

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

Choroba

Synonimy

GAMT deficiency

Niedobór GAMT

Kod ORPHA

382

Kod OMIM

612736

Kod ICD10

E72.8

Kod ICD11

5C53.4

*Źródło

orphanet