Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome with intellectual disability characterized by infantile onset of global developmental delay, severe intellectual disability, growth deficiency, microcephaly, strabismus, blue-gray sclerae, and extensive Mongolian spots. Some patients also present with epilepsy. Brain imaging may demonstrate variable abnormalities including cerebral atrophy, thin corpus callosum, ventriculomegaly, or arachnoid cysts.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
488627

Kod OMIM
617051

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet