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Review on Albinism

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Abstract: *Albinism affects the eyes and the visual system. As a result, individuals with albinism may experience photophobia, nystagmus, and poor visual acuity. They frequently have refractive errors, poor binocular, or stereoscopic vision and they may have strabismus. These functional changes are the result of the anatomical alterations associated with albinism. A reduction in melanin synthesis results in hypopigmentation of the following components of the eye: the iris (the diaphragm that controls the access of light to the retina), the retinal pigment epithelium (that has an integral supportive role to the neurosensory retina), and the choroid. Furthermore, in albinism, there is poor development of the fovea (the region of the retina responsible for central vision and best acuity) and anatomical changes in the visual pathway that connects the eye to the brain. With the appropriate management, individuals with albinism may maximize their visual potential. General measures, such as reducing photophobia and glare, refractive correction, and visual aids, may improve visual acuity. Strabismus and nystagmus may respond to surgical correction.*

Keywords: Albinism, Oculocutaneous, Kertosis, Actinic, Skin neoplasms, Social stigma.

I. INTRODUCTION

Albinism is a genetic condition of hypopigmentation caused by an abnormality in melanin pigment production. The absence or reduction of melanin has a severe impact on the development of the eye and visual system, such that persons with albinism display a variety of ophthalmic deficits, including foveal hypoplasia, translucency of the iris, nystagmus, reduced visual acuity and an abnormal decussation pattern at the optic chiasm.[1,2]

Different Type of Albinism

Albinism has two main types: ocular albinism (OA), which primarily affects the eyes, and oculocutaneous albinism (OCA), which affects the skin, hair, and eyes.

Doctors further subdivide OCA into a number of subgroups depending on the specific genes it affects.

These subdivisions includeTrusted Source:

OCA type 1: Individuals tend to have milky skin, white hair, and blue eyes. With age, some individuals' skin and hair may darken.

OCA type 2: Less severe than type 1, this occurs most often in sub-Saharan Africans, African Americans, and some Native American communities.

OCA type 3: Vision problems are usually milder in type 3 than in other types. This type mostly affects Black South Africans.

OCA type 4: This type is most common among East Asian populations. It presents similarly to type 2.

X-linked ocular albinism: A genetic mutation in the X chromosome causes X-linked ocular albinism, which mainly affects males. Vision problems are present, but eye, hair, and skin colour are generally within the normal range.

Hermansky-Pudlak syndrome: This rare variant is most common Trusted Source in Puerto Rico. The symptoms are similar to those of oculocutaneous albinism, but bowel, heart, kidney, and lung diseases or bleeding disorders, such as hemophilia, are more likely.[4]

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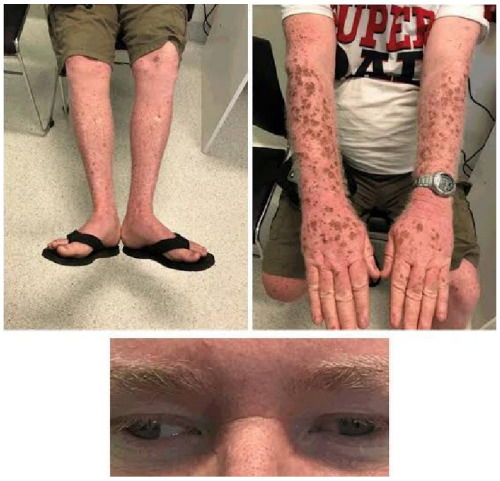

Fig No.2 Hermansky-Pudlak syndrome

Chediak-Higashi syndrome: This is a very rare form of albinism resulting from a mutation in the CHS1 gene. The symptoms may resemble those of oculocutaneous albinism, but a person's hair can appear silvery, and their skin can look slightly gray. The white blood cells can have defects, making the person more prone to infections.[3,5]

Fig No.3 Chediak-Higashi syndrome

Symptoms

The main symptoms of albinism affect the vision and the color of the skin, hair, and eyes.

Skin: The most obvious sign of albinism is a lighter skin tone, although this is not always the case. In some people, levels of melanin **slowly increaseTrusted Source** over time, darkening the skin tone as the person ages.

An individual's skin may burn easily in the sun, and it does not usually tan. After sun exposure, some people with albinism might develop:

- freckles
- moles, which are usually pink in color due to the reduced quantities of pigment
- lentigines, which are large freckle-like spots[6]

Hair: In people with albinism, hair color can range from white to brown. Those of African or Asian descent tend to have yellow, brown, or reddish hair.As the individual ages, their hair color may slowly darken.[7]

Eye colour: Eye colour may also change with age and can vary from very light blue to brown.

Low levels of melanin in the iris mean that the eyes can appear slightly translucent and, in a certain light, look red or pink as the light reflects off the retina at the back of the eye.

