

Opis choroby *

Definicja

Microlissencephaly-micromelia syndrome is a syndrome of abnormal cortical development, characterized by severe prenatal polyhydramnios, postnatal microcephaly, lissencephaly, upper limb micromelia, dysmorphic facies (coarse face, hypertrichosis, and short nose with long philtrum), intractable seizures, and early death. Hypoparathyroidism was noted in one case.

Dane

Klasyfikacja	Synonimy
Zespół wad wrodzonych	Basel-Vanagaite-Sirota syndrome
	Zespół Basela, Vanagaite i Sirota

Kod ORPHA	Kod OMIM	Kod ICD10
50810	-	Q04.3

Kod ICD11

-

*Źródło

orphanet