

Opis choroby *

Definicja

Microcephaly-brachydactyly-kyphoscoliosis syndrome is characterized by profound intellectual deficit in association with microcephaly, short stature, brachydactyly type D, a flattened occiput, downslanting palpebral fissures, low-set large ears, a broad prominent nose and kyphoscoliosis. It has been described in three sisters. The disorder is likely to be transmitted as an autosomal recessive trait.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Viljoen-Kallis-Voges syndrome
	Zespół Viljoena, Kallisa i Vogesa

Kod ORPHA	Kod OMIM	Kod ICD10
3433	-	Q87.8

Kod ICD11
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*Źródło

orphanet