DISEASE:
Guanidinoacetate methyltransferase deficiency

NAME:	Guanidinoacetate methyltransferase deficiency
DESCRIPTION:	Guanidinoacetate methyltransferase (GAMT) deficiency is a creatine deficiency syndrome characterized by global developmental delay/intellectual disability (DD/ID), prominent speech delay, autistic/hyperactive behavioral disorders, seizures, and various types of pyramidal and/or extra-pyramidal manifestations.
ORPHACODE:	382
SYNONYMS:	GAMT deficiency
XREF(S):	Orphanet MeSH OMIM ICD-10
ANALYTE(S):	GAMT
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Creatine deficiency by Guanidinoacetate methyltransferase deficiency (2 genes)

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman

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