

Opis choroby *

Definicja

A rare non-syndromic cerebral malformation characterized by congenital partial or complete absence of the corpus callosum. Patients are often asymptomatic but may also present with intellectual disability, visual impairment, delayed speech development, seizures, feeding difficulties, impaired hand-eye coordination, and behavioral abnormalities. Patients may have a normal intelligence quotient while exhibiting specific cognitive deficits, such as reduced interhemispheric transfer of sensorimotor information, reduced cognitive processing speed, and deficits in complex reasoning and novel problem-solving.

Dane

Klasyfikacja

Wada morfologiczna

Kod ORPHA

200

Kod OMIM

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Kod ICD10

Q04.0

Kod ICD11

LA05.3

*Źródło

orphanet