The lack of pigment prevents the iris from fully blocking sunlight, so the person is sensitive to light. Doctors call this photosensitivity.

Vision: All types of albinism affect the vision to a certain degree. Possible changes to eye function includeTrusted Source:

- **Nystagmus:** The eyes move rapidly and uncontrollably back and forth.

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- **Strabismus:** The eyes do not align.
- **Amblyopia:** This is the medical name for a lazy eye.
- **Myopia or hypermetropia:** The person may have extreme nearsightedness or farsightedness.
- **Photophobia:** The eyes are particularly sensitive to light.
- **Optic nerve hypoplasia:** Visual impairment happens because an individual's optic nerve is underdeveloped.
- **Optic nerve misrouting:** Nerve signals from the retina to the brain follow unusual nerve routes.

Astigmatism**:** An abnormal inflexibility of the front surface of the eye or lens results in blurred vision.[8]

Causes

Albinism is caused by a genetic mutation that is usually passed from parents to child.
There are two main cause of albinism

Autosomal recessive inheritance

Autosomal recessive inheritance disorder is a condition of passing the genetic trait to their children. It can be inherited when the child carries the defective or mutated copy of the gene.
According to data published in NCBI, titled "Albinism", states that oculocutaneous albinism and ocular albinism are passed to their children through autosomal recessive inheritance.
Additionally, it states that the children should carry one copy of the mutated gene from each parent get this condition.[9]

X-linked inheritance

Ocular albinism is usually passed through X-linked inheritance. When albinism is passed through X-linked inheritance, it affects girls and boys in a different pattern.
Girl children will become carriers of the mutated gene, whereas boys will be affected with albinism. For example, when the maternal is affected with albinism, there is a chance of her offspring getting affected with albinism in one in two chances.
When the paternal is affected with albinism, his daughters will be carriers while his sons will neither have albinism nor be carriers.
If your family has albinism, it is recommended to talk to a genetic counsellor before getting pregnant. This will help to avoid the carrying the mutated genes to their offspring and prevent the defected genes from running through families.[10]

Diagnose Albinism

Albinism can be diagnosed merely by observation of major or total absence of pigmentation of the skin, hair and eyes.
Other tests to diagnose albinism include -
Genetic Testing -
Genetic testing is the most accurate way to diagnose albinism and its specific type. This is helpful in families with albinism and is useful for specific, isolated populations who carry the trait in them.
Eye Examination -
An ophthalmologist should perform a **complete examination of the eye** of an affected individual. An **electroretinogram test** should be done to determine vision problems in albinism.
Chemical Testing of Hair -
Chemical testing of hair also provides an easy confirmation of the diagnosis of albinism.
Hairbulb pigmentation test- It can be used to identify carriers. It is done by incubating a piece of the person's hair in a solution of tyrosine, an amino acid the body uses to make melanin. If the hair turns dark, it means the hair is making melanin. Light hair means there is no melanin synthesis.
Tyrosinase test- It is more precise than the hairbulb pigmentation test. It measures the rate at which hair converts tyrosine into another chemical (DOPA), which is then made into pigment.
Blood Test -
Recently, a blood test has been developed that can identify carriers of the gene for some types of albinism.

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Prenatal Diagnosis Of Albinism-

Amniocentesis and **chorionic villus sampling** can also diagnose some types of albinism in pregnancy.[11]

PATHOPHYSIOLOGY

Mortality/Morbidity

The main cause for morbidity in patients with albinism is decreased visual acuity.

In tropical regions, there can be higher mortality among these patients secondary to an increased incidence of skin cancer due to sun exposure.

Mortality also is increased in patients with Hermansky-Pudlak syndrome and Chediak-Higashi syndrome. Patients with Hermansky-Pudlak syndrome have a bleeding diathesis secondary to platelet dysfunction and also experience restrictive lung disease, inflammatory bowel disease, cardiomyopathy, and renal disease. Patients with Chediak-Higashi syndrome are susceptible to infection and also can develop lymphofollicular malignancy.[12]

Race

Albinism affects all racial groups. However, type II oculocutaneous albinism occurs more frequently in African American and African populations. Similarly, there is a much higher incidence of Hermansky-Pudlak syndrome among people of Puerto Rican origin.

Sex

Oculocutaneous albinism affects both sexes equally. Ocular albinism is a disease primarily of males because of its sex-linked transmission.

Age

Most people with albinism are diagnosed during infancy or early childhood.

Prognosis

Visual prognosis in patients with albinism is quite variable. Usually, no improvement in visual acuity occurs in patients with type I oculocutaneous albinism. Visual acuity may improve with increased pigmentation in other forms of albinism as the patient grows older.[13]

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