

Opis choroby *

Definicja

X-linked intellectual disability, Stocco Dos Santos type is characterised by severe intellectual deficit with hyperactivity, language delay, congenital hip luxation, short stature, kyphosis and recurrent respiratory infections. Aggressive behaviour and frequent epileptic seizures may also be present. The syndrome has been described in four boys from the same family. Transmission is X-linked and is caused by mutations in the *KIAA1202* gene, localised to the Xp11.2 region.

Dane

Klasyfikacja

Zespół wad wrodzonych

Kod ORPHA

85288

Kod OMIM

300434

Kod ICD10

Q87.8

Kod ICD11

LD90

*Źródło

orphanet