

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