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[Marker Primary Antibody]

Rabbit Anti-GAMT/FITC Conjugated antibody

Catalog Number: SL13278R-FITC

FOR RESEARCH USE ONLY

[DATASHEET]

Host:	Rabbit
Isotype:	IgG
Clonality:	Polyclonal
Target:	Anti-GAMT/FITC
Immunogen:	KLH conjugated synthetic peptide derived from human GAMT
Purification:	affinity purified by Protein A
Concentration:	1mg/ml
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 1M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.
Applications:	ICC=1:50-200 IF=1:50-200 not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Reactivity:	Human,Mouse,Rat(predicted:Dog,Pig)
Background:	background: In the creatine biosynthesis pathway, glycine is converted to guanidinoacetate by amidinotransferase, and guanidinoacetate is then converted to creatine by Guanidinoacetate N-methyltransferase (GAMT). GAMT, a methyltransferase, uses S-adenosylmethionine as the methyl donor for this reaction. Methyltransferases are a type of transferase enzyme which transfers a methyl group to nucleic bases in DNA or amino acids in protein. Encoding a 236 amino acid protein, the human GAMT gene maps to chromosome 19p13.3. Defects in the GAMT gene leads to GAMT deficiency, which is associated with guanidinoacetate accumulation and decreased levels of creatine excretion in brain. Such biochemical changes are thought to lead to various neurological syndromes and muscular hypotonia. **Tissue Specificity:** Expressed in liver. DISEASE: Defects in GAMT are the cause of guanidinoacetate methyltransferase deficiency (GAMT deficiency) [MIM:612736]. GAMT deficiency is an autosomal recessive disorder characterized by developmental

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	delay/regression, mental retardation, severe disturbance of expressive and cognitive speech, intractable seizures and movement disturbances, severe depletion of creatine/phosphocreatine in the brain, and accumulation of guanidinoacetic acid (GAA) in brain and body fluids. **Similarity:** Belongs to the RMT2 methyltransferase family. **Database links:** Clone ID: NP_000147.1 Human **Entrez Gene:** 2593 Human **Entrez Gene:** 25257 Rat Omim: 601240 Human **SwissProt:** Q14353 Human **SwissProt:** P10868 Rat **Unigene:** 81131 Human **Unigene:** 33890 Rat Important **Note:** This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.