

Opis choroby *

Definicja

X-linked epilepsy-learning disabilities-behavior disorders syndrome is characterized by epilepsy, learning difficulties, macrocephaly, and aggressive behaviour. It has been described in males from a four-generation kindred. It is transmitted as an X-linked recessive trait and is likely to be caused by mutations in the gene encoding synapsin I (Xp11.3-q12).

Dane

Klasyfikacja

Choroba

Kod ORPHA

85294

Kod OMIM

300491

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet