

Opis choroby *

Definicja

A rare genetic disease characterized by mild intellectual deficit, congenital cataract, progressive sensorineural hearing impairment, ataxia, peripheral neuropathy, and short stature. There have been no further descriptions in the literature since 1991.

Dane

Klasyfikacja

Choroba

Synonimy

Cataract-ataxia-hearing loss syndrome

Kod ORPHA

1368

Kod OMIM

212710

Kod ICD10

G11.2

Kod ICD11

LD2H.Y

*Źródło

orphanet