Opis choroby *

Definicja

A rare autosomal dominant, multiple congenital anomalies syndrome characterized by facial dysmorphism (flat facial profile with normal calvarium, hypertelorism, small downslanting palpebral fissures, hypoplastic nose with button tip and slitlike nares, and small, pursed mouth), profound sensorineural deafness, ulnar deviations and contractures of the hand. This disorder is allelic to Waardenburg syndrome, and distinguished by the imaging findings and distinct facial features.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
CDHS
CDHS
Zespół Sommera, Young, Wee i Frye
Craniofacial-hearing loss-hand syndrome
Sommer-Young-Wee-Frye syndrome

Kod ORPHA
1529

Kod OMIM
122880

Kod ICD10
Q87.0

Kod ICD11
LD2H.Y

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*Źródło

orphanet