

Opis choroby *

Definicja

Dysmorphism-short stature-deafness-disorder of sex development syndrome is characterized by dysmorphism (including facial asymmetry, arched eyebrows, hypertelorism, broad and flat nasal bridge, microtia, small nose with anteverted nostrils, micrognathia), deafness, cleft palate, male pseudohermaphroditism, and growth and psychomotor retardation. It has been described in two siblings. It is transmitted as an autosomal recessive trait.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Dysmorphism-short stature-hearing loss-disorder of sex development syndrome
Dysmorfia - niski wzrost - głuchota - pseudohermafrodytyzm
Ieshima-Koeda-Inagaki syndrome
Dysmorphism-short stature-deafness-disorder of sex development syndrome

Kod ORPHA

2282

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet