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

KlasyfikacjaSynonimyZespół wad wrodzonych Kennedy-Teebi syndromeZespół Kennedy'ego i Teebiego

<u>*Źródło</u>

orphanet