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
Zespół wad wrodzonych

Synonimy
Basel-Vanagaite-Sirota syndrome
Zespół Basela, Vanagaite i Sirota

Kod ORPHA
50810

Kod OMIM
-

Kod ICD10
Q04.3

Kod ICD11
-

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*Źródło

orphanet