Oculocutaneous Albinism

National Cancer Institute

Source

National Cancer Institute. Oculocutaneous Albinism. NCI Thesaurus. Code C84941.

An autosomal recessive inherited disorder caused by mutations of the OCA2, SLC45A2, TYR and TYRP1 genes. It is characterized by hypopigmentation of the skin, hair, and eyes, resulting in very fair skin, white colored hair, and reduced pigmentation in the iris and retina. Individuals may have vision disturbances and photophobia.