

## Opis choroby \*

### Definicja

Endosteal sclerosis-cerebellar hypoplasia syndrome is characterized by congenital cerebellar hypoplasia, endosteal sclerosis, hypotonia, ataxia, mild to moderate developmental delay, short stature, hip dislocation, and tooth eruption disturbances. It has been described in four patients. Less common manifestations are microcephaly, strabismus, nystagmus, optic atrophy, and dysarthria. It is appears to be transmitted as an autosomal recessive trait.

### Dane

### Klasyfikacja

Zespół wad wrodzonych

#### Kod ORPHA

85186

#### Kod OMIM

614381

#### Kod ICD10

Q87.8

#### Kod ICD11

LD2F.1Y

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### \*Źródło

orphanet