

## Product datasheet for **RC402353**

### 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:	Q478X
Affected Codon#:	478
Affected NT#:	1432
Nucleotide Mutation:	TSC2 Mutant (Q478X), 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:	1431 bp
Restriction Sites:	SgfI-XhoI



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**ORF Nucleotide Sequence:**

>RC402353 representing NM\_000548  
 Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC  
 GCC**GCGATCGCC**

ATGGCCAAACCAACAAGCAAAGATTAGGCTTGAAGGAGAAGTTAAGATTCTGTTGGGACTGGGAACAC  
 CGAGGCCAAATCCCAGGTCTGCAGAGGGTAAACAGACGGAGTTTATCATCACCGGAAAATACTGAGAGA  
 ACTGAGCATGGAATGTGGCCTCAACAATCGCATCCGGATGATAGGGCAGATTTGTGAAGTCGAAAAACC  
 AAGAAATTTGAAGAGCACGCAGTGAAGCACTCTGGAAGGCGGTGCGGATCTGTTGCAGCCGGAGCGGC  
 CGCTGGAGGCCCGGCACGCGGTCTGGCTCTGCTGAAGGCCATCGTGCAGGGCAGGGCAGCGTTTGGG  
 GGTCTCAGAGCCCTCTCTTTAAGGTCATCAAGGATTACCTTCCAACGAAGACCTTACGAAAGGCTG  
 GAGTTTTCAAGGCCCTCACAGACAATGGAGACACATCACCTACTTGGAGGAAGAGCTGGCTGACTTTG  
 TCCTGCAGTGGATGGATGTTGGCTTGTCTCGAATTCCTTCTGGTGTGGTGAAGTGGTCAAATCAA  
 TAGCTGTTACCTCGACGAGTACATCGCAAGGATGGTTCAGATGATCTGTCTGCTGTGCGTCCGGACCGCG  
 TCCTCTGTGGACATAGAGGTCTCCCTGCAGGTGCTGGACGCCGTGGTCTGCTACAACCTGCCGCGCTG  
 AGAGCCTCCCGCTGTTATCGTTACCCTCTGTGCGACCATCAACGTCAAGGAGCTCTGCGAGCCTTGCTG  
 GAAGCTGATGCGGAACCTCCTTGGCACCCACCTGGGCCACAGCGCCATCTACAACATGTGCCACCTCATG  
 GAGGACAGAGCCTACATGGAGGACGCGCCCTGCTGAGAGGAGCCGTGTTTTTGTGGGCATGGCTCTCT  
 GGGGAGCCACCGGCTCTATTCTCTCAGGAACCTCGCCGACATCTGTGTTGCCATCATTTTACCAGGCCAT  
 GGCATGTCCGAACGAGGTGGTGTCTATGAGATCGTCTGTCCATCACCAGGCTCATCAAGAAGTATAGG  
 AAGGAGCTCCAGTGGTGGCTGGGACATCTGCTGAACATCATCGAACGGCTCCTTACGAGCTCCAGA  
 CCTTGGACAGCCCGGAGCTCAGGACCATCGTCCATGACCTGTTGACCACGGTGGAGGAGCTGTGTGACCA  
 GAACGAGTCCACGGGTCTCAGGAGAGATACTTTGAACCTGGTGGAGAGATGTGCGGACCGAGGCCCTGAG  
 TCCTCCCTCTGAACCTGATCTCTATAGAGCGCAGTCCATCCACCCGGCCAAGGACGGCTGGATTGAGA  
 ACCTGCAGGCGCTGATGGAGAGATTCTCAGGAGCGAGTCCCAGGCGCCGTGCGCATCAAGGTGCTGGA  
 CGTGCTGTCTTTGTGCTGCTCATCAACAGG

AG**GCGACCG**ACGCGTACGCGGCCGCTCGAGCAGAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCC  
 TGGATTACAAGGATGACGACGA TAAGGTTTAA

**Protein Sequence:**

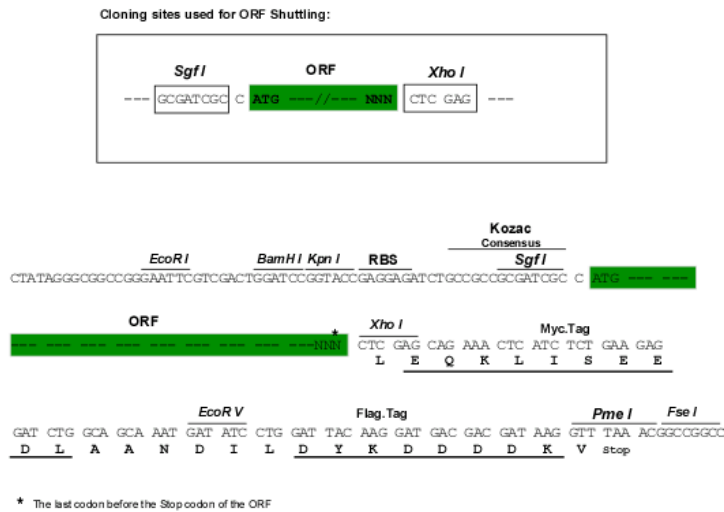
>RC402353 representing NM\_000548  
 Red=Cloning site Green=Tags(s)

MAKPTSKDSSLKEKFKILLGLGTPRPNRPSAEGKQTEFIITAEILRELSMEGLNLRIRMIGQICEVAKT  
 KKFEHAEVALWKAVADLLQPERPLEARHAVLALLKAIYQGQGERLGVLRALFFKVIKDYPSNEDLHERL  
 EVFKALTDNGRHITYLEELADFVLQWMDVGLSSEFLLVLVNLVKFNLCYLDEYIARMVQMICLLCVRTA  
 SSVDIEVSLQVLDVAVVCYNCLPAESLPLFIVTLCRTINVKELCEPCWKLNRNLLGTHLGHSAIYNMCHLM  
 EDRAYMEDAPLLRGAVFFVGMALWGAHRLYSLRNSPTSVLPSFYQAMACPNVSVYEVLSITRLIKKYR  
 KELQVVAWDILLNIIERLLQQLQTLDSPELRTIVHDLLTTVEELCDQNEFHGSQERYFELVERCADQRPE  
 SLLNLLISYRAQSIHPAKDGIQNLQALMERFFRSESRGAVRIKVLVDVLSFVLLINR

SGP**TRRRLEQKLI**SEEDLAANDILDYKDDDDKV

**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:**

1431 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:**

52.5 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]