

Opis choroby *

Definicja

X-linked intellectual disability-epilepsy-progressive joint contractures-dysmorphism syndrome is characterised by intellectual deficit, epilepsy, facial dysmorphism and progressive joint contractures. It has been described in two boys. Hypotonia and feeding problems at birth were also reported. The mode of transmission is X-linked.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA	Kod OMIM	Kod ICD10
85319	-	Q87.8

Kod ICD11

LD90

*Źródło

orphanet