DISEASE:
Microphthalmia, Lenz type

NAME:	Microphthalmia, Lenz type
DESCRIPTION:	A rare X-linked inherited form of syndromic microphthalmia 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), 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.
ORPHACODE:	568
SYNONYMS:	Lenz microphthalmia
XREF(S):	Orphanet OMIM OMIM ICD-10
ANALYTE(S):	BCOR NAA10
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