Opis choroby *

Definicja

A rare multiple congenital anomalies/dysmorphic syndrome characterized by moderate to severe intellectual disability, neurologic signs and symptoms (such as seizures, spasticity, strabismus), characteristic dysmorphic facial features (including broad forehead, hypertelorism, downslanting palpebral fissures, broad and flat nasal bridge, midline notch of upper lip, lack of upper central incisors, incomplete oral cleft, and prominent mandible), and acne scars. Hearing impairment, pseudo-bulbar palsy, growth retardation, and skeletal anomalies (camptodactyly, clinodactyly, bilateral cubitus valgus, pes cavus/planus) have also been described.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Pallister-W syndrome
Zespół Pallistera i W

Kod ORPHA
2804

Kod OMIM
311450

Kod ICD10
Q87.8

Kod ICD11
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*Źródło

orphanet