

## Product datasheet

# Recombinant human BTK (mutated C481R) protein (Active) ab268372

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### Description

<b>Product name</b>	Recombinant human BTK (mutated C481R) protein (Active)
<b>Biological activity</b>	The specific activity of ab268372 was 2.5 nmol/min/mg in a kinase assay using Poly (4:1 Glu, Tyr) synthetic peptide as substrate.
<b>Purity</b>	> 75 % SDS-PAGE. Affinity purified.
<b>Expression system</b>	Baculovirus infected Sf9 cells
<b>Accession</b>	<b><u>Q06187</u></b>
<b>Protein length</b>	Full length protein
<b>Animal free</b>	No
<b>Nature</b>	Recombinant
<b>Species</b>	Human
<b>Molecular weight information</b>	SDS-PAGE molecular weight: ~75kDa
<b>Modifications</b>	mutated C481R
<b>Tags</b>	His tag N-Terminus
<b>Additional sequence information</b>	GenBank: NM_000061

### Specifications

Our **Abpromise guarantee** covers the use of **ab268372** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

<b>Applications</b>	Functional Studies SDS-PAGE
<b>Form</b>	Liquid

### Preparation and Storage

<b>Stability and Storage</b>	Shipped on Dry Ice. Upon delivery aliquot. Store at -80°C. Avoid freeze / thaw cycle. pH: 7.00 Preservative: 1.02% Imidazole
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Constituents: 0.82% Sodium phosphate, 1.74% Sodium chloride, 0.002% PMSF, 0.004% DTT, 25% Glycerol (glycerin, glycerine)

This product is an active protein and may elicit a biological response in vivo, handle with caution.

## General Info

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### Function

Plays a crucial role in B-cell ontogeny. Transiently phosphorylates GTF2I on tyrosine residues in response to B-cell receptor cross-linking. Required for the formation of functional ARID3A DNA-binding complexes.

### Involvement in disease

Defects in BTK are the cause of X-linked agammaglobulinemia (XLA) [MIM:300755]; also known as X-linked agammaglobulinemia type 1 (AGMX1) or immunodeficiency type 1 (IMD1). XLA is a humoral immunodeficiency disease which results in developmental defects in the maturation pathway of B-cells. Affected boys have normal levels of pre-B-cells in their bone marrow but virtually no circulating mature B-lymphocytes. This results in a lack of immunoglobulins of all classes and leads to recurrent bacterial infections like otitis, conjunctivitis, dermatitis, sinusitis in the first few years of life, or even some patients present overwhelming sepsis or meningitis, resulting in death in a few hours. Treatment in most cases is by infusion of intravenous immunoglobulin.

Defects in BTK may be the cause of X-linked hypogammaglobulinemia and isolated growth hormone deficiency (XLA-IGHD) [MIM:307200]; also known as agammaglobulinemia and isolated growth hormone deficiency or Fleisher syndrome or isolated growth hormone deficiency type 3 (IGHD3). In rare cases XLA is inherited together with isolated growth hormone deficiency (IGHD).

### Sequence similarities

Belongs to the protein kinase superfamily. Tyr protein kinase family. TEC subfamily.

Contains 1 Btk-type zinc finger.

Contains 1 PH domain.

Contains 1 protein kinase domain.

Contains 1 SH2 domain.

Contains 1 SH3 domain.

### Post-translational modifications

Autophosphorylated on Tyr-223 and Tyr-551. Phosphorylation of Tyr-223 may create a docking site for a SH2 containing protein.

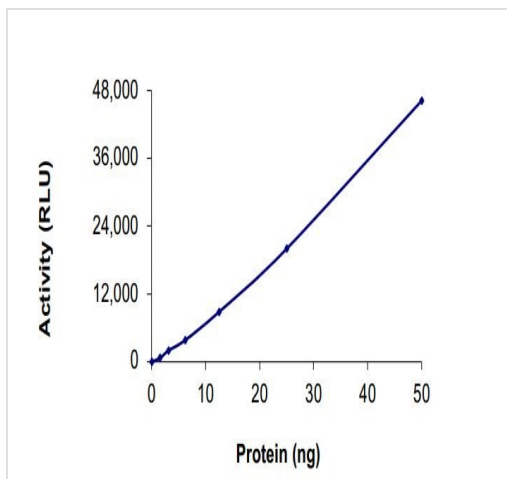
### Cellular localization

Cytoplasm. Membrane. Nucleus.

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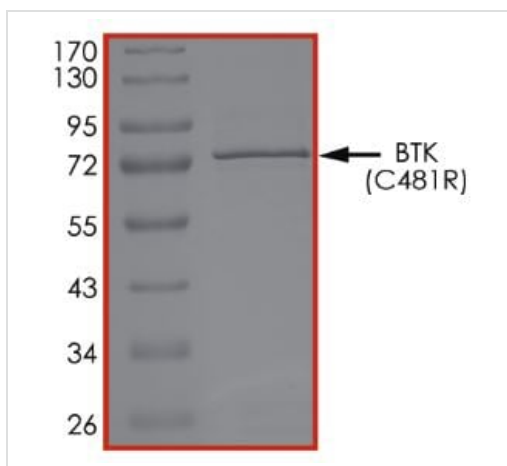
## Images

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Functional Studies - Recombinant human BTK (mutated C481R) protein (Active) (ab268372)

The specific activity of ab268372 was 2.5 nmol/min/mg in a kinase assay using Poly (4:1 Glu, Tyr) synthetic peptide as substrate.



SDS-PAGE - Recombinant human BTK (mutated C481R) protein (Active) (ab268372)

SDS-PAGE analysis of ab268372.

**Please note:** All products are "FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC PROCEDURES"

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