Albinism

PLAY PICMONIC

Pathophysiology

Normal Melanocyte Number
Normal Melon (#) Number
Albinism is characterized by a normal melanocyte number but with a decrease in melanin production, also known as defective melanocytes.

Decreased or Absent Melanin Production
Down-arrow Melon-color Production
When melanin production decreases, the body may not be able to produce enough pigment, leading to a pale complexion.

Decreased or Absent Tyrosinase Activity
Down-arrow Tire
Melanin synthesis involves the conversion of tyrosine to DOPA, which is then converted to melanin by the enzyme tyrosinase. Reduction or absence of tyrosinase activity impairs melanin synthesis.

Defective Tyrosine Transport
Broken Tire
In albinism, melanin synthesis can also be affected by defective tyrosine transport.

Etiology

Genetic Mutation
Genetic Mutant
Albinism occurs due to a genetic mutation, which is classified into several types. OCA1 to OCA7 are inherited autosomal recessive types, while OA1, or ocular albinism, is inherited through an X-linked pattern.

Signs and Symptoms

Hypopigmentation
Hippo-pig with Hypopigmentation
The decrease in melanin production in albinism causes a pale skin manifestation, known as hypopigmentation. Patients present with white or fair hair, as well as greenish-blue or light brown eyes that can appear red in certain light due to reflection of blood vessels in the retina.

Ocular Disturbances
Wavy-eyes
Ocular disturbances can occur in patients with albinism. Patients can experience photophobia, refractive errors, strabismus, nystagmus, amblyopia, or abnormalities of the optic nerve.

Sunburn

Sun

Patients with albinism are prone to experiencing severe damage to their skin due to a lack of melanin. The majority of this damage happens in the areas of the body most exposed to the sun, including the face, shoulders, ears, and neck. Skin damage manifestations may include sunburns, blisters, ephelides, solar elastosis/keratosis, lentigo, and skin cancer.

Treatment

Avoid UV Light

Avoid-sign UV Sun-light

The risk of skin cancer increases in patients with higher ultraviolet exposure. Patients should especially avoid critical hours of high incidence of radiation between 10 am and 3 pm. Applying sunscreen (SPF +30) every 2 hours under outdoor activities should be suggested. Patients should also avoid medications that can increase photosensitivity.

Nitisinone

Night-sin

Nitisinone is FDA-approved to treat hereditary tyrosinemia type 1 by increasing plasma tyrosine levels and enhancing pigmentation in hair and eyes. It may also have potential as a treatment for ocular albinism.

Consideration

Increased Risk of Skin Cancer

Up-arrow Risk Skin Tumor-guy

Patients with albinism are at a higher risk of developing skin cancer due to their increased susceptibility to the harmful effects of ultraviolet radiation and actinic damage. The most common is squamous cell carcinomas.