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# X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome

INSERM

## Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome. ORPHA:431140*

X-linked colobomatous microphthalmia-microcephaly-intellectual disability-short stature syndrome is a rare syndromic microphthalmia disorder characterized by microphthalmia with coloboma (which may involve the iris, ciliary body, choroid, retina and/or optic nerve), microcephaly, short stature and intellectual disability. Other eye abnormalities such as pendular nystagmus, esotropia and ptosis may also be present. Additional associated abnormalities include kyphoscoliosis, anteverted pinnae with minimal convolutions, diastema of the incisors and congenital pes varus.