

Recombinant Full Length Human AHI1 Protein, GST-tagged

Cat. No. AHI1-945HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Human AHI1 full-length ORF (AAH94800.1, 1 a.a. - 609 a.a.) recombinant protein with GST-tag at N-terminal.
Species	Human
Source	In Vitro Cell Free System
ProteinLength	609 amino acids
Description	This gene is apparently required for both cerebellar and cortical development in humans. This gene mutations cause specific forms of Joubert syndrome-related disorders. Joubert syndrome (JS) is a recessively inherited developmental brain disorder with several identified causative chromosomal loci. Alternatively spliced transcript variants encoding different isoforms have been identified. [provided by RefSeq, Oct 2008]
Molecular Mass	96.4 kDa
AA Sequence	MPTAESEAKV KTKVRFEE LL KTHSDLMREK KKLKKKL VRS EENISPDTIR SNLHYMKETT SDDPDTIRSN LPHIKETTSD DVSAANTNNL KKSTRVTKNK LRNTQLATEN PNGDASVEED KQGKPNKKVI KTVPQLTTQD LKPETPENKV DSTHQKTHTK PQPGVDHQKS EKANEGREET DLEEDEELMQ AYQCHVTEEM AKEIKRKIRK KLKEQLTYFP SDTLFHDDKL SSEKRKKKKE VPVFSKAETS TLTISGDTVE GEQKKESSVR SVSSDSHQDD EISSMEQSTE DSMQDDTKPK PKKTKKKT KA VADNNEDVDG DGVHEITSRD SPVYPKCLLD DDLVLGVYIH

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RTDRLKSDFM ISHPMVKIHV VDEHTGQYVK KDDSGRPVSS YYEKENVDYI
 LPIMTQPYDF KQLKSRLPEW EEQIVFNENF PYLLRGSDDES PKVILFFEIL
 DFLSVDEIKN NSEVQNQECG FRKIAWAFLK LLGANGNANI NSKLRLQLYY
 PPTKPRSPLS VVEAFEWWSK CPRNHYPSTL YVTVRGLKVP DCIKPSYRSM
 MALQEEKGKP VHCERHHESS SVDTEPGLEE SKEVIKWKRL PGQENEDVFV
 LISPTMEEY

Applications

Enzyme-linked Immunoabsorbent Assay
 Western Blot (Recombinant protein)
 Antibody Production
 Protein Array

Storage

Store at -80 centigrade. Aliquot to avoid repeated freezing and thawing.

Storage Buffer

50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.

GENE INFORMATION

Gene Name

AHI1 Abelson helper integration site 1 [Homo sapiens]

Official Symbol

AHI1

Synonyms

AHI1; Abelson helper integration site 1; Abelson helper integration site; jouberin;
 FLJ20069; JBTS3; Jouberin; ORF1; contatins SH3 and WD40 domains; abelson
 helper integration site 1 protein homolog; AHI-1; dJ71N10.1; FLJ14023;
 DKFZp686J1653

Gene ID

54806

mRNA Refseq

NM_001134830

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Protein Refseq	NP_001128302
MIM	608894
UniProt ID	Q8N157

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