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
Zespół wad wrodzonych

Synonimy
Da Silva syndrome
Zespół Da Silva

Kod ORPHA
1495

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet