UNC13D cDNA Clone – Unc-13 Homolog D (C. Elegans) (Human)

PRODUCT OVERVIEW

Product Name	UNC13D, cDNA Clone
Full Product Name	UNC13D cDNA Clone
Product Gene Name	UNC13D cdna clone
Sequence	atggcgacac tcctctccca tccgcagcag cgccctccct tcttgcgcca ggccatcaag ataaggcgcc gcagagtcag agatctacag gatcccccgc cccaaatggc cccggagatc cagcctccat cccaccactt ctcccccgag cagcgggccc tgctctacga ggacgcactc tacactgtct tgcaccgcct gggtcatcct gagcccaacc atgtgacgga ggcctctgag ctgctgcgat acctgcagga ggccttccac gtggagcccg aggagcacca gcagacactg cagcgggtca gggagcttga gaagccaata ttttgtctga aggcaacagt gaaacaggcc aagggcattc tgggcaaaga tgtcagtggg ttcagcgacc cctactgcct gctgggcatt gagcaggggg taggtgtgcc agggggcagc cccgggtccc ggcatcggca gaaggctgtg gtgaggcaca ccatccccga ggaggagacc caccgcacgc aggtcatcac ccagacactc aaccccgtct gggacgagac cttcatcctg gagtttgagg acatcaccaa tgcgagcttt catctggaca tgtgggacct ggacactgtg gagtctgtcc gacagaagct tggggagctc acggatctgc atgggcttcg caggatcttt aaagaggccc ggaaggacaa aggccaggac gactttctgg ggaacgtggt tctgaggctg caggacctgc gctgccgaga ggaccagtgg taccccctgg aaccccgcac tgagacctac ccagaccgag gccagtgcca cctccagttc caactcatcc ataagcggag agccacttcg gccagccgct cgcagccgag ctacaccgtg cacctccacc tcctgcagca gcttgtgtcc cacgaggtca cccagcacga ggcgggaagc acctcctggg acgggtcgct gagtccccag gctgccaccg tcctctttct gcacgccaca cagaaggacc tatccgactt ccaccagtcc atggcgcagt ggctggccta cagccgcctc taccagagcc tggagttccc cagcagctgc ctcctgcacc ccatcaccag catcgagtac cagtggatcc agggtcggct caaggcagaa cagcaggagg agctggccgc ctcattcagc tccctgctga cctacggcct ctccctcatc cggaggttcc gctctgtctt ccccctctct gtctcggact ccccagcccg gctgcagtct cttctcaggg tcctggtaca gatgtgcaag atgaaggcct ttggagaact gtgccccaac accgccccat tgccccagct ggtgactgag gccctgcaga ctggcaccac tgaatggttc cacctgaagc agcagcacca tcaacccatg gtgcagggca tcccggaggc aggcaaggcc ttgctgggcc tggtacagga tgtcattggc gacctgcacc agtgccagcg cacatgggac aagatcttcc acaataccct caagatccac ctcttctcca tggctttccg ggagctgcag tggctggtgg ccaagcgggt gcaggaccac acgacggttg tgggtgatgt agtgtcccca gagatgggcg agagtctgtt ccagctctac atcagcctca aggagctctg ccagctgcgc atgagctcct cagagaggga tggagtcctg gccctggata atttccaccg ctggttccag ccggccatcc cctcctggct gcagaagacg tacaacgagg ccctggcgcg ggtgcagcgc gctgtgcaga tggatgagct ggtgcccctg ggtgaactga ccaagcacag cacatcagcg gtggatctat ccacctgctt tgcccagatc agccacactg cccggcagct ggactggcca gacccagagg aggccttcat gattaccgtc aagtttgtgg aggacacctg tcgcctggcc ctggtgtact gcagccttat aaaggcccgg gcccgcgagc tctcttcagg ccagaaggac caaggccagg cagccaacat gctgtgtgtg gtggtgaatg acatggagca gctgcggctg gtgatcggca agttgcccgc ccagctggca tgggaggccc tggagcagcg ggtaggggcc gtgctggagc aggggcagct gcagaacacg ctgcatgccc agctgcagag cgcgctggcc gggctgggcc atgagatccg cactggcgtc cgcaccctgg ccgagcagtt ggaggtgggc atcgccaagc acatccagaa actggtgggc gtcagggagt ctgtcctgcc tgaggatgcc attctgcccc tgatgaagtt cctggaggtg gagctttgct acatgaacac caacttggtg caggagaact tcagcagcct cctgaccctg ctctggaccc acacactcac agtgctggtg gaggcggccg cctcccagcg cagctcatcc ctggcttcca acaggctgaa gattgccctg cagaacctgg agatctgctt ccacgctgag ggctgtggcc tgccacccaa ggccctgcac actgccacct tccaggctct gcagagggac ctggagctgc aggcggcctc cagccgggaa ctcatccgga agtacttctg cagccgaatc cagcagcagg cagaaaccac ctctgaggag ctgggggctg tgacagtcaa ggcctcctac cgcgcctctg agcagaagct gcgtgtggag ctgctcagcg cctccagcct gctgcccctg gactccaatg gctccagcga ccccctttgtc cagctgacct tggagcccag gcatgagttc cctgagctgg ccgcccggga gacccagaag cacaagaagg accttcaccc attgtttgat gagacctttg aattcctggt gcctgctgag ccgtgccgca aggctggggc atgcctcctg ctcaccgtgc tggactacga cacgctgggg gccgacgacc tggaaggcga ggccttcctg ccgctgcgtg aggtgcccgg gctgagtggc tctgaggagc ctggtgaggt gcctcagacc cgcctgcccc tcacgtaccc cgcacccaac ggggacccaa tcctgcagct gctggagggc cggaagggtg accgagaagc ccaggtcttt gtgaggctgc ggcggcaccg ggccaagcag gcctcccagc atgccttgcg gccggcaccg tag
Molecular Weight	128,819 Da
OMIM	608897
Vector	Please Inquire
ISO Certification	Manufactured in an ISO 9001:2015 Certified Laboratory.
Related Product Information for UNC13D cdna clone	MBS1267531 is a cDNA clone targeting UNC13D.
Other Notes	Small volumes of UNC13D cdna clone vial(s) may occasionally become entrapped in the seal of the product vial during shipment and storage. If necessary, briefly centrifuge the vial on a tabletop centrifuge to dislodge any liquid in the container`s cap. Certain products may require to ship with dry ice and additional dry ice fee may apply.

