Opis choroby *

Definicja

A rare developmental defect during embryogenesis malformation syndrome characterized by proportionate short stature, sensorineural deafness, mutism, facial dysmorphism and recurrent infections as a result of abnormal neutrophil chemotaxis. There have been no further descriptions in the literature since 1978.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Short stature-hearing loss-neutrophil dysfunction-dysmorphism syndrome
Zespół Thonga, Douglasa i Ferrante
Thong-Douglas-Ferrante syndrome

Kod ORPHA
2866

Kod OMIM
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Kod ICD10
Q87.1

Kod ICD11
LD2H.Y

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*Źródło

orphanet