

## 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<br>GPAA1-related biosynthesis defect |

| Kod ORPHA | Kod OMIM | Kod ICD10 |
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| 529665    | 617810   | Q87.8     |

### Kod ICD11

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### \*Źródło

orphanet