

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

Kod ORPHA	Kod OMIM	Kod ICD10
2866	-	Q87.1

Kod ICD11  
LD2H.Y

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### \*Źródło

orphanet