Opis choroby *

Definicja

A rare syndromic type of cerebral malformation characterized by aprosencephaly (absence of telencephalon and diencephalon), oculo-facial anomalies (i.e. ocular hypotelorism or cyclopia, malformation/absence of nasal structures, cleft lip), preaxial limb defects (i.e. hypoplastic hands, absent halluces) and various other anomalies including ambiguous genitalia, imperforate anus, and vertebral anomalies. The syndrome is thought to have an autosomal recessive mode of inheritance.

Dane

Klasyfikacja
Zespół wad wrodzonych

Synonimy
Garcia-Lurie syndrome
Atelencephaly
Zespół Garcia i Lurie
XK syndrome
XK-aprosencephaly

Kod ORPHA
3469

Kod OMIM
207770

Kod ICD10
Q04.3

Kod ICD11
LD20.3

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*Źródło

orphanet