

Recombinant Human HMBS

Cat. No. HMBS-29326TH **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant full length Human HMBS with a proprietary N terminal tag. Predicted MW 65.82kDa
Species	Human
Source	Wheat Germ
ProteinLength	361 amino acids
Description	This gene encodes a member of the hydroxymethylbilane synthase superfamily. The encoded protein is the third enzyme of the heme biosynthetic pathway and catalyzes the head to tail condensation of four porphobilinogen molecules into the linear hydroxymethylbilane. Mutations in this gene are associated with the autosomal dominant disease acute intermittent porphyria. Alternatively spliced transcript variants encoding different isoforms have been described.
Molecular Weight	65.820kDa inclusive of tags
Tissue specificity	Isoform 1 is ubiquitously expressed. Isoform 2 is found only in erythroid cells.
Form	Liquid
Purity	Proprietary Purification
Storage buffer	pH: 8.00Constituents:0.3% Glutathione, 0.79% Tris HCl

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Storage	Shipped on dry ice. Upon delivery aliquot and store at -80oC. Avoid freeze / thaw cycles.
Sequences of amino acids	MSGNGNAAATAEENSPKMRVIRVGTRKSQLARIQTDSVVA TLKASYPGLQFEIIAMS TTGDKIPDTALSKIGEKSFLTKE LEHALEKNEVDLVVHSLKDLPTVLP PGFTIGAICKR ENPH DAVVFHPKFVVGKTLETLP EKSVVGTSSLRRAAQLQRKFPH LEFRSIRGNLNT RLRKLDEQQEFSAILATAGLQRMGWHN RVGQILHPEECMYAVGQ GALGVEVR AK DQDILDVLGV LHD PETLLRCIAERAFLRHLEGGCSVPVAVHTAMKDGQLYLTG GVW SLDGSDSIQETMQATIHVPAQHEDGPEDDPQLVGITA RNIPRGPQLAAQNLGISLAN LLLSKGAKNILDVARQLNDA H
Sequence Similarities	Belongs to the HMBS family.
GENE INFORMATION	
Gene Name	HMBS hydroxymethylbilane synthase [Homo sapiens]
Official Symbol	HMBS
Synonyms	HMBS; hydroxymethylbilane synthase; PBGD, PORC, porphobilinogen deaminase , porphyria, acute; Chester type , UPS, uroporphyrinogen I synthase; porphobilinogen deaminase;
Gene ID	3145
mRNA Refseq	NM_000190
Protein Refseq	NP_000181
MIM	609806

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Uniprot ID	P08397
Chromosome Location	11q23.2-qter
Pathway	Heme Biosynthesis, organism-specific biosystem; Heme biosynthesis, organism-specific biosystem; Metabolic pathways, organism-specific biosystem; Metabolism, organism-specific biosystem; Metabolism of porphyrins, organism-specific biosystem;
Function	hydroxymethylbilane synthase activity; hydroxymethylbilane synthase activity; transferase activity;

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