Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay and moderate to severe intellectual disability, as well as variable other manifestations, such as macro- or microcephaly, epilepsy, hypotonia, behavioral problems, stereotypic movements, and facial dysmorphism (including arched eyebrows, long palpebral fissures, prominent nasal bridge, upturned nose, dysplastic ears, and broad mouth), among others. Brain imaging may show cerebellar anomalies, hypoplastic corpus callosum, enlarged ventricles, polymicrogyria, or white matter abnormalities.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
500159

Kod OMIM
617751

Kod ICD10
Q87.0

Kod ICD11
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*Źródło

orphanet