Condition: Albinism (oculocutaneous albinism)

Inheritance:

Genetically heterogeneous, autosomal recessive or X-linked.

Genetic etiology:

The two most common forms are OCA1, due to mutation in the *OCA1* gene, which encodes the enzyme tyrosinase, and OCA2, due to mutation in the *OCA2* gene that encodes the P protein. Deficient tyrosinase results in failure of conversion of tyrosine to melanin. The P protein is a transmembrane protein of unknown function. The X-linked form causes ocular albinism (XLOA), due to mutation in the *GPR143* gene, which encodes a G-protein coupled membrane receptor that may be involved in melanogenesis in the iris and retina.

Frequency:

OCA1 affects 1/40,000; OCA2 affects 1/40,000, though the frequency is higher in Africans; XLOA affects 1/60,000 males.

Clinical features:

Oculocutaneous albinism is characterized by deficiency or lack of melanin pigment in skin, hair, and eyes. Lack of skin pigmentation leads to high risk of sunburn and development of skin cancer. Different alleles are associated with different degrees of skin and hair pigmentation. Lack of ocular pigmentation results in abnormal ocular development , with poor visual acuity, nystagmus, lack of binocular vision, and sensitivity to bright light. Ocular albinism affects only the eye.

Management:

Use of sunscreen to protect the skin; corrective lenses to augment vision; dark lenses to protect eyes from bright light.

Genetic counseling:

Depends on mode of genetic transmission (autosomal recessive or sex-linked). Genetic testing is available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.