

Opis choroby *

Definicja

X-linked intellectual disability, Wilson type is characterised by severe intellectual deficit with mutism, epilepsy, growth retardation and recurrent infections. It has been described in three males from three generations of one family. The causative gene has been localised to the 11p region of the X chromosome.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

85290

Kod OMIM

309545

Kod ICD10

Q87.8

Kod ICD11

LD90

*Źródło

orphanet