Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by psychomotor and growth delay, severe intellectual disability, microcephaly, and hypoplastic corpus callosum. Additional reported manifestations include increased muscle tonus, seizures, cardiac anomalies, recurrent bronchopneumonia, camptodactyly, preauricular skin tag, and dysmorphic facial features (such as broad forehead, hypertelorism, flat nasal bridge, anteverted nostrils, and prominent ears), among others.

Dane

Klasyfikacja Synonimy

Zespół wad wrodzonych Da Silva syndrome

Zespół Da Silva

Kod ORPHA

Kod OMIM

Kod ICD10

1495

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Q87.8

Kod ICD11

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*Źródło

orphanet