

Recombinant Human GAMT Protein (His Tag)

Catalog Number: PKSH032515

Note: Centrifuge before opening to ensure complete recovery of vial contents.

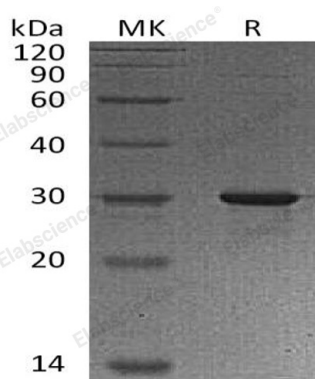
Description

Species	Human
Mol_Mass	29.5 kDa
Accession	Q14353
Bio-activity	Not validated for activity

Properties

Purity	> 90 % as determined by reducing SDS-PAGE.
Endotoxin	< 1.0 EU per µg of the protein as determined by the LAL method.
Storage	Store at < -20°C, stable for 6 months. Please minimize freeze-thaw cycles.
Shipping	This product is provided as liquid. It is shipped at frozen temperature with blue ice/gel packs. Upon receipt, store it immediately at < -20°C.
Formulation	Supplied as a 0.2 µm filtered solution of 20mM Tris-HCl, 1mM DTT, pH 8.0.
Reconstitution	Not Applicable

Data



> 90 % as determined by reducing SDS-PAGE.

Background

GAMT is a methyltransferase which belongs to the class I-like SAM-binding methyltransferase superfamily. It contains one RMT2 (arginine N-methyltransferase 2-like) domain and is expressed in liver. GAMT converts guanidoacetate to creatine, using S-adenosylmethionine as the methyl donor. Defects in GAMT are the cause of guanidinoacetate methyltransferase deficiency, which is an autosomal recessive disorder characterized by developmental 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 in brain and body fluids.

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