

Opis choroby *

Definicja

Deafness-genital anomalies-metacarpal and metatarsal synostosis syndrome is characterised by sensorineural deafness, bilateral synostosis of the 4th and 5th metacarpals and metatarsals, genital anomalies (hypospadias in males), psychomotor delay and abnormal dermatoglyphics. So far, it has been described in two unrelated patients. Facial dysmorphism was noted in both patients (prominent forehead, ear anomalies, facial asymmetry and an open mouth appearance).

Dane

Klasyfikacja

Zespół wad wrodzonych Hearing loss-genital anomalies-metacarpal and metatarsal synostosis syndrome
Zespół Pfeiffera i Kapferera
Pfeiffer-Kapferer syndrome

Synonimy

Kod ORPHA
3224

Kod OMIM
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Kod ICD10
Q87.8

Kod ICD11
LD2H.Y

*Źródło

orphanet