Opis choroby *

Definicja

A rare, genetic intellectual disability syndrome characterized by severe global developmental delay with intellectual disability, microcephaly, growth retardation, ocular defects such as congenital cataract, and nevus flammeus simplex on the forehead. Cardiac, urogenital, and skeletal abnormalities, as well as seizures are present in most patients. Dysmorphic craniofacial features include sparse hair, downslanting palpebral fissures, hypertelorism, broad and overhanging nasal tip and short philtrum, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
464738

Kod OMIM
616449

Kod ICD10
Q87.8

Kod ICD11
-

---

*Źródło

orphanet