Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by intrauterine growth retardation, multiple congenital malformations (such as brain malformations including ectopic neuropituitary gland, hypoplastic adenopituitary, and hypoplastic cerebellar vermis, cardiac and renal anomalies, and postaxial polydactyly), abnormal hair structure with temporal balding, and dysmorphic facial features with hypoplastic nasal bridge, anteverted nostrils, dysplastic ears, long and smooth philtrum, narrow upper lip, and prominent, asymmetric lower lip. Postnatal growth retardation and severe developmental delay have also been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Goossens-Devriendt syndrome
Zespół Goossensa i Devriendta

Kod ORPHA
75389

Kod OMIM
-

Kod ICD10
Q87.8

Kod ICD11
-

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*Źródło

orphanet