

Product datasheet for RC400814

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: W124X

Affected Codon#: 124

Affected NT#: 371

Nucleotide Mutation: BTK Mutant (W124X), 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: 369 bp

Restriction Sites: Sgfl-Mlul

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BTK (NM_000061) Human Mutant ORF Clone - RC400814

ORF Nucleotide Sequence:

>RC400814 representing NM_000061

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

TTTTGTAATACGACTCACTATAGGGCGGCCGGGAATTCGTCGACTGGATCCGGTACCGAGGAGATCTGCCGCCGCGATCGCC

ATGGCCGCAGTGATTCTGGAGAGCATCTTTCTGAAGCGATCCCAACAGAAAAAGAAAACATCACCTCTAA ACTTCAAGAAGCGCCTGTTTCTCTTGACCGTGCACAAACTCTCCTACTATGAGTATGACTTTGAACGTGG GAGAAGAGGCAGTAAGAAGGGTTCAATAGATGTTGAGAAGATCACTTGTGTTGAAACAGTGGTTCCTGAA AAAAATCCTCCTCCAGAAAGACAGATTCCGAGAAGAGGGTGAAGAGTCCAGTGAAATGGAGCAAATTTCAA TCATTGAAAGGTTCCCTTATCCCTTCCAGGTTGTATATGATGAAGGGCCTCTCTACGTCTTCTCCCCAAC TGAAGAACTAAGGAACGG

AGCGGACCGACGCGTACGCGGCCGCTCGAGCAGAAACTCATCTCAGAAGAGGATCTGGCAGCAAATGATATCCTGGATTACAAGGATGACGACGA TAAGGTTTAA

Protein Sequence:

>RC400814 representing NM_000061
Red=Cloning site Green=Tags(s)

 ${\tt MAAVILESIFLKRSQQKKKTSPLNFKKRLFLLTVHKLSYYEYDFERGRRGSKKGSIDVEKITCVETVVPEKNPPPERQIPRRGEESSEMEQISIIERFPYPFQVVYDEGPLYVFSPTEELRKR}$

SGPTRTRRLEQKLISEEDLAANDILDYKDDDDK**V**

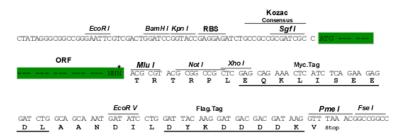
Restriction Sites:

Sgfl-Mlul

Cloning Scheme:

Cloning sites used for ORF Shuttling:





^{*} 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 customercom 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: 369 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: 13.5 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]