ADDITIONAL DETAILS

NCBI/Uniprot data below describe general gene information for UNC13D. It may not necessarily be applicable to this product.

NCBI GI #	45501048
NCBI GeneID	201294
NCBI Accession #	BC067084
UniProt Secondary Accession #	Q9H7K5; B4DWG9
UniProt Related Accession #	Q70J99
NCBI Official Full Name	Homo sapiens unc-13 homolog D (C. elegans), mRNA
NCBI Official Synonym Full Names	unc-13 homolog D
NCBI Official Symbol	UNC13D
NCBI Official Synonym Symbols	FHL3; HLH3; HPLH3; Munc13-4
UniProt Protein Name	Protein unc-13 homolog D
UniProt Synonym Protein Names	Munc13-4
Protein Family	Protein
UniProt Gene Name	UNC13D
UniProt Entry Name	UN13D_HUMAN
NCBI Protein Information	protein unc-13 homolog D
NCBI Summary for UNC13D	This gene encodes a protein that is a member of the UNC13 family, containing similar domain structure as other family members but lacking an N-terminal phorbol ester-binding C1 domain present in other Munc13 proteins. The protein appears to play a role in vesicle maturation during exocytosis and is involved in regulation of cytolytic granules secretion. Mutations in this gene are associated with familial hemophagocytic lymphohistiocytosis type 3, a genetically heterogeneous, rare autosomal recessive disorder. [provided by RefSeq, Jul 2008]
UniProt Comments for UNC13D	UNC13D: Plays a role in cytotoxic granule exocytosis in lymphocytes. Required for both granule maturation and granule docking and priming at the immunologic synapse. Regulates assembly of recycling and late endosomal structures, leading to the formation of an endosomal exocytic compartment that fuses with perforin-containing granules at the immunologic synapse and licences them for exocytosis. Regulates Ca(2+)-dependent secretory lysosome exocytosis in mast cells. Defects in UNC13D are the cause of familial hemophagocytic lymphohistiocytosis type 3 (FHL3); also known as HPLH3. Familial hemophagocytic lymphohistiocytosis (FHL) is a genetically heterogeneous, rare autosomal recessive disorder. It is characterized by immune dysregulation with hypercytokinemia and defective natural killer cell function. The clinical features of the disease include fever, hepatosplenomegaly, cytopenia, hypertriglyceridemia, hypofibrinogenemia, and neurological abnormalities ranging from irritability and hypotonia to seizures, cranial nerve deficits, and ataxia. Hemophagocytosis is a prominent feature of the disease, and a non-malignant infiltration of macrophages and activated T-lymphocytes in lymph nodes, spleen, and other organs is also found. Belongs to the unc-13 family. 3 isoforms of the human protein are produced by alternative splicing. Chromosomal Location of Human Ortholog: 17q25.1 Cellular Component: lysosome Molecular Function: protein binding; Rab GTPase binding Biological Process: positive regulation of exocytosis; regulation of mast cell degranulation Disease: Hemophagocytic Lymphohistiocytosis, Familial, 3
Research Articles on UNC13D	**1.** Munc13-4 conveys Ca(2+) sensitivity to platelet SNARE-mediated membrane fusion and reveal a potential mechanism by which Munc13-4 bridges and stabilizes apposing membranes destined for fusion.

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