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X-linked lissencephaly with abnormal genitalia

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked lissencephaly with abnormal genitalia. ORPHA:452*

X-linked lissencephaly with abnormal genitalia (XLAG) is a rare, genetic, central nervous system malformation disorder characterized, in males, by lissencephaly (with posterior predominance and moderately thickened cortex), complete absence of corpus callosum, neonatal-onset (mainly perinatal) intractable seizures, postnatal microcephaly, severe hypotonia, poor responsiveness and hypogonadism (micropenis, hypospadias, cryptorchidism, small scrotal sac). Defective temperature regulation and chronic diarrhea may be additionally observed.