

## 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

<b>Klasyfikacja</b>	<b>Synonimy</b>
Zespół wad wrodzonych	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