Opis choroby *

Definicja

L-Arginine:glycine amidinotransferase (AGAT) deficiency is a very rare type of creatine deficiency sydrome characterized by global developmental delay, intellectual disability, and myopathy.

Dane

Klasyfikacja
Choroba

Synonimy
AGAT deficiency
Niedobór AGAT

Kod ORPHA
35704

Kod OMIM
612718

Kod ICD10
E72.8

Kod ICD11
5C53.4

---

*Źródło

orphanet