



Recombinant Protein Technical Manual

Recombinant Human PHYH Protein

RPES0130

Product Data:

Product SKU: RPES0130

Size: 20µg

Species: Human

Expression host: E. coli

Uniprot: O14832

Protein Information:

Molecular Mass: 35.6 kDa

AP Molecular Mass: 26-32 kDa

Tag:

Bio-activity:

Purity: > 80 % as determined by reducing SDS-PAGE.

Endotoxin: Please contact us for more information.

Storage: Lyophilized proteins are stable for up to 12 months when stored at -20 to -80°C. Reconstituted protein solution can be stored at 4-8°C for 2-7 days. Aliquots of reconstituted samples are stable at < -20°C for 3 months.

Shipping: This product is provided as lyophilized powder which is shipped with ice packs.

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

Reconstitution: Please refer to the printed manual for detailed information.

Application:

Synonyms: LN1;LNAP1;PAHX;PHYH1;RD

Immunogen Information:

Sequence: Ser31-Leu338

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.