

Opis choroby *

Definicja

Spastic ataxia with congenital miosis is a rare hereditary ataxia characterized by an apparently non-progressive or slowly progressive symmetrical ataxia of gait, pyramidal signs in the limbs, spasticity and hyperreflexia (especially in the lower limbs) together with dysarthria and impaired pupillary reaction to light, presenting as a fixed miosis (with pupils that seldom exceed 2 mm in diameter and dilate poorly with mydriatics). Nystagmus may also be present.

Dane

Klasyfikacja

Choroba

Synonimy

Autosomal dominant spastic ataxia type 7

SPAX7

Autosomalna dominująca ataksja spastyczna

typu 7

SPAX7

Kod ORPHA

1182

Kod OMIM

108650

Kod ICD10

G11.4

Kod ICD11

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*Źródło

orphanet