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# Product datasheet for TP721002XL

## GAMT (NM_000156) Human Recombinant Protein

### Product data:

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|---|---|
| **Product Type:** | Recombinant Proteins |
| **Description:** | Purified recombinant protein of Human guanidinoacetate N-methyltransferase (GAMT), transcript variant 1 |
| **Species:** | Human |
| **Expression Host:** | E. coli |
| **Expression cDNA Clone or AA Sequence:** | Met1-Gly236 |
| **Tag:** | N-His&C-His |
| **Predicted MW:** | 29.5 kDa |
| **Purity:** | >95% as determined by SDS-PAGE and Coomassie blue staining |
| **Buffer:** | Provided lyophilized from a 0.2 μm filtered solution of 20 mM Tris-HCl, 150 mM NaCl |
| **Endotoxin:** | Endotoxin level is < 0.1 ng/μg of protein (< 1 EU/μg) |
| **Storage:** | Store at -80°C. |
| **Stability:** | Stable for at least 3 months from date of receipt under proper storage and handling conditions. |
| **RefSeq:** | NP_000147 |
| **Locus ID:** | 2593 |
| **UniProt ID:** | Q14353, V9HWB2 |
| **RefSeq Size:** | 1138 |
| **Cytogenetics:** | 19p13.3 |
| **RefSeq ORF:** | 708 |
| **Synonyms:** | CCDS2; HEL-S-20; PIG2; TP53I2 |

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| **Summary:** | 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. Pseudogenes of this gene are found on chromosomes 2 and 13. [provided by RefSeq, Feb 2012] |
| **Protein Families:** | Druggable Genome |
| **Protein Pathways:** | Arginine and proline metabolism, Glycine, serine and threonine metabolism, Metabolic pathways |