

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 - małowłowie - zez

Kod ORPHA

1824

Kod OMIM

226960

Kod ICD10

Q87.5

Kod ICD11

LD24.6Y

*Źródło

orphanet