Opis choroby *

Definicja

A rare, genetic, syndromic intellectual disability characterized by global developmental delay, early-onset seizures, cerebellar atrophy, osteopenia, nystagmus and dysmorphic facial features, including bitemporal narrowing, prominent forehead, anteverted nares. Dysarthria, dysmetria, ataxic gait, spasticity and dysmorphic features have also been associated.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
GPAA1-related biosynthesis defect
GPAA1-related biosynthesis defect

Kod ORPHA
529665

Kod OMIM
617810

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet