

Opis choroby *

Definicja

Colobomatous microphthalmia-obesity-hypogonadism-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, hypogonadism and, more variably, nystagmus, cataracts and developmental delay.

Dane

Klasyfikacja

Choroba

Kod ORPHA

363741

Kod OMIM

601794

Kod ICD10

Q87.8

Kod ICD11

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*Źródło

orphanet