abcam

Product datasheet

Recombinant Human XPNPEP3 protein ab173067

Description

Product name	Recombinant Human XPNPEP3 protein
Purity	> 95 % SDS-PAGE. ab173067 is greater than 95% pure, as determined by SEC-HPLC and reducing SDS-PAGE.
Endotoxin level	< 1.000 Eu/µg
Expression system	Escherichia coli
Accession	Q9NQH7
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MGSSHHHHHHSSGLVPRGSHMPWLLSAPKLVPAVANVR GLSGCMLCSQRR YSLQPVPERRIPNRYLGQPSPFTHPHLLRPGEVTPGLSQV EYALRRHKLM SLIQKEAQGQSGTDQTVVVLSNPTYYMSNDIPYTFHQDNN FLYLCGFQEP DSILVLQSLPGKQLPSHKAILFVPRRDPSRELWDGPRSGT DGAIALTGVD EAYTLEEFQHLLPKMKAETNMVWYDWMRPSHAQLHSDY MQPLTEAKAKSK NKVRGVQQLIQRLRLIKSPAEIERMQIAGKLTSQAFIETMFT SKAPVEEA FLYAKFEFECRARGADILAYPPVVAGGNRSNTLHYVKNNQ LIKDGEMVLL DGGCESSCYVSDITRTWPVNGRFTAPQAELYEAVLEIQRD CLALCFPGTS LENIYSMMLTLIGQKLKDLGIMKNIKENNAFKAARKYCPHHV GHYLGMDV HDTPDMPRSLPLQPGMVITIEPGIYIPEDDKDAPEKFRGLG VRIEDDVVV TQDSPFILSADCPKEMNDIEQICSQASLEHHHHHH
Predicted molecular weight	60 kDa including tags
Amino acids	1 to 507
Tags	His tag C-Terminus , His tag N-Terminus

Specifications

Our **Abpromise guarantee** covers the use of **ab173067** in the following tested applications.

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

Applications	HPLC SDS-PAGE
Form	Liquid

Preparation and Storage

Stability and Storage	Shipped on Dry Ice. Store at -20°C or -80°C. Avoid freeze / thaw cycle. pH: 7.30 Constituents: 0.02% DTT, 0.3% Tris It is supplied as an 0.2 µM filtered solution.

General Info

Tissue specificity	Isoform 1 and isoform 2 are widely expressed, with isoform 1 being more abundant.
Involvement in disease	Defects in XPNPEP3 are the cause of nephronophthisis-like nephropathy type 1 (NPHPL1) [MIM:613159]. A disorder with features of nephronophthisis, a cystic kidney disease leading to end-stage renal failure. Nephronophthisis is histologically characterized by modifications of the tubules with thickening of the basement membrane, interstitial fibrosis and, in the advanced stages, medullary cysts. Typical clinical manifestation are chronic renal failure, anemia, polyuria, polydipsia, isosthenuria, and growth retardation. Associations with extrarenal symptoms are frequent. In NPHPL1 patients, extrarenal symptoms include hypertension, essential tremor, sensorineural hearing loss and gout. Severely affected individuals can manifest a mitochondrial disorder with isolated complex I deficiency activity in muscle, seizures, mental retardation and hypertrophic dilated cardiomyopathy.
Sequence similarities	Belongs to the peptidase M24B family.
Cellular localization	Mitochondrion.

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

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