

## Opis choroby \*

### Definicja

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.

### Dane

Klasyfikacja	Synonimy
Choroba	GAMT deficiency
	Niedobór GAMT
<b>Kod ORPHA</b>	<b>Kod OMIM</b>
382	612736
<b>Kod ICD11</b>	<b>Kod ICD10</b>
5C53.4	E72.8

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### \*Źródło

orphanet