

## Product datasheet for **RC402494**

### 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:     | Y1593X   |
| Affected Codon#:          | 1593   |
| Affected NT#:             | 4779   |
| Nucleotide Mutation:      | TSC2 Mutant (Y1593X), Myc-DDK-tagged ORF clone of Homo sapiens tuberous sclerosis 2 (TSC2), transcript variant 1 as transfection-ready DNA |
| Effect:                   | Tuberous sclerosis   |
| Symbol:                   | Tuberin  |
| 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:                 | 4776 bp  |
| Restriction Sites:        | SgfI-XhoI  |
| ORF Nucleotide Sequence:  | >RC402494 representing NM_000548<br>Red=Cloning site Blue=ORF Green=Tags(s)  |

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GCC**CGATCGCC**

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TCCTGCAGTGGATGGATGTTGGCTTGTCTCGGAATTCCTTCTGGTGCTGGTGAACCTGGTCAAATTCAA  
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**Protein Sequence:**

>RC402494 representing NM\_000548

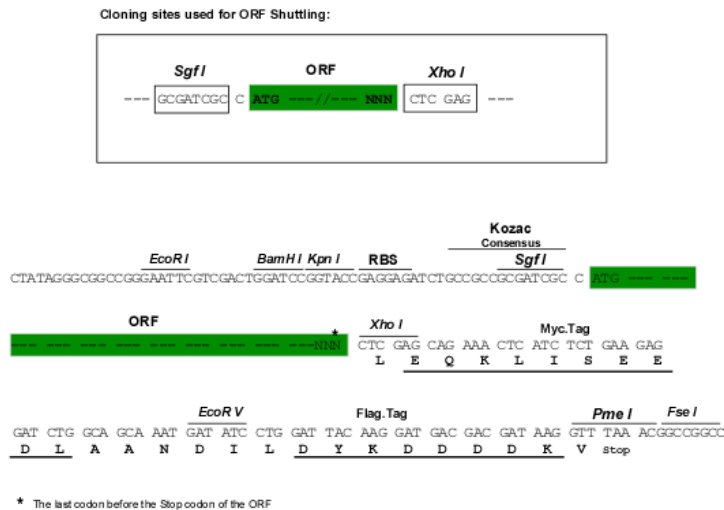
Red=Cloning site Green=Tags(s)

MAKPTSKDSSLKEFKILLGLGTPRPNRPAEKGQTEFIITAEILRELSMECGLNRRMIRMIQICEVAKT  
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 SQLSHIPEDKDHQVRKLATQLLDLAEGCHTHFNSLLDIIKVMARSLSPPELEERDVAAYSASLEDV  
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 LPNKDGVVRFSPYCVCDYMEPERGSEKKTSGPLSPPTGPPGAPAGPAVRLGSPYSLLFRVLLQCLKQE  
 SDWKVLKLVLRPELRYKVLIFTSPCSVDQLCSALCSMLSGPKTLERLRGAPEGFRTDLHLAVVPVL  
 TALISYHNYLDKTKQREMYCLEQGLIHRCASQCVALSICSVEMPDI I IKALPVLVVKLTHISATASMA  
 VPLLEFLSTLARLPHLYRNFAAEQYASVFAISLPYTNPSKFNQYIVCLAHHVIAWVIRCLPFRKDFVP  
 FITKGLRSNVLVSFDDTPEKDSFRARSTSLNERPKSLRIARPPKQGLNNSPPVKEFKESSAAEFRCRSI  
 SVSEHVRSRIQTSLSASLGSADENSAQADDLKNLHLELTETCLDMARYVFSNFTAVPKRSPVGEF  
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 AKPPPLPRSNVASFSSLYQSSCQQLHRSVSWADSAVMEEGSPGEVPLVEPPGLEDEAALGMDRRT  
 DAYSRSSSVSSQEEKSLHAEELVGRGIPIERVVSSEGGSPVDLSFQPSQPLSKSSSPELQTLQDILGD  
 PGDKADVGRLSPEVKARSQSGTLDGESAAWSASGEDSRGQPEGPLPSSSPRSPGLRPRGYTISDSAPSR  
 RGKRVERDALKSRATASNAEKVPGINPSFVFLQLYHSPFFGDESNKPILLPNESQSFERSVQLLDQIPSY  
 DTHKIAVLYVGEQSNSELAAILSNEHGSYRYTEFLTGLGRLIELKDCQDPKV

SGPTRRRLKQLISEEDLAANDILDYKDDDDKV

**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).

**Note:**

Plasmids are not sterile. For experiments where strict sterility is required, filtration with 0.22um filter is required.

**RefSeq:**

[NP\\_000539](#)

**RefSeq Size:**

4776 bp

**RefSeq ORF:**

5424 bp

**Locus ID:**

7249

**Cytogenetics:**

16p13.3

|                          |  |
|--------------------------|--|
| <b>Domains:</b>          | Rap_GAP, Tuberin   |
| <b>Protein Families:</b> | Druggable Genome   |
| <b>Protein Pathways:</b> | Insulin signaling pathway, mTOR signaling pathway, p53 signaling pathway   |
| <b>MW:</b>               | 175.1 kDa  |
| <b>Gene Summary:</b>     | 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] |