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Colobomatous microphthalmia-obesityhypogenitalism-intellectual disability syndrome

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Colobomatous microphthalmia-obesity-hypogenitalism-intellectual disability syndrome.

ORPHA:363741

Colobomatous microphthalmia-obesity-hypogenitalism-intellectual disability syndrome is a rare, genetic, syndromic microphthalmia disorder characterized by bilateral, usually asymmetrical, microphthalmia associated typically with a unilateral coloboma, truncal obesity, borderline to mild intellectual disability, hypogenitalism and, more variably, nystagmus, cataracts and developmental delay.

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