Uveal coloboma-cleft lip and palate-intellectual disability

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Uveal coloboma-cleft lip and palate-intellectual disability. ORPHA:1473

Uveal coloboma-cleft lip and palate-intellectual disability is characterised by coloboma of the iris, bilateral cleft lip and palate, and intellectual deficiency of varying degree. A wide variability in clinical expression is observed. Some patients also present with microphthalmia, cataract, glaucoma, ptosis, sensorineural hearing loss and haematuria. To date, 12 cases have been described from three generations of a single family. Transmission is autosomal dominant.