

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
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Kod ORPHA 1824	Kod OMIM 226960	Kod ICD10 Q87.5
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Kod ICD11
LD24.6Y

*Źródło

orphanet