

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

Kod ORPHA	Kod OMIM	Kod ICD10
3224	-	Q87.8

Kod ICD11
LD2H.Y

*Źródło

orphanet