

Recombinant Human GAMT cell lysate

Cat. No. GAMT-684HCL Lot. No. (See product label)

SPECIFICATION

Species	Human
Description	The protein encoded by this gene is a methyltransferase that converts guanidoacetate to creatine, using S-adenosylmethionine as the methyl donor. Defects in this gene have been implicated in neurologic syndromes and muscular hypotonia, probably due to creatine deficiency and accumulation of guanidinoacetate in the brain of affected individuals. Two transcript variants encoding different isoforms have been described for this gene.
Size	100 ul
Storage Buffer	1X Sample Buffer (50 mM Tris-HCl, 2% SDS, 10% glycerol, 300 mM 2-mercaptoethanol, 0.01% Bromophenol blue)
Applications	Western Blot;

GENE INFORMATION

Gene Name	GAMT guanidinoacetate N-methyltransferase [Homo sapiens]
Official Symbol	GAMT
Synonyms	GAMT; guanidinoacetate N-methyltransferase; PIG2; TP53I2;
Gene ID	2593

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mRNA Refseq	NM_000156
Protein Refseq	NP_000147
MIM	601240
UniProt ID	Q14353
Chromosome Location	19p13.3
Pathway	Arginine and proline metabolism, organism-specific biosystem; Arginine and proline metabolism, conserved biosystem; Creatine metabolism, organism-specific biosystem; Creatine pathway, organism-specific biosystem; Creatine pathway, conserved biosystem; Glycine, serine and threonine metabolism, organism-specific biosystem; Glycine, serine and threonine metabolism, conserved biosystem;
Function	guanidinoacetate N-methyltransferase activity; methyltransferase activity; transferase activity;

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