Opis choroby *

Definicja

Teebi-Shaltout syndrome is a rare, genetic, development defect during embryogenesis malformation syndrome characterized by association of characteristic facial features (including abnormal head shape with narrow forehead, hypertelorism, telecanthus, small earlobes, broad nasal bridge and tip, underdeveloped ala nasi, small/wide mouth and high/cleft palate), ectodermal dysplasia (including oligodontia with delayed dentition, slow growing hair and reduced sweating) and skeletal abnormalities including camptodactyly and caudal appendage. Short stature and abnormal palmar creases are additional clinical features.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
3291

Kod OMIM
272950

Kod ICD10
Q82.4

Kod ICD11
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*Źródło

orphanet