Opis choroby *

Definicja

A rare genetic multiple congenital anomalies/dysmorphic syndrome characterized by pre- and postnatal growth restriction, microcephaly, mild to severe intellectual disability, sensorineural hearing loss with cochlear abnormalities, and facial dysmorphism (with small and elongated face, bifrontal narrowing, epicanthus, short nose, small nares, dysplastic ears, and short neck). Additional variable features include limb malformations, cardiac anomalies, abnormal skin pigmentation, and recurrent infections, among others.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
WABS
WABS

Kod ORPHA
280558

Kod OMIM
613398

Kod ICD10
Q87.8

Kod ICD11
LD2F.1Y

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*Źródło

orphanet