

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	Synonimy
Zespół wad wrodzonych SINO syndrome	SINO syndrome
	Zespół SINO

Kod ORPHA	Kod OMIM	Kod ICD10
521390	617296	G11.4

Kod ICD11
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*Źródło

orphanet