

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	Synonimy
Zespół wad wrodzonych GPAA1	GPAA1-related biosynthesis defect GPAA1-related biosynthesis defect

Kod ORPHA	Kod OMIM	Kod ICD10
529665	617810	Q87.8

Kod ICD11

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*Źródło

orphanet