

HMBS Polyclonal Antibody

Purified Rabbit Polyclonal Antibody (Pab) Catalog # AP59129

Product Information

Application IHC-P, IHC-F, IF, E

Primary Accession P08397

Reactivity Rat, Pig, Cat, Bovine

Host Rabbit
Clonality Polyclonal
Calculated MW 39330
Physical State Liquid

Immunogen KLH conjugated synthetic peptide derived from human HMBS

Epitope Specificity 21-120/361 **Isotype** IgG

Purity affinity purified by Protein A

Buffer 0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.

SUBCELLULAR LOCATION Cytoplasm.

SIMILARITY Belongs to the HMBS family.

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.

Important Note This product as supplied is intended for research use only, not for use in

human, therapeutic or diagnostic applications.

Background Descriptions 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]

Additional Information

Gene ID 3145

Other Names Porphobilinogen deaminase, PBG-D, 2.5.1.61, Hydroxymethylbilane synthase,

HMBS, Pre-uroporphyrinogen synthase, HMBS, PBGD, UPS

Target/Specificity Isoform 1 is ubiquitously expressed. Isoform 2 is found only in erythroid cells.

Dilution IHC-P=1:100-500,IHC-F=1:100-500,IF=1:50-200,ELISA=1:5000-10000

Storage Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name HMBS

Synonyms PBGD, UPS

Function As part of the heme biosynthetic pathway, catalyzes the sequential

polymerization of four molecules of porphobilinogen to form hydroxymethylbilane, also known as preuroporphyrinogen (PubMed:18004775, PubMed:18936296, PubMed:19138865, PubMed:23815679). Catalysis begins with the assembly of the dipyrromethane cofactor by the apoenzyme from two molecules of

porphobilinogen or from preuroporphyrinogen. The covalently linked cofactor

acts as a primer, around which the tetrapyrrole product is assembled

(PubMed:<u>18936296</u>). In the last step of catalysis, the product,

preuroporphyrinogen, is released, leaving the cofactor bound to the

holodeaminase intact (PubMed: 18936296).

Cytoplasm, cytosol {ECO:0000250 | UniProtKB:P22907}

Tissue Location [Isoform 1]: Is ubiquitously expressed.

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.