Human PHYH Protein

Catalog Number: **13368-HNAE**

General Information

Gene Name Synonym:

LN1; LNAP1; PAHX; PHYH1; RD

Protein Construction:

A DNA sequence encoding the human PHYH (O14832) (Ser31-Leu338) was expressed, with a N-terminal Met.

Source: Human

Expression Host: E. coli

QC Testing

Purity: > 80 % as determined by SDS-PAGE

Endotoxin:

Please contact us for more information.

Stability:

Samples are stable for up to twelve months from date of receipt at -70 ℃

Predicted N terminal: Met

Molecular Mass:

The recombinant human PHYH consists of 309 amino acids and predicts a molecular mass of 35.6 KDa. It migrates as an approximately 26-32 KDa band in SDS-PAGE under reducing conditions.

Formulation:

Lyophilized from sterile 20mM mops, 10% glycerol, 2mM DDT, 1mM EDTA, 0.2mM PMSF, 0.2M NaCl, pH 7.2

Normally 5 % - 8 % trehalose, mannitol and 0.01% Tween80 are added as protectants before lyophilization. Specific concentrations are included in the hardcopy of COA. Please contact us for any concerns or special requirements.

Usage Guide

Storage:

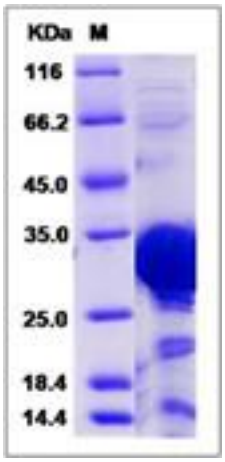
Store it under sterile conditions at -20℃ to -80℃ upon receiving. Recommend to aliquot the protein into smaller quantities for optimal storage.

Avoid repeated freeze-thaw cycles.

Reconstitution:

Detailed reconstitution instructions are sent along with the products.

SDS-PAGE:



Protein Description

PHYH belongs to the family of iron(II)-dependent oxygenases, which typically incorporate one atom of dioxygen into the substrate and one atom into the succinate carboxylate group. PHYH is expressed in liver, kidney, and T-cells, but not in spleen, brain, heart, lung and skeletal muscle. It converts phytanoyl-CoA to 2-hydroxyphytanoyl-CoA. Defects in PHYH can cause 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.

References

1.Mihalik SJ, *et al.* (1997) Identification of PAHX, a Refsum disease gene. Nat Genet. 17(2): 185-9. 2.McDonough MA, *et al.* (2005) Structure of human phytanoyl-CoA 2-hydroxylase identifies molecular mechanisms of Refsum disease. J Biol Chem. 280(49):41101-10. 3.Jansen GA, *et al.* (1998) Characterization of phytanoyl-Coenzyme A hydroxylase in human liver and activity measurements in patients with peroxisomal disorders. Clin Chim Acta. 271 (2):203-11.