PHYH cDNA Clone – Phytanoyl-Coa 2-Hydroxylase (Human)

PRODUCT OVERVIEW

Product Name	PHYH, cDNA Clone
Full Product Name	PHYH cDNA Clone
Product Gene Name	PHYH cdna clone
Sequence	atggagcagc ttcgcgccgc cgcccgtctg cagattgttc tgggccacct cggccgcccc tcggccgggg ctgtcgtagc tcatcccact tcagggacta tttcctctgc cagtttccat cctcaacaat tccagtatac tctggataat aatgttctaa ccctggaaca gagaaaattt tatgaagaaa atgggtttct agtaatcaaa aatcttgtac ctgatgccga tattcaacgc tttcggaatg agtttgaaaa aatctgcaga aaggaggtga aaccattagg attaacagta atgagagatg tgaccatttc gaaatccgaa tatgctccaa gtgagaagat gatcacgaag gtccaggatt tccaggaaga taaggagctc ttcagatact gcactctccc cgagattctg aaatatgtgg agtgcttcac tggacctaat attatggcca tgcacacaat gttgataaac aaacctccag attctggcaa gaagacgtcc cgtcaccccc tgcaccagga cctgcactat ttccccttca ggcccagcga tctcatcgtt tgcgcctgga cggcgatgga gcacatcagc cggaacaacg gctgtctggt tgtgctccca ggcacacaca agggctccct gaagccccac gattacccca agtgggaggg gggagttaac aaaatgttcc acgggatcca ggactacgag gaaaacaagg cccgggtgca cctggtgatg gagaagggcg acactgtttt cttccatcct ttgctcatcc acggatctgg tcagaataaa acccagggat tccggaaggc aatttcctgc catttcgcca gtgccgattg ccactacatt gacgtgaagg gcaccagtca agaaaacatc gagaaggaag ttgtaggaat agcacataaa ttctttggag ctgaaaatag cgtgaacttg aaggatattt ggatgtttcg agctcgactt gtgaaaggag aaagaaccaa tctttga
Molecular Weight	27,291 Da
OMIM	266500
Vector	pENTR223.1 or pUC
Clone Sequence Report	Provided with product shipment
ISO Certification	Manufactured in an ISO 9001:2015 Certified Laboratory.
Related Product Information for PHYH cdna clone	MBS1267960 is a cDNA clone targeting PHYH.
Other Notes	Small volumes of PHYH 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 PHYH. It may not necessarily be applicable to this product.

NCBI GI #	20809598
NCBI GeneID	5264
NCBI Accession #	BC029512
UniProt Secondary Accession #	A8MTS8; B1ALH5
UniProt Related Accession #	O14832
NCBI Official Full Name	Homo sapiens phytanoyl-CoA 2-hydroxylase, mRNA
NCBI Official Synonym Full Names	phytanoyl-CoA 2-hydroxylase
NCBI Official Symbol	PHYH
NCBI Official Synonym Symbols	RD; LN1; PAHX; LNAP1; PHYH1
UniProt Protein Name	Phytanoyl-CoA dioxygenase, peroxisomal

UniProt Synonym Protein Names	Phytanic acid oxidase; Phytanoyl-CoA alpha-hydroxylase; PhyH
Protein Family	Phytanoyl-CoA dioxygenase
UniProt Gene Name	PHYH
UniProt Synonym Gene Names	PAHX; PhyH
UniProt Entry Name	PAHX_HUMAN
NCBI Protein Information	phytanoyl-CoA dioxygenase, peroxisomal
NCBI Summary for PHYH	This gene is a member of the PhyH family and encodes a peroxisomal protein that is involved in the alpha-oxidation of 3-methyl branched fatty acids. Specifically, this protein converts phytanoyl-CoA to 2-hydroxyphytanoyl-CoA. Mutations in this gene have been associated with Refsum disease (RD) and deficient protein activity has been associated with Zellweger syndrome and rhizomelic chondrodysplasia punctata. Alternate transcriptional splice variants, encoding different isoforms, have been characterized. [provided by RefSeq, Jul 2008]
UniProt Comments for PHYH	PHYH: Converts phytanoyl-CoA to 2-hydroxyphytanoyl-CoA. Defects in PHYH are a cause of Refsum disease (RD). RD is an autosomal recessive disorder characterized clinically by a tetrad of abnormalities: retinitis pigmentosa, peripheral neuropathy, cerebellar ataxia, and elevated protein levels in the cerebrospinal fluid (CSF). Patients exhibit accumulation of the branched-chain fatty acid, phytanic acid, in blood and tissues. Less constant features are nerve deafness, anosmia, skeletal abnormalities, ichthyosis, cataracts and cardiac impairment. Manifestations of the disease appear in the second or third decade of life. Belongs to the PhyH family. Protein type: EC 1.14.11.18; Oxidoreductase Chromosomal Location of Human Ortholog: 10p13 Cellular Component: peroxisomal matrix; peroxisome Molecular Function: cofactor binding; phytanoyl-CoA dioxygenase activity; protein binding Biological Process: fatty acid alpha-oxidation; isoprenoid metabolic process Disease: Refsum Disease, Classic
Research Articles on PHYH	**1.** Observational study of gene-disease association. (HuGE Navigator)
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