Recombinant Human Hydroxymethylbilane Synthase, His-tagged

Cat. No. HMBS-1929H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant humanHMBS protein, fused to His-tag at N-terminus, was expressed in E. coli andpurified by using conventional chromatography techniques.
Species	Human
Description	Porphobilinogendeaminase, also known as HMBS, is a member of the hydroxymethylbilanesynthase superfamily. It is a cytoplasmic enzyme found in the heme synthesispathway. Deficiency of HMBS causes errors in pyrrole metabolism which in turnleads to an inherited autosomal disorder called acute intermittent porphyria(AIP) which is characterized by acute attacks of neurological dysfunctionswith hypertension, tachycardia, peripheral neurologic disturbances, abdominalpain and excessive amounts of aminolevulinic acid and porphobilinogen in theurine.
Form	Liquid. In 20mMTris-HCl buffer (pH 8.0) containing 1mM DTT, 10% glycerol, 0.1M NaCl.
Molecular Weight	41.9kDa (385aa)confirmed by MALDI-TOF.
Purity	>95%by SDS-PAGE
Concentration	1 mg/ml(determined by Bradford assay)
Sequences of	MGSSHHHHHHSSGLVPRGSH MGSHMSGNGN AAATAEENSP KMRVIRVGTR KS

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aminoacids	QLARIQTD SVVATLKASYPGLQFEIIAM STTGDKILDT ALSKIGEKSL FTKELEHALE KNEVDLVVHSLKDLPTVLPP GFTIGAICKR ENPHDAVVFH PKFVGKTLET LPEKSV VGTS SLRRAAQLQR KFPHLEFRSIRGNLNTRLRK LDEQQEFSAI ILATAGLQRM G WHNRVGQIL HPEECMYAVG QGALGVEVRA KDQDILDLVGVLHDPETLLRCIAERA FLRHLEGGCSVPVA VHTAMKDGQL YLTGGVWSLD GSDSIQETMQ ATIHVPAQHE DGPEDDPQLV GITARNIPRGPQLAAQNLGI SLANLLLSKG AKNILDVARQ LNDAH
Storage	Can be storedat +4°C short term (1-2 weeks). For long term storage, aliquot and store at-20°C or -70°C. Avoid repeated freezing and thawing cycles.
Pathways	Heme Biosynthesis; Metabolic pathways;Metabolism; Metabolism of porphyrins; Porphyrin and chlorophyll metabolism

GENE INFORMATION

Gene Name	HMBS hydroxymethylbilane synthase[Homo sapiens]
Official Symbol	HMBS
Synonyms	HMBS; hydroxymethylbilane synthase; UPS;PBGD; PORC; PBG-D; porphobilinogen deaminase; uroporphyrinogen I synthase;pre-uroporphyrinogen synthase; uroporphyrinogen I synthetase; porphyria,acute; Chester type; EC 2.5.1.61
Gene ID	3145
mRNA Refseq	NM_000190
Protein Refseq	NP_000181
MIM	609806

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UniProt ID	P08397
Chromosome Location	11q23.3
Function	hydroxymethylbilane synthase activity;transferase activity
Rendering based on PDB 3ECR.	

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