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

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*Źródło

orphanet