Opis choroby *

Definicja

A rare hereditary ataxia characterized by progressive truncal and limb ataxia resulting in gait instability. Dysarthria, dysphagia, nystagmus, spasticity of the lower limbs, mild peripheral sensory neuropathy, cognitive impairment and accelerated ageing have also been associated.

Dane

Klasyfikacja
Choroba

Synonimy
SCAR16
Autosomalna recesywna ataksja rdzeniowo-móżdżkowa typu 16
SCAR16
Spinocerebellar ataxia autosomal recessive type 16

Kod ORPHA
412057

Kod OMIM
615768

Kod ICD10
G11.1

Kod ICD11
-

---

*Źródło

orphanet