

Product datasheet for **SC309448**

Aspartate beta hydroxylase (ASPH) (NM_020164) Human Untagged Clone

Product data:

Product Type:	Expression Plasmids
Product Name:	Aspartate beta hydroxylase (ASPH) (NM_020164) Human Untagged Clone
Tag:	Tag Free
Symbol:	ASPH
Synonyms:	AAH; BAH; CASQ2BP1; FDLAB; HAAH; JCTN; junctin
Vector:	<u>pCMV6 series</u>
Fully Sequenced ORF:	>NCBI ORF sequence for NM_020164, the custom clone sequence may differ by one or more nucleotides ATGGCTGAAGATAAAGAGACAAAGCATGGAGGACACAAGAATGGGAGGAAAGCGGACTC TCAGGAAC TTCATTCTTCACGTGGTTTATGGTGATTGCATTGCTGGCGTCTGGACATCT GTAGCTGTCGTTTGGTTTGATCTGTTGACTATGAGGAAGTTCTAGCCAAAGCAAAGGAC TTCCGTTATAACTTATCAGAGGTGCTTCAAGGAAAAGTGAAGTCTATGATGCTGATGGT GATGGAGATTTTGTGATGTGGATGATGCCAAAGTTTTATTAGAAGGCCAGTGGGGTAGCC AAGAGAAAAGTAAAGCTAAAGTAAAGAACTCACTAAAGAAGAGCTCAAGAAGGAGAAA GAGAAACCTGAGTCAAGGAAGGAAAGTAAAGTGAAGAGAGAAAAAGGGGAAGAAGAG GATGTCCGAAAGGATAAGAAAATTGCTGATGCAGACCTATCCAGGAAGGAGTCTCCTAAG GGTAAAAAGGACAGAGAAAAAGAGAAAGTGGACCTAGAAAAAAGTGTAAAACCAAGGAA AATAGGAAAAATCAACAAATATGAAGGATGTTTCTAGTAAAAATGGCATCCCGAGACAAA GATGACAGAAAGGAAAGTAGAAGTTCTACCAGATATGCACACTTAACAAAGGAAATACC CAGAAAAGAAACGGCTAA
Restriction Sites:	Please inquire
ACCN:	NM_020164
OTI Disclaimer:	Our molecular clone sequence data has been matched to the reference identifier above as a point of reference. Note that the complete sequence of our molecular clones may differ from the sequence published for this corresponding reference, e.g., by representing an alternative RNA splicing form or single nucleotide polymorphism (SNP).
OTI Annotation:	This TrueClone is provided through our Custom Cloning Process that includes sub-cloning into OriGene's pCMV6 vector and full sequencing to provide a non-variant match to the expected reference without frameshifts, and is delivered as lyophilized plasmid DNA.
Components:	The ORF clone is ion-exchange column purified and shipped in a 2D barcoded Matrix tube containing 10ug of transfection-ready, dried plasmid DNA (reconstitute with 100 ul of water).



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Reconstitution Method:

1. Centrifuge at 5,000xg for 5min.
2. Carefully open the tube and add 100ul of sterile water to dissolve the DNA.
3. Close the tube and incubate for 10 minutes at room temperature.
4. Briefly vortex the tube and then do a quick spin (less than 5000xg) to concentrate the liquid at the bottom.
5. Store the suspended plasmid at -20°C. The DNA is stable for at least one year from date of shipping when stored at -20°C.

RefSeq: [NM_020164.2](#), [NP_064549.1](#)

RefSeq Size: 1254 bp

RefSeq ORF: 678 bp

Locus ID: 444

UniProt ID: [Q12797](#)

Cytogenetics: 8q12.3

Protein Families: Druggable Genome, Transmembrane

Gene Summary: This gene is thought to play an important role in calcium homeostasis. The gene is expressed from two promoters and undergoes extensive alternative splicing. The encoded set of proteins share varying amounts of overlap near their N-termini but have substantial variations in their C-terminal domains resulting in distinct functional properties. The longest isoforms (a and f) include a C-terminal Aspartyl/Asparaginyl beta-hydroxylase domain that hydroxylates aspartic acid or asparagine residues in the epidermal growth factor (EGF)-like domains of some proteins, including protein C, coagulation factors VII, IX, and X, and the complement factors C1R and C1S. Other isoforms differ primarily in the C-terminal sequence and lack the hydroxylase domain, and some have been localized to the endoplasmic and sarcoplasmic reticulum. Some of these isoforms are found in complexes with calsequestrin, triadin, and the ryanodine receptor, and have been shown to regulate calcium release from the sarcoplasmic reticulum. Some isoforms have been implicated in metastasis. [provided by RefSeq, Sep 2009]

Transcript Variant: This variant (5) represents use of an alternate promoter and 5' UTR, uses a distinct start codon, includes an alternate in-frame exon, and uses a distinct 3' splice pattern that lacks many coding exons, compared to variant 1. The encoded isoform (e) has a slightly shorter and distinct N-terminus and some overlap with the N-terminus of isoform a. This isoform lacks the catalytic domain and enzymatic function of isoform a. This isoform has also been referred to as junctin isoform 1 and forms a complex with calsequestrin, triadin, and the ryanodine receptor by direct interaction at the C-terminal part of the molecule. This protein appears to stabilize the complex and plays a crucial role in the regulation of calcium release from the sarcoplasmic reticulum. This variant is expressed in cardiac and skeletal muscle.

Sequence Note: This RefSeq record was created from transcript and genomic sequence data to make the sequence consistent with the reference genome assembly. The genomic coordinates used for the transcript record were based on transcript alignments.