Opis choroby *

Definicja

A rare, genetic, neurological disorder characterized by intrauterine growth retardation, failure to thrive, infantile onset of sensorineural deafness, severe global developmental delay or absent psychomotor development, paraplegia or quadriplegia with dystonia and pyramidal signs, microcephaly, ocular abnormalities (strabismus, optic atrophy), mildly dysmorphic features (deep-set eyes, prominent nasal bridge, micrognathia), seizures and abnormalities of brain morphology (hypomyelinating white matter changes, cerebral atrophy).

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Severe motor and intellectual disabilities-sensorineural hearing loss-dystonia syndrome
Severe motor and intellectual disabilities-sensorineural hearing loss-dystonia syndrome

Kod ORPHA
369939

Kod OMIM
300475

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet