

Opis choroby *

Definicja

X-linked spinocerebellar ataxia type 3 is a form of spinocerebellar degeneration characterized by onset in infancy of hypotonia, ataxia, sensorineural deafness, developmental delay, esotropia, and optic atrophy, and by a progressive course leading to death in childhood. It has been described one family with at least six affected males from five different sibships (connected through carrier females). It is transmitted as an X-linked recessive trait.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

SCAX3

SCAX3

Zespół ataksja sprzężona z chromosomem X -
głuchota

X-linked ataxia-deafness syndrome

X-linked ataxia-hearing loss syndrome

Kod ORPHA

85297

Kod OMIM

301790

Kod ICD10

G11.1

Kod ICD11

8A03.1Y

*Źródło

orphanet