

## Recombinant Human GAMT cell lysate

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

### SPECIFICATION

#### Product Overview

|                |                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
|----------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| 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;  |

 Tel: 1-631-559-9269    1-516-512-3133

 Email: [info@creative-biomart.com](mailto:info@creative-biomart.com)     Fax: 1-631-938-8127

 45-1 Ramsey Road, Shirley, NY 11967, USA

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|----------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|
| <b>Gene ID</b>             | 2593                                                                                                                                                                                                                                                                                                                                                                                              |
| <b>mRNA Refseq</b>         | NM_000156                                                                                                                                                                                                                                                                                                                                                                                         |
| <b>Protein Refseq</b>      | NP_000147                                                                                                                                                                                                                                                                                                                                                                                         |
| <b>MIM</b>                 | 601240                                                                                                                                                                                                                                                                                                                                                                                            |
| <b>UniProt ID</b>          | Q14353                                                                                                                                                                                                                                                                                                                                                                                            |
| <b>Chromosome Location</b> | 19p13.3                                                                                                                                                                                                                                                                                                                                                                                           |
| <b>Pathway</b>             | 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; |
| <b>Function</b>            | guanidinoacetate N-methyltransferase activity; methyltransferase activity; transferase activity;                                                                                                                                                                                                                                                                                                  |