

## **Product datasheet for RC400807**

## BTK (NM 000061) Human Mutant ORF Clone

## **Product data:**

**Product Type:** Mutant ORF Clones

**Product Name:** BTK (NM 000061) Human Mutant ORF Clone

Mutation Description: Y112X

Affected Codon#: 112

Affected NT#: 336

**Nucleotide Mutation:** BTK Mutant (Y112X), Myc-DDK-tagged ORF clone of Homo sapiens Bruton

agammaglobulinemia tyrosine kinase (BTK) as transfection-ready DNA

Effect: Ammlobulinemi

Symbol: BTK

Synonyms: AGMX1; AT; ATK; BPK; IGHD3; IMD1; PSCTK1; XLA

E. coli Selection: Kanamycin (25 ug/mL)

Mammalian Cell Neomycin

Selection:

**Vector:** pCMV6-Entry (PS100001)

Tag: Myc-DDK
ACCN: NM 000061

ORF Size: 333 bp

**Restriction Sites:** Sgfl-Mlul

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## BTK (NM\_000061) Human Mutant ORF Clone - RC400807

**ORF Nucleotide** Sequence:

>RC400807 representing NM\_000061

Red=Cloning site Blue=ORF Green=Tags(s)

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCC GCCGCGATCGCC

ATGGCCGCAGTGATTCTGGAGAGCATCTTTCTGAAGCGATCCCAACAGAAAAAGAAAACATCACCTCTAA ACTTCAAGAAGCGCCTGTTTCTCTTGACCGTGCACAAACTCTCCTACTATGAGTATGACTTTGAACGTGG GAGAAGAGCAGTAAGAAGGGTTCAATAGATGTTGAGAAGATCACTTGTGTTGAAACAGTGGTTCCTGAA AAAAATCCTCCTCCAGAAAGACAGATTCCGAGAAGAGGTGAAGAGTCCAGTGAAATGGAGCAAATTTCAA TCATTGAAAGGTTCCCTTATCCCTTCCAGGTTGTATATGATGAAGGGCCTCTC

**AGCGGACCG**ACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGATCTGGCAGCAAATGATATCC TGGATTACAAGGATGACGACGA TAAGGTTTAA

**Protein Sequence:** 

>RC400807 representing NM\_000061 Red=Cloning site Green=Tags(s)

MAAVILESIFLKRSQQKKKTSPLNFKKRLFLLTVHKLSYYEYDFERGRRGSKKGSIDVEKITCVETVVPE KNPPPERQIPRRGEESSEMEQISIIERFPYPFQVVYDEGPL

**SGPTRTRRL**EQKLISEEDLAANDILDYKDDDDK**V** 

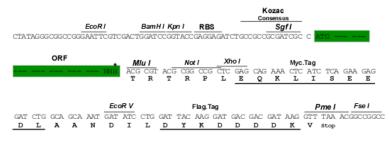
**Restriction Sites:** 

Sgfl-Mlul

**Cloning Scheme:** 







<sup>\*</sup> The last codon before the Stop codon of the ORF



**OTI Disclaimer:** 

Due to the inherent nature of this plasmid, standard methods to replicate additional amounts of DNA in E. coli are highly likely to result in mutations and/or rearrangements. Therefore, OriGene does not guarantee the capability to replicate this plasmid DNA. Additional amounts of DNA can be purchased from OriGene with batch-specific, full-sequence verification at a reduced cost. Please contact our customer care team at <a href="mailto:customport@origene.com">customport@origene.com</a> or by calling 301.340.3188 option 3 for pricing and delivery.

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. <u>More info</u>

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 000052

RefSeq Size: 333 bp
RefSeq ORF: 1980 bp
Locus ID: 695

Cytogenetics: Xq22.1

**Domains:** pkinase, SH2, TyrKc, SH3, BTK, PH, S\_TKc

**Protein Families:** Druggable Genome, Protein Kinase

**Protein Pathways:** B cell receptor signaling pathway, Fc epsilon RI signaling pathway, Primary immunodeficiency

**MW:** 12.2 kDa

**Gene Summary:** The protein encoded by this gene plays a crucial role in B-cell development. Mutations in this

gene cause X-linked agammaglobulinemia type 1, which is an immunodeficiency

characterized by the failure to produce mature B lymphocytes, and associated with a failure of Ig heavy chain rearrangement. Alternative splicing results in multiple transcript variants

encoding different isoforms. [provided by RefSeq, Dec 2013]