Recombinant Human AHI1 cell lysate

Cat. No. AHI1-41HCL **Lot. No.** (See product label)

SPECIFICATION

Product Overview	
Species	Human
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.
Size	100 ul
Storage Buffer	1X Sample Buffer (50 mM Tris-HCl, 2% SDS, 10% glycerol, 300 mM 2-mercaptoethanol, 0.01% Bromophenol blue)
Applications	Western Blot;

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;

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	DKFZp686J1653;
Gene ID	54806
mRNA Refseq	NM_001134830
Protein Refseq	NP_001128302
MIM	608894
UniProt ID	Q8N157
Chromosome Location	6q23.2
Function	protein binding;

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