Oculocutaneous albinism

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Oculocutaneous albinism. ORPHA:55

Oculocutaneous albinism (OCA) describes a group of inherited disorders of melanin biosynthesis characterized by a generalized reduction in pigmentation of hair, skin and eyes and variable ocular findings including nystagmus, reduced visual acuity and photophobia. Variants include OCA1A (the most severe form), OCA1B, OCA1-minimal pigment (OCA1-MP), OCA1-temperature sensitive (OCA1-TS), OCA2, OCA3, OCA4, OCA5, OCA6 and OCA7.