

## 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