Opis choroby *

Definicja

A rare genetic neurological disorder characterized by the association of congenital spastic paraplegia with global developmental delay and intellectual disability, ophthalmologic abnormalities (including nystagmus, reduced visual acuity, or hypermetropia), and obesity. Additional manifestations are brachyplagiocephaly and dysmorphic facial features. Brain imaging may show dilated ventricles, abnormal myelination, and mild generalized atrophy. Homozygous loss-of-function variants of KIDINS220 associated with a fetal lethal phenotype with ventriculomegaly and limb contractures have been reported.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
SINO syndrome
Zespół SINO

Kod ORPHA
521390

Kod OMIM
617296

Kod ICD10
G11.4

Kod ICD11
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*Źródło

orphanet