Opis choroby *

Definicja

A rare disorder characterized by the association of epiphyseal dysplasia, short stature, microcephaly and, in the first reported cases, congenital nystagmus. So far, less than 10 cases have been described in the literature. Variable degrees of intellectual deficit have also been reported. Other occasional features include retinitis pigmentosa and coxa vara. Transmission appears to be autosomal recessive.

Dane

Klasyfikacja
Choroba

Synonimy
Epiphyseal dysplasia-microcephaly-nystagmus syndrome
Dysplazja nasadowa - małogłowie - zez

Kod ORPHA
1824

Kod OMIM
226960

Kod ICD10
Q87.5

Kod ICD11
LD24.6Y

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*Źródło

orphanet