

Opis choroby *

Definicja

Partington syndrome is a form of syndromic X-linked mental retardation (S-XLMR) characterised by the association of mild to moderate intellectual deficit, dysarthria and dystonic hand movements. So far, less than 20 cases have been described in the literature. The syndrome is caused by mutations in the Aristaless-related homeobox (<i>ARX</i>) gene (Xp22.13). Transmission is X-linked recessive.

Dane

Klasyfikacja

Zespół wad wrodzonych

Synonimy

Partington-Mulley syndrome

Niepełnosprawność sprzężona z chromosomem

X - dystonia - dyzartria

Zespół Partingtona i Mulley'a

X-linked intellectual disability-dystonia-dysarthria syndrome

Kod ORPHA

94083

Kod OMIM

309510

Kod ICD10

G93.4

Kod ICD11

LD90.Y

*Źródło

orphanet