

## PTS Protein, Human, Recombinant (His)

### General Information

Synonyms:	PTPS;PTS;6-pyruvoyltetrahydropterin synthase
Protein Construction:	A DNA sequence encoding the mature form of human PTS (Q03393) (Met1-Glu145) was expressed with a polyhistide tag at the N-terminus. Predicted N terminal: His
Species:	Human
Expression Host:	E. coli
Accession:	Q03393
Molecular Weight:	18.2 kDa (predicted); 27 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:	> 85 % as determined by SDS-PAGE
Endotoxin:	Please contact us for more information.
Formulation:	Supplied as sterile PBS, 40% Glycerol, pH, 7.4.

### 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 the product under sterile conditions at -20°C to -80°C. Samples are stable for up to 12 months. Please avoid multiple freeze-thaw cycles and store products in aliquots.

Actual storage temperature shall be subject to the COA.

#### Shipping:

Proteins are shipped with blue ice.

### Protein Background

PTS(6-pyruvoyltetrahydropterin synthase) belongs to the PTPS family. It catalyzes the elimination of inorganic triphosphate from dihydroneopterin triphosphate, which is the second and irreversible step in the biosynthesis of tetrahydrobiopterin from GTP. Tetrahydrobiopterin, also known as BH(4), is an essential cofactor and regulator of various enzyme activities, including enzymes involved in serotonin biosynthesis and NO synthase activity. Mutations in this gene result in hyperphenylalaninemia. PTS is involved in the biosynthesis of tetrahydrobiopterin, an essential cofactor of aromatic amino acid hydroxylases. PTS also catalyzes the transformation of 7,8-dihydroneopterin triphosphate into 6-pyruvoyl tetrahydropterin. Defects in PTS are the cause of BH4-deficient hyperphenylalaninemia type A (HPABH4A), also called 6-pyruvoyl-tetrahydropterin synthase deficiency (PTS

deficiency) or hyperphenylalaninemia tetrahydrobiopterin-deficient due to PTS deficiency. HPABH4A is an autosomal recessive disorder characterized by depletion of the neurotransmitters dopamine and serotonin, and clinically by severe neurological symptoms unresponsive to the classic phenylalanine-low diet.

### Reference

Ashida A, et al. (1994) A missense mutation (A to G) of 6-pyruvoyltetrahydropterin synthase in tetrahydrobiopterin-deficient form of hyperphenylalaninemia. *Genomics*. 24:408-10.

Ashida A, et al. (1993) cDNA cloning, expression in *Escherichia coli* and purification of human 6-pyruvoyl-tetrahydropterin synthase. *Biochem. Biophys Res Commun*. 195:1386-93.

Thoeny B, et al. (1992) Human 6-pyruvoyltetrahydropterin synthase: cDNA cloning and heterologous expression of the recombinant enzyme. *Biochem Biophys Res Commun*. 189:1437-43.

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