCCCGCTGTTATCGTTACCCTCTGTGCAACCATCAACGTCAAGGAGCTCTGCGAGCCTTGCTG
 GAAGCTGATGCGGAACCTCCTTGGCACCCACCTGGGCCACAGCGCCATCTACAACATGTGCCACCTCATG
 GAGGACAGAGCCTACATGGAGGACGCGCCCTGCTGAGAGGAGCCGTGTTTTTGTGGGCATGGCTCTCT
 GGGGAGCCACCGGCTCTATTCTCTCAGGAACCTGCGGACATCTGTGTTGCCATCATTTTACCAGGCCAT
 GGCATGTCCGAACGAGGTGGTGTCTATGAGATCGTCTGTCCATCACCAGGCTCATCAAG

AG**CGGACCG**ACGCGTACGCGGCCGCTCGAGCAGAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC
 TGGATTACAAGGATGACGACGA TAAGGTTTAA

Protein Sequence:

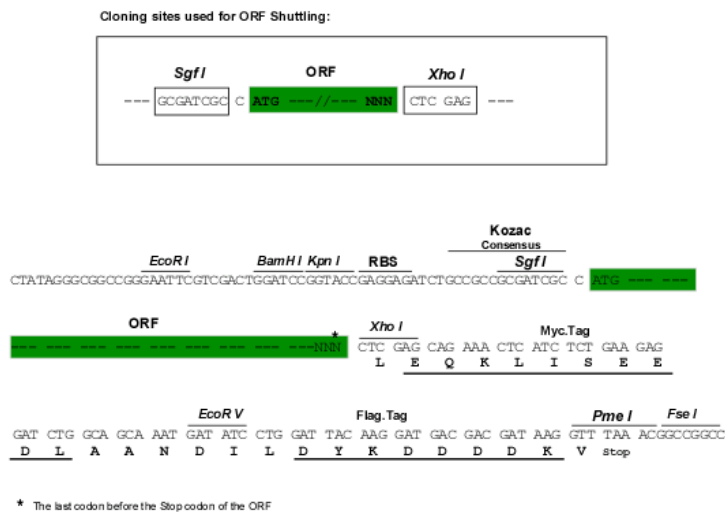
>RC402339 representing NM_000548
 Red=Cloning site Green=Tags(s)

MAKPTSKDSSLKEKFKILLGLGTPRPNRPSAEGKQTEFIITAEILRELSMECGLNNRIRMIGQICEVAKT
 KKFEEHAVEALWKAVADLLQPERPLEARHAVLALLKAIVQGGERLGVLRALFFKVIKDYPSNEDLHERL
 EVFKALTDNGRHITYLEELADFVLQWMDVGLSSEFLLVNLVKFNSCYLDEYIARMVQMICLLCVRTA
 SSVDIIEVSLQVLDVAVVCYNLPAESLPLFIVTLCRTINVKELCEPCWKLNRNLLGTHLGHSAIYNMCHLM
 EDRAYMEDAPLLRGAVFFVGMALWGAHRLYSLRNSPTSVLPSFYQAMACPNEVVSYEIVLSITRLIK

SGPTRRRRLEQKLISEEDLAANDILDYKDDDDKV

Restriction Sites:

Sgfl-XhoI

Cloning Scheme:

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:

1041 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:

38.2 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]