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 Synonimy

Zespół wad wrodzonych Goossens-Devriendt syndrome Zespół Goossensa i Devriendta

 Kod ORPHA
 Kod OMIM
 Kod ICD10

 75389
 O87.8

Kod ICD11

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*Źródło

orphanet