DISEASE:
L-Arginine:glycine amidinotransferase deficiency

NAME:	L-Arginine:glycine amidinotransferase deficiency
DESCRIPTION:	L-Arginine:glycine amidinotransferase (AGAT) deficiency is a very rare type of creatine deficiency sydrome characterized by global developmental delay, intellectual disability, and myopathy.
ORPHACODE:	35704
SYNONYMS:	AGAT deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	GATM
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

Related Analytes

- glycine amidinotransferase