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