

HAX1 Rabbit pAb

Catalog # AP96252

Product Information

Application	WB, IHC-P, IHC-F, IF, E
Predicted	Rat, Pig, Human, Mouse, Rabbit, Dog, Horse
Host	Rabbit
Clonality	Polyclonal
Calculated MW	31 KDa
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human HAX1
Epitope Specificity	191-279/279
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Mitochondrion. Endoplasmic reticulum. Nucleus membrane. Cytoplasmic vesicle (By similarity). Sarcoplasmic reticulum (By similarity).
SIMILARITY	Belongs to the HAX1 family.
SUBUNIT	Interacts with ABCB1, ABCB4 and ABCB11 (By similarity). Directly associates with HCLS1/HS1, through binding to its N-terminal region. Interacts with CTTN. Interacts with PKD2. Interacts with GNA13. Interacts with CASP9. Interacts with ITGB6. Interacts with PLN and ATP2A2; these interactions are inhibited by calcium. Interacts with GRB7. Interacts (via C-terminus) with XIAP/BIRC4 (via BIR 2 domain and BIR 3 domain) and this interaction blocks ubiquitination of XIAP/BIRC4.
Post-translational modifications	Proteolytically cleaved by caspase-3 during apoptosis.
DISEASE	Defects in HAX1 are the cause of neutropenia severe congenital autosomal recessive type 3 (SCN3) [MIM:610738]; also known as Kostmann disease. A disorder of hematopoiesis characterized by maturation arrest of granulopoiesis at the level of promyelocytes with peripheral blood absolute neutrophil counts below $0.5 \times 10^9/l$ and early onset of severe bacterial infections. Some patients affected by severe congenital neutropenia type 3 have neurological manifestations such as psychomotor retardation and seizures. Note=The clinical phenotype due to HAX1 deficiency appears to depend on the localization of the mutations and their influence on the transcript variants. Mutations affecting exclusively isoform 1 are associated with isolated congenital neutropenia, whereas mutations affecting both isoform 1 and isoform 5 are associated with additional neurologic symptoms.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	RelevanceHAX1 associates with HS1, binding to its N-terminal region. It is also known to associate with PKD2 (involved in polycystic kidney disease) and with cortactin/EMS1. HAX1 is also reported to bind to hairpin structures in vimentin and DNA polymerase beta mRNAs, so may play a role in mRNA stability and transport. It may also function in promoting cell survival. Defects in HAX1 are the cause of autosomal recessive severe congenital neutropenia 3 (SCN3) also called Kostmann disease.

Additional Information

Gene ID	10456
Other Names	HCLSBP1; HS1BP1; SCN3; HCLS1 associated protein X-1; HCLS1 (and PKD2) associated protein
Target/Specificity	Ubiquitous. Up-regulated in oral cancers.
Dilution	WB=1:500-2000,IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500,ELISA=1:5000-10000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
Name	HAX1 Rabbit pAb
Function	Promotes cell survival. Potentiates GNA13-mediated cell migration. Involved in the clathrin-mediated endocytosis pathway. May be involved in internalization of ABC transporters such as ABCB11. May inhibit CASP9 and CASP3. May regulate intracellular calcium pools.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.