Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by global developmental delay or regression, variable congenital heart defects (such as patent ductus arteriosus, atrial or ventricular septal defects, and double outlet right ventricle, among others), and dysmorphic features (including ptosis, epicanthal folds, abnormally set/dysplastic ears, low hairline or excess nuchal skin, wide-spaced/inverted nipples, umbilical hernia or diastasis recti, and digital anomalies). Additional variable manifestations are hyper- or hypotonia, seizures, hearing loss, cortical blindness, and optic atrophy. Brain imaging may show cerebral and cerebellar atrophy and hydrocephalus.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
592570

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet