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

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*Źródło

orphanet