Opis choroby *

Definicja

Lissencephaly (LIS) due to TUBA1A mutation is a congenital cortical development anomaly due to abnormal neuronal migration involving neocortical and hippocampal lamination, corpus callosum, cerebellum and brainstem. A large clinical spectrum can be observed, from children with severe epilepsy and intellectual and motor deficit to cases with severe cerebral dysgenesis in the antenatal period leading to pregnancy termination due to the severity of the prognosis.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
171680

Kod OMIM
611603

Kod ICD10
Q04.3

Kod ICD11
-

---

*Źródło

orphanet