

ORIGIN AND PATHOGENESIS OF THE CURRENTLY COMMON ALBINISM DISEASE

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Annotation: The term albinism usually refers to oculocutaneous (ok-u-low-ku-TAY-nee-us) albinism (OCA). OCA is a group of disorders passed down in families where the body makes little or none of a substance called melanin. The type and amount of melanin in your body determines the color of your skin, hair and eyes. Melanin also plays a role in the development and function of the eyes, so people with albinism have vision problems.

Key words: Albinism, skin, eyes, melanin.

Symptoms of albinism are usually seen in a person's skin, hair and eye color, but sometimes differences are slight. People with albinism are also sensitive to the effects of the sun, so they're at higher risk of getting skin cancer.

Although there's no cure for albinism, people with the disorder can take steps to protect their skin and eyes and get proper eye and skin care.

Symptoms of albinism involve skin, hair and eye color, as well as vision.

The easiest form of albinism to see results in white hair and very light-colored skin compared with siblings or other blood relatives. But skin coloring, also called pigmentation, and hair color can range from white to brown. People of African descent who have albinism may have skin that is light brown or red brown and have freckles. For some people, skin color may be nearly the same as that of parents or siblings without albinism.

With exposure to the sun, some people may develop:

- Freckles.
- Moles, with or without color, which are sometimes pink.
- Large freckle-like spots, called solar lentigines (len-TIJ-ih-nee-z).
- Sunburn and no ability to tan.

For some people with albinism, skin coloring never changes. For others, melanin production may begin or increase during childhood and the teen years, resulting in slight changes in color.

Hair color can range from very white to brown. People of African or Asian descent who have albinism may have hair color that's yellow, red or brown. Hair color also may darken by early

adulthood. Or hair may stain from contact with minerals in water and the environment, making hair appear darker with age.

Eyelashes and eyebrows are often pale. Eye color can range from very light blue to brown and may change with age.

With albinism, the colored parts of the eyes, called the irises, usually don't have enough pigment. This allows light to shine through the irises and makes the eyes extremely sensitive to bright light. Because of this, very light-colored eyes may appear red in some lighting.

Vision problems are a key feature of all types of albinism. Eye problems may include:

- Rapid, back-and-forth movement of the eyes that can't be controlled, called nystagmus.
- An uncommon head position or head posture, such as tilting the head to try to reduce eye movements and see better.
- Eyes that can't look at the same direction at the same time or they appear to be crossed, a condition called strabismus.
- Problems seeing near objects or far objects, called farsightedness or nearsightedness.
- Extreme sensitivity to light, called photophobia.
- A difference in the curve of the front surface of the eye or the lens inside the eye, called astigmatism, which causes blurred vision.
- Differences in the development of the thin layer of tissue on the inside back wall of the eye, called the retina. This difference results in reduced vision.
- Nerve signals from the retina to the brain that don't follow the usual nerve pathways in the eye. This is called misrouting of the optic nerve.
- Poor depth perception, which means not being able to see things in three dimensions and judge how far away an object is.
- Legal blindness — vision less than 20/200 — or complete blindness.

Albinism is a group of genetic conditions in which a person has no or very little melanin pigment in their skin, hair and eyes. Albinism occurs in all racial and ethnic groups throughout the world. In Australia, about 1 in 17,000 people have some type of albinism.

It is important for people with albinism to protect their skin and eyes from the sun, and have their eyes checked regularly.

The genes for albinism are passed down from both parents, who may not be directly affected by it themselves. However, there is no simple test to determine whether a person carries a gene for albinism. Some children with albinism are born with pinkish-white skin and white hair. Their eyes are usually light grey, blue or hazel, although they can look pink in the light.

People with albinism usually have poor vision. Glasses can help, but some have poor vision even with glasses. Several eye conditions can affect people with albinism including nystagmus, which is horizontal back and forth movement of the eyes, and photophobia, which is sensitivity to bright light and glare.

Albinism is a group of inherited disorders that results in little or no production of the pigment melanin, which determines the color of the skin, hair and eyes. Melanin also plays a role in the development of certain optical nerves, so all forms of Albinism cause problems with the development and function of the eyes. Other symptoms can include light skin or changes in skin color; very white to brown hair; very light blue to brown eye color that may appear red in some light and may change with age; sensitivity to sun exposure; and increased risk of developing skin cancer. Albinism is caused by genetic changes in one of several genes, and most types are inherited in an autosomal recessive manner.

Patient organizations can help patients and families connect. They build public awareness of the disease and are a driving force behind research to improve patients' lives. They may offer online and in-person resources to help people live well with their disease. Many collaborate with medical experts and researchers.

Services of patient organizations differ, but may include:

- Ways to connect to others and share personal stories
- Easy-to-read information
- Up-to-date treatment and research information
- Patient registries
- Lists of specialists or specialty centers
- Financial aid and travel resources

Please note: GARD provides organizations for informational purposes only and not as an endorsement of their services. Please contact an organization directly if you have questions about the information or resources it provides.

