

Opis choroby *

Definicja

Spastic paraplegia-glaucoma-intellectual disability syndrome is characterized by progressive spastic paraplegia, glaucoma and intellectual deficit. It has been described in two families. The second described sibship was born to consanguineous parents. The mode of inheritance is autosomal recessive.

Dane

Klasyfikacja

Choroba

Kod ORPHA

2818

Kod OMIM

270850

Kod ICD10

G11.4

Kod ICD11

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*Źródło

orphanet