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

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*Źródło

orphanet