

Recombinant Human HAX1 protein ab140544

1 图像

|           |                                                                                                                                                                                                                                                                                                                                                                         |
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| 描述        |                                                                                                                                                                                                                                                                                                                                                                         |
| 产品名称      | 重组人HAX1蛋白                                                                                                                                                                                                                                                                                                                                                               |
| 纯度        | > 80 % SDS-PAGE.<br>ab140544 is purified using conventional chromatography techniques.                                                                                                                                                                                                                                                                                  |
| 表达系统      | Escherichia coli                                                                                                                                                                                                                                                                                                                                                        |
| Accession | <u>O00165</u>                                                                                                                                                                                                                                                                                                                                                           |
| 蛋白长度      | Full length protein                                                                                                                                                                                                                                                                                                                                                     |
| 无动物成分     | No                                                                                                                                                                                                                                                                                                                                                                      |
| 性质        | Recombinant                                                                                                                                                                                                                                                                                                                                                             |
| 种属        | Human                                                                                                                                                                                                                                                                                                                                                                   |
| 序列        | MGSSHHHHHH SSGLVPRGSH MSLFDLFRGF<br>FGFPGPRSHR DPFFGGMTRD EDDDEEEEEE<br>GGSWGRGNPR FHSPQHPPPEE FGFGFSFSPG<br>GGIRFHDFNG FDDLVRDFNS IFSDMGAWTL<br>PSHPPPELPGP ESETPGERLR EGQTLRDSML<br>KYPDSHQPRI FGGVLESDAR SESPQPAPDW<br>GSQRPFHRFD DVWPMDPHPR TREDNDLDSQ<br>VSQEGLGPVL QPQPKSYFKS ISVTKITKPD<br>GIVEERRTVV DSEGRTEETTV TRHEADSSPR<br>GDPESPRPPA LDDAFSILD L FLGRWFRSR |
| 预测分子量     | 34 kDa including tags                                                                                                                                                                                                                                                                                                                                                   |
| 氨基酸       | 1 to 279                                                                                                                                                                                                                                                                                                                                                                |
| 标签        | His tag N-Terminus                                                                                                                                                                                                                                                                                                                                                      |

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| 技术指标                                                                                                                                 |                               |
| Our <b>Abpromise guarantee</b> covers the use of <b>ab140544</b> in the following tested applications.                               |                               |
| The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user. |                               |
| 应用                                                                                                                                   | SDS-PAGE<br>Mass Spectrometry |
| 质谱法                                                                                                                                  | MALDI-TOF                     |
| 形式                                                                                                                                   | Liquid                        |

## 制备和贮存

### 稳定性和存储

Shipped at 4°C. Store at +4°C short term (1-2 weeks). Upon delivery aliquot. Store at -20°C or -80°C. Avoid freeze / thaw cycle.

pH: 8.00

Constituents: 0.32% Tris HCl, 10% Glycerol (glycerin, glycerine), 0.58% Sodium chloride

## 常规信息

### 功能

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.

### 组织特异性

Ubiquitous. Up-regulated in oral cancers.

### 疾病相关

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.

### 序列相似性

Belongs to the HAX1 family.

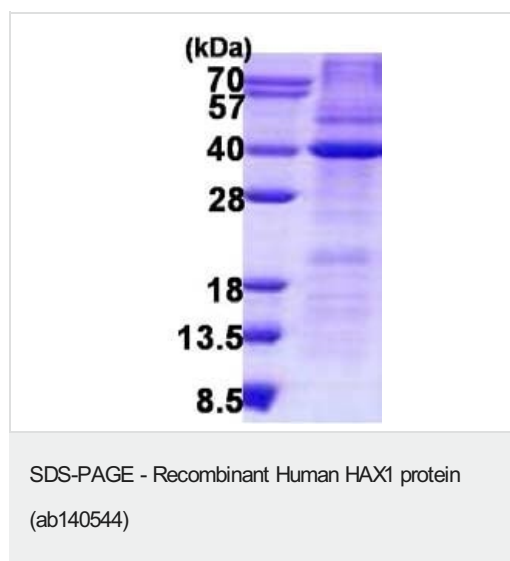
### 翻译后修饰

Proteolytically cleaved by caspase-3 during apoptosis.

### 细胞定位

Mitochondrion. Endoplasmic reticulum. Nucleus membrane. Cytoplasmic vesicle. Sarcoplasmic reticulum.

## 图片



15% SDS-PAGE analysis of ab140544 (3μg).

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