Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by variable developmental delay, intellectual disability, early-onset seizures, and facial dysmorphism (including arched eyebrows, long palpebral fissures, prominent nasal bridge, large ears, thin upper lip, and high arched palate). Other reported features are microcephaly, hypotonia, growth retardation, congenital heart defects, and malformations of the fingers and toes, as well as additional neurologic manifestations (such as ataxia or spastic quadriplegia). Brain imaging may show hypoplastic corpus callosum, white matter abnormalities, or cortical atrophy.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
505237

Kod OMIM
617452

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet