Microphthalmia, Lenz type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Microphthalmia, Lenz type. ORPHA:568

Lenz microphthalmia syndrome is a very rare X-linked inherited form of syndromic microphthalmia (see this term) characterized by unilateral or bilateral microphthalmia (and/or clinical anophthalmia) with or without coloboma in addition to a range of extraocular manifestations such as microcephaly, malformed ears, dental abnormalities (i.e. irregular shape of incisors), skeletal anomalies (duplicated thumbs, syndactyly, clinodactyly, camptodactyly (see these terms)), urogenital anomalies (hypospadias, cryptorchidism, renal dysgenesis, hydroureter) and mild to severe intellectual disability. It is allelic to two disorders: oculofaciocardiodental syndrome and premature aging appearance-developmental delay-cardiac arrhythmia syndrome (see these terms).