

## PHYH Protein, Human, Recombinant

### General Information

|                       |  |
|-----------------------|--|
| Synonyms:             | PHYH1;RD;PAHX;LN1;phytanoyl-CoA 2-hydroxylase;LNAP1  |
| Protein Construction: | A DNA sequence encoding the human PHYH (O14832) (Ser31-Leu338) was expressed, with a N-terminal Met. Predicted N terminal: Met |
| Species:              | Human  |
| Expression Host:      | E. coli  |
| Accession:            | O14832   |
| Molecular Weight:     | 35.6 kDa (predicted); 26-32 kDa (reducing conditions)  |

### QC Testing

|                      |  |
|----------------------|--|
| Biological Activity: | Activity testing is in progress. It is theoretically active, but we cannot guarantee it. If you require protein activity, we recommend choosing the eukaryotic expression version first.   |
| Purity:              | > 80 % as determined by SDS-PAGE   |
| Endotoxin:           | Please contact us for more information.  |
| Formulation:         | Lyophilized from a solution filtered through a 0.22 µm filter, containing 20 mM mops, 10% glycerol, 2 mM DDT, 1 mM EDTA, 0.2 mM PMSF, 0.2M NaCl, pH 7.2. Typically, a mixture containing 5% to 8% trehalose, mannitol, and 0.01% Tween 80 is incorporated as a protective agent before lyophilization. |

### Preparation and Storage

**Reconstitution:**  
A Certificate of Analysis (CoA) containing reconstitution instructions is included with the products. Please refer to the CoA for detailed information.

**Stability & Storage:**

It is recommended to store recombinant proteins at -20°C to -80°C for future use. Lyophilized powders can be stably stored for over 12 months, while liquid products can be stored for 6-12 months at -80°C. For reconstituted protein solutions, the solution can be stored at -20°C to -80°C for at least 3 months. Please avoid multiple freeze-thaw cycles and store products in aliquots.

Actual storage temperature shall be subject to the COA.

**Shipping:**

In general, lyophilized powders are shipped with blue ice, while solutions are shipped with dry ice.

### Protein Background

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.

### Reference

Mihalik SJ, et al. (1997) Identification of PAHX, a Refsum disease gene. *Nat Genet.* 17(2): 185-9.

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.

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.

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