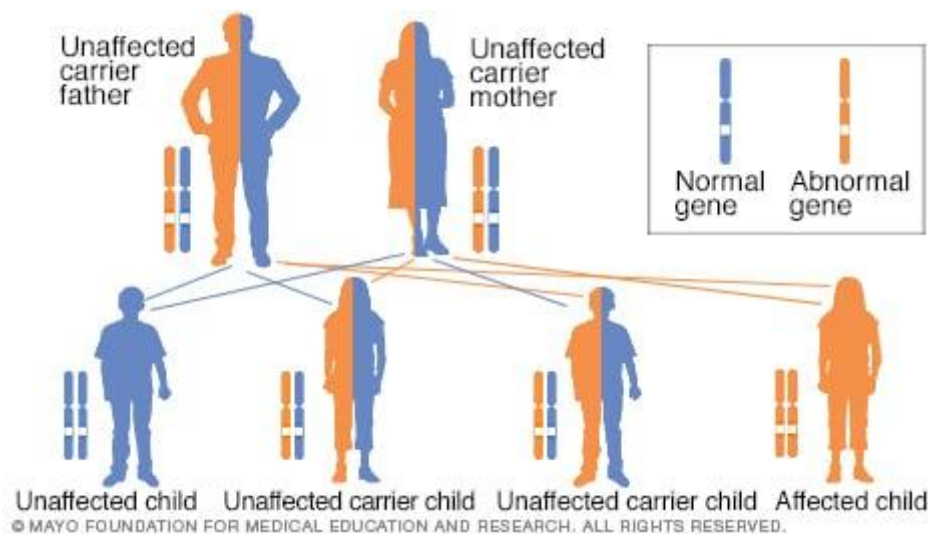
Albinism is a rare condition. It is a genetic disorder which is characterized by the inadequate production of the pigment melanin. The disorder may be inherited from the parents. The condition is not life threatening, but those suffering from albinism may have to limit their outdoor activities as they cannot tolerate being in the sun for long.

Melanin is the chemical that gives the skin, hair and the iris of the eye their color. Those suffering from albinism will have extremely pale skin, hair and eyes. They may also have some patches of skin or hair that have a darker shade than other parts. Some serious complications that may crop up include problems with the vision, and a higher risk of developing skin cancer.

There are primarily two basic types of albinism. These are oculocutaneous albinism and ocular albinism. Oculocutaneous albinism or OCA affects the pigment in the eyes, hair and skin. Those suffering from this condition will have white or pink hair, extremely pale skin and pale iris color.

They usually suffer from eyesight problems because the photoreceptor cells are unable to process the light waves and send conflicting messages to the brain.

Ocular albinism or OA, is not as obvious as the skin and hair do not have such extreme lack of pigmentation. However, the primary problem here lies with the eye sight. An examination of the eye will show lack of pigmentation in the iris and retina . This is the rarer form of albinism and is caused by a mutation in an X chromosome. Its inheritance therefore follows an X-linked pattern. It is not as obvious at birth as OCA.



Several genes give instructions for making one of several proteins involved in producing melanin. Melanin is made by cells called melanocytes that are found in your skin, hair and eyes.

Albinism is caused by a change in one of these genes. Different types of albinism can occur, based mainly on which gene change caused the disorder. The gene change may result in no melanin at all or a big decrease in the amount of melanin.

Symptoms of Albinism

The most obvious symptoms are the pale color of the hair and the skin. The lack of melanin reduces the skin's ability to protect itself from UV rays. This results in increased vulnerability to skin damage by solar exposure. However, a number of symptoms of albinism can be related to associated eye problems. The list below includes some possible symptoms related to vision in a person with albinism.

Strabismus: There is a misalignment of the eyes leading to a squint. Also known as being cross-eyed, it happens when one eye looks straight at an object while the other turns inwards, outwards, upwards or downwards. Both eyes do not move in a synchronized manner. This hampers optimal eye functioning.

Nystagmus: A condition where the patient's eyes move rapidly and uncontrollably. Often a preferred head position is developed to optimize vision and deal with the involuntary eye motion. This leads to strained muscles in the neck.

Photophobia: A high degree of sensitivity to bright light from sources such as the sun, fluorescent bulbs or incandescent lighting. Because of the intense discomfort, the patient feels the need to squint or close the eyes for relief.

Refractive Errors: Hyperopia or far sightedness, myopia or near sightedness and astigmatism or defect in the curvature of the cornea are the common types of refractive errors.

Monocular Vision: A dependence on only one eye for vision, with the other eye not being used to send messages to the brain. This may thus become a lazy eye.

Foveal Hypoplasia: Improper development of the retina during birth or childhood leading to poor vision.

Faulty Optic Nerve: The nerve signals that must travel from the retina to the brain fail to develop properly and therefore do not transmit information as required.

Transillumination Issues with Iris: The colored diaphragm between the anterior chamber of the eye and the lens is known as the iris. When it is lacking in the pigment melanin, it is unable to screen out the extra light entering the eye. This causes vision issues.

Treatment Options for Albinism

The production of melanin cannot be regulated artificially. For this reason, there is no cure for the subnormal synthesis of melanin which causes albinism. The skin may be protected from sun exposure to reduce the chances of sunburn as well as of skin cancer. Using sun screen of SPF 30 or more, good-quality sunglasses, long-sleeved garments and wide-brimmed hats are a good start to protect the patient.

There is no cure for the eye problems caused by albinism. However, treatment options such as corrective glasses and contact lenses may help in some cases to improve the vision. Using correct lighting can help reduce the strain on the eye sight within the home. Children in school will need additional help to see distant objects in the classroom. Training and counseling may be required to equip teachers who handle children with albinism.

Albinism does not worsen with age. It has no impact on the intelligence of a person. Genetic counseling of patients suffering from albinism helps them understand the disorder. It also helps deal with the possibility of future generations being affected with the condition. Having a support group makes it easier to deal with the challenges of albinism. Keeping a positive mental attitude and developing social skills to cope with the stigma attached to the condition help the patient immensely.

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