

# Zespół Lowry'ego i Wooda

Kod Orpha: 1824 Kod OMIM: 226960

## 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

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.