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

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*Źródło

orphanet