XK aprosencephaly syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. XK aprosencephaly syndrome. ORPHA:3469

XK aprosencephaly syndrome is a very 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.