

Opis choroby *

Definicja

A congenital, X-linked, clinical subtype of L1 syndrome, characterized by variable spastic paraplegia, mild to moderate intellectual disability, and dysplasia, hypoplasia or aplasia of the corpus callosum. In this subtype hydrocephalus, adducted thumbs, or absent speech are not observed.

Dane

Klasyfikacja

Podtyp kliniczny

Kod ORPHA

1497

Kod OMIM

304100

Kod ICD10

Q04.8

Kod ICD11

LD20.Y

*Źródło

orphanet