Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay, intellectual disability, hypotonia, seizures, and autism spectrum disorder. Variable associated features include ophthalmologic anomalies, congenital heart defects, genitourinary defects, and craniofacial dysmorphism (including frontal bossing, epicanthal folds, low-set, posteriorly rotated ears, anteverted nares, and micrognathia). Brain imaging may show thinning of the corpus callosum, white matter abnormalities, ventriculomegaly, and a small cerebellar vermis.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
494344

Kod OMIM
616975

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet