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Rabbit Anti-HMBS antibody

SL9068R

Product Name:	HMBS
Chinese Name:	□卜胆原脱氨酶抗体
Alias:	HEM3_HUMAN; HMBS; Hydroxymethylbilane synthase; PBG D; PBG-D; PBGD; Porphobilinogen deaminase; Pre uroporphyrinogen synthase; Pre-uroporphyrinogen synthase; UPS; Uroporphyrinogen I synthase; Uroporphyrinogen I synthetase.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Chicken,Pig,Cow,Rabbit,Sheep,Cat,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:50-200（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	39kDa
Cellular localization:	cytoplasmic
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human HMBS:21-120/361
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
PubMed:	PubMed
Product Detail:	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. [provided by RefSeq, Jul 2008]

Function:
Tetrapolymerization of the monopyrrole PBG into the hydroxymethylbilane pre-uroporphyrinogen in several discrete steps.

Subcellular Location:
Cytoplasm.

Tissue Specificity:
Isoform 1 is ubiquitously expressed. Isoform 2 is found only in erythroid cells.

DISEASE:
Defects in HMBS are the cause of acute intermittent porphyria (AIP) [MIM:176000]. AIP is a form of porphyria. Porphyrias are inherited defects in the biosynthesis of heme, resulting in the accumulation and increased excretion of porphyrins or porphyrin precursors. They are classified as erythropoietic or hepatic, depending on whether the enzyme deficiency occurs in red blood cells or in the liver. AIP is an autosomal dominant form of hepatic porphyria characterized by acute attacks of neurological dysfunctions with abdominal pain, hypertension, tachycardia, and peripheral neuropathy. Most attacks are precipitated by drugs, alcohol, caloric deprivation, infections, or endocrine factors.

Similarity:
Belongs to the HMBS family.

SWISS:
P08397

Gene ID:
3145

Database links:

Entrez Gene: 3145 Human

Entrez Gene: 15288 Mouse

Entrez Gene: 396581 Pig

Entrez Gene: 25709 Rat

Omim: 609806 Human

SwissProt: P08397 Human

	SwissProt: P22907 Mouse SwissProt: P19356 Rat Unigene: 82609 Human Unigene: 247676 Mouse Unigene: 11080 Rat **Important Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications. 卟胆原脱氨酶，为血红素等四吡咯环化合物合成通路中的催化酶。PBGD催化卟胆原(Porphobilingen，PBG)底物的线性四聚化，反应过程中四分子卟胆原底物有序性的共价连接到酶的辅基上，最后水解形成具有四联吡咯环结构的尿卟啉原前体。PBGD的活力缺陷会引起急性间歇性卟啉症(Acute Intermittant Porphyria，AIP)的发生。