SunLong Biotech Co.,LTD
Tel: 0086-571- 56623320 Fax:0086-571- 56623318
E-mail:sales@sunlongbiotech.com
www.sunlongbiotech.com

Rabbit Anti-AHI1 antibody

SL7854R

Product Name:	AHI1
Chinese Name:	白血病相关蛋白AHI1抗体
Alias:	Abelson helper integration site 1 protein homolog; Abelson helper integration site 1; Abelson helper integration site; AHI 1; AHI-1; Ahi1; AHI1_HUMAN; Contatins SH3 and WD40 domains; JBTS3; Jouberin; ORF1.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,Rabbit,Sheep,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	137kDa
Cellular localization:	cytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human AHI1:801-900/1196
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. 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.
PubMed:	PubMed
Product Detail:	Highly expressed in the most primitive normal hematopoietic cells. Expressed in brain, particularly in neurons that give rise to the crossing axons of the corticospinal tract and superior cerebellar peduncles. Expressed in kidney (renal collecting duct cells) (at protein level). Involvement in disease:Defects in AHI1 are the cause of Joubert syndrome type 3

	(JBTS3) . JBTS is an autosomal recessive disorder presenting with cerebellar ataxia, oculomotor apraxia, hypotonia, neonatal breathing abnormalities and psychomotor delay. Neuroradiologically, it is characterized by cerebellar vermian hypoplasia/aplasia, thickened and reoriented superior cerebellar peduncles, and an abnormally large interpeduncular fossa, giving the appearance of a molar tooth on transaxial slices (molar tooth sign). Additional variable features include retinal dystrophy and renal disease. JBTS3 shows minimal extra central nervous system involvement and appears not to be associated with renal dysfunction. **Function:** Component of the tectonic-like complex, a complexlocalized at the transition zone of primary cilia and acting as abarrier that prevents diffusion of transmembrane proteins betweenthe cilia and plasma membranes (By similarity). **Subunit:** Part of the tectonic-like complex (also named B9complex). Interacts with MKS1 (By similarity). Interacts withNPHP1. **Subcellular Location:** Cytoplasm, cytoskeleton, cilium basal body(By similarity). Cell junction, adherens junction. **Tissue Specificity:** Highly expressed in the most primitive normalhematopoietic cells. Expressed in brain, particularly in neuronsthat give rise to the crossing axons of the corticospinal tract andsuperior cerebellar peduncles. Expressed in kidney (renalcollecting duct cells) (at protein level). **DISEASE:** Defects in AHI1 are the cause of Joubert syndrome type 3(JBTS3) [MIM:608629]. JBTS is an autosomal recessive disorderpresenting with cerebellar ataxia, oculomotor apraxia, hypotonia,neonatal breathing abnormalities and psychomotor delay.Neuroradiologically, it is characterized by cerebellar vermianhypoplasia/aplasia, thickened and reoriented superior cerebellarpeduncles, and an abnormally large interpeduncular fossa, givingthe appearance of a molar tooth on transaxial slices (molar toothsign). Additional variable features include retinal dystrophy andrenal disease. JBTS3 shows minimal extra central nervous systeminvolvement and appears not to be associated with renaldysfunction. **Similarity:** Contains 1 SH3 domain. Contains 7 WD repeats. **SWISS:** Q8N157

	Gene ID: 54806 **Database links:** Entrez Gene: 54806Human Entrez Gene: 52906Mouse Entrez Gene: 308923Rat Omim: 608894Human SwissProt: Q8N157Human SwissProt: Q8K3E5Mouse SwissProt: Q6DTM3Rat Unigene: 386684Human Unigene: 253280Mouse Unigene: 155144Rat **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications. 脑组织**表达**