

Product datasheet for RC402512

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:	Q1665X
Affected Codon#:	1665
Affected NT#:	4993
Nucleotide Mutation:	TSC2 Mutant (Q1665X), 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:	4992 bp
Restriction Sites:	SgfI-XhoI
ORF Nucleotide Sequence:	>RC402512 representing NM_000548 Red=Cloning site Blue=ORF Green=Tags(s)

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Protein Sequence:

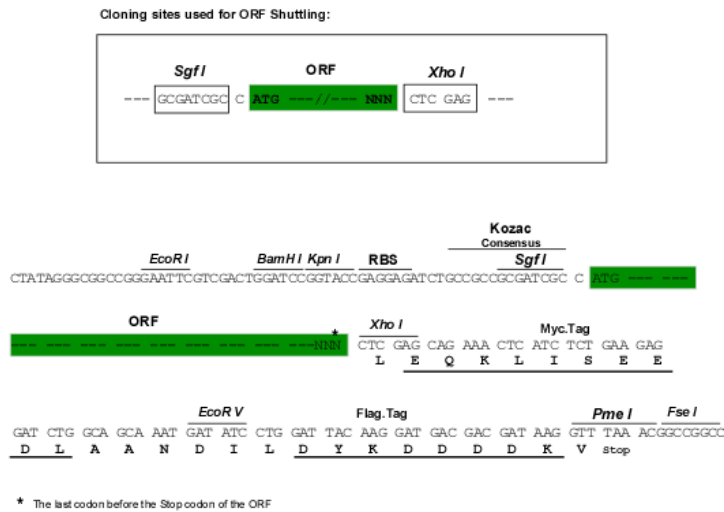
>RC402512 representing NM_000548
 Red=Cloning site Green=Tags(s)

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 HDDIMQAVFHIAITLMPKDVDKHRCDDKRHLGNDFVSIYVNDSEDFKLGTIKG

SGPTRRRLEQKLI SEEDLAANDILDYKDDDDKV

Restriction Sites:

SgfI-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:

4992 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:

183 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]