

HMBS Antibody (monoclonal) (M01)

Mouse monoclonal antibody raised against a full length recombinant HMBS. Catalog # AT2381a

Product Information

| Application | WB, E |
|-------------------|-----------------|
| Primary Accession | <u>P08397</u> |
| Other Accession | <u>BC000520</u> |
| Reactivity | Human |
| Host | mouse |
| Clonality | monoclonal |
| Isotype | IgG2a Kappa |
| Clone Names | 3E9 |
| Calculated MW | 39330 |

Additional Information

| Gene ID | 3145 |
|--------------------|---|
| Other Names | Porphobilinogen deaminase, PBG-D, Hydroxymethylbilane synthase, HMBS, Pre-uroporphyrinogen synthase, HMBS, PBGD, UPS |
| Target/Specificity | HMBS (AAH00520.1, 1 a.a. ~ 361 a.a) full-length recombinant protein with GST tag. MW of the GST tag alone is 26 KDa. |
| Dilution | WB~~1:500~1000 E~~N/A |
| Format | Clear, colorless solution in phosphate buffered saline, pH 7.2 . |
| Storage | Store at -20°C or lower. Aliquot to avoid repeated freezing and thawing. |
| Precautions | HMBS Antibody (monoclonal) (M01) is for research use only and not for use in diagnostic or therapeutic procedures. |

Background

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.

References

Variation at the NFATC2 Locus Increases the Risk of Thiazolinedinedione-Induced Edema in the Diabetes

REduction Assessment with ramipril and rosiglitazone Medication (DREAM) Study. Bailey SD, et al. Diabetes Care, 2010 Jul 13. PMID 20628086.[Molecular genetic study of acute intermittent porphyria in Russia: mutation analysis and functional polymorphism search in porphobilinogen deaminase gene] Surin VL, et al. Genetika, 2010 Apr. PMID 20536026.Gene-centric association signals for lipids and apolipoproteins identified via the HumanCVD BeadChip. Talmud PJ, et al. Am J Hum Genet, 2009 Nov. PMID 19913121.Acute intermittent porphyria--impact of mutations found in the hydroxymethylbilane synthase gene on biochemical and enzymatic protein properties. Ulbrichova D, et al. FEBS J, 2009 Apr. PMID 19292878.Structure of human porphobilinogen deaminase at 2.8 A: the molecular basis of acute intermittent porphyria. Gill R, et al. Biochem J, 2009 Apr 28. PMID 19207107.





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