Opis choroby *

Definicja

A rare acrofacial dysostosis due to the presence of manifestations not usually seen in Nager syndrome (NS) such as microcephaly, blepharophimosis, microtia, a peculiar beakednose, cleft lip and palate, symmetrical involvement of the thumbs and great toes and developmental delay. It has since been suggested that these features can also be a part of the NS phenotype.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Kennedy-Teebi syndrome
Zespół Kennedy'ego i Teebiego

Kod ORPHA
64542

Kod OMIM
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Kod ICD10
Q75.4

Kod ICD11
LD25.2

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*Źródło

orphanet