

Opis choroby *

Definicja

X-linked intellectual disability, Stevenson type is characterised by intellectual deficit, hypotonia, absent deep tendon reflexes, tapered fingers and excessive fingerprint arches, genu valgum, a characteristic face and small teeth. It has been described in four males from two generations of one family. The causative gene appears to be located in the q13 region of the X chromosome.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

85325

Kod OMIM

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Kod ICD10

Q87.8

Kod ICD11

LD90

*Źródło

orphanet