Ocular albinism with congenital sensorineural deafness

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Ocular albinism with congenital sensorineural deafness. ORPHA:352740

Ocular albinism with congenital sensorineural deafness is a rare, genetic, oculocutaneous disorder characterized by profound, congenital, sensorineural hearing loss in association with moderate to severe hypopigmentation of the ocular fundus, blue irides or partial heterochromia, and patchy or generalized hypopigmentation of the skin. White forelock, premature graying of hair, freckles, lentigines and café-au-lait macules are frequently associated. Other highly variable features include reduced visual acuity, strabismus, and an iris transillumination defect.