

What Is Albinism?

Albinism is an inherited genetic condition that reduces the amount of melanin pigment formed in the **skin, hair and/or eyes**. Albinism occurs in all racial and ethnic groups throughout the world. In the U.S., approximately one in 18,000 to 20,000 people has some type of albinism. In other parts of the world, the occurrence can be as high as one in 3,000. Most children with albinism are born to parents whose hair and eye color are typical for their ethnic backgrounds.

A common myth is that people with albinism have red eyes. Although lighting conditions can allow the blood vessels at the back of the eye to be seen, which can cause the eyes to look reddish or violet, most people with albinism have blue eyes, and some have hazel or brown eyes. There are different types of albinism and the amount of pigment in the eyes varies. However, vision problems are associated with all types of albinism.

Vision Considerations

People with albinism have vision problems that are not correctable with eyeglasses, and many have low vision. It's the abnormal development of the retina and abnormal patterns of nerve connections between the eye and the brain that cause vision problems. The presence of these eye problems defines the diagnosis of albinism.

The degree of impairment varies with the different types of albinism. Although people with albinism may be considered "legally blind" with a corrected visual acuity of 20/200 or worse, most learn to use their



Photo courtesy of Positive Exposure, Rick Guidotti

vision in a variety of ways and are able to perform innumerable activities such as reading, riding a bike or fishing. Some have sufficient vision to drive a car.

Dermatological Considerations

Because most people with albinism have fair complexions, it's important to avoid sun damage to the skin and eyes by taking precautions such as wearing sunscreen or sunblock, hats, sunglasses and sun-protective clothing.

Types of Albinism

While most people with albinism have very light skin and hair, levels of pigmentation can vary depending on one's type of albinism. Oculocutaneous (pronounced ock-you-low-kew-TAIN-ee-us) albinism (OCA) involves the eyes, hair and skin. Ocular albinism (OA), which is much less



Photo courtesy of Positive Exposure, Rick Guidotti

common, involves only the eyes, while skin and hair may appear similar or slightly lighter than that of other family members.

Over the years, researchers have used various systems for classifying oculocutaneous albinism. In general, these systems contrasted types of albinism having almost no pigmentation with types having slight pigmentation. In less pigmented types of albinism, hair and skin are cream-colored and vision is often in the range of 20/200. In types with slight pigmentation, hair appears more yellow or has a reddish tinge and vision may be better.



Photo courtesy of Positive Exposure, Rick Guidotti

DNA tests can determine the precise type of albinism. Research on albinism genes is ongoing. **To date as many as seven forms of oculocutaneous albinism are now recognized – OCA1, OCA2, OCA3, OCA4, OCA5, OCA6 and OCA7.** Some are further divided into subtypes.

- **OCA1**, or tyrosinase-related albinism, results from a genetic defect in an enzyme called tyrosinase. This enzyme helps the body to change the amino acid, tyrosine, into pigment. (An amino acid is a “building block” of protein.) There are two subtypes of OCA1. In OCA1A, the enzyme is inactive and no melanin is produced, leading to white hair and very light skin. In OCA1B, the enzyme is minimally active and a small amount of melanin is produced, leading to hair that may darken to blond, yellow/orange or even light brown, as well as slightly more pigment in the skin.

- **OCA2**, or P gene albinism, results from a genetic defect in the P protein that helps the tyrosinase enzyme to function. People with OCA2 make a minimal amount of melanin pigment and can have hair color ranging from very light blond to brown.
- **OCA3** is rarely described and results from a genetic defect in TYRP1, a protein related to tyrosinase. People with OCA3 can have substantial pigment.
- **OCA4** results from a genetic defect in the SLC45A2 protein that helps the tyrosinase enzyme to function. People with OCA4 make a minimal amount of melanin pigment similar to people with OCA2.
- **OCA5–7** were recognized in humans in 2012 and 2013. They have reported mutations on three additional causative genes. As gene testing becomes available, and more people with these types of albinism are identified, the complete range of physical manifestations will be recognized, and may overlap with other known types of OCA. Currently, these types of albinism are considered to be uncommon.

Researchers have also identified several other genes that result in albinism with other features. One group includes at least 10 genes leading to **Hermansky-Pudlak Syndrome** (HPS). In addition to albinism, HPS is associated with bleeding problems and bruising. Some forms are also associated with lung and bowel disease. HPS is a less common form of albinism but should be suspected if a person with albinism shows unusual bruising or bleeding or if

a genetic test for a type of OCA produces inconclusive results.

Other albinism-related syndromes include **Chediak-Higashi Syndrome** and **Griscelli Syndrome**.



Photo courtesy of Positive Exposure, Rick Guidotti

Genetics of Albinism

The genes for OCA are located on “autosomal” chromosomes. Autosomes are the 22 pairs of chromosomes that contain genes for our general body characteristics, compared to the one pair of sex chromosomes. We normally have two copies of these chromosomes and the many genes on them – one inherited from our father, the other inherited from our mother. For a recessive trait (like most types of albinism) to occur, both of the person’s chromosomes must carry that trait. That means that most

types of albinism result from inheriting an albinism trait from both the mother and the father who often have typical pigmentation. In this case, the mother and father are considered to be carriers of the albinism trait because they each carry a recessive gene for the condition but do not manifest the condition themselves. When both parents carry the albinism gene (and neither parent has albinism) there is a one in four chance at each pregnancy that the baby will be born with albinism. This type of inheritance is called “autosomal recessive” inheritance.



Photo courtesy of Positive Exposure, Rick Guidotti

Ocular albinism (OA1) is caused by a change in the GPR143 gene that plays a signaling role that is especially important to pigmentation in the eye. OA1 follows a simpler pattern of inheritance because the gene for OA1 is on the X chromosome. Females have two copies of the X chromosome while males have only one copy (and a Y chromosome that makes them male). To have ocular albinism, a male only needs to inherit one changed copy of the gene for ocular albinism from his carrier mother. Therefore almost all of the people



Photo courtesy of Positive Exposure, Rick Guidotti

with OA1 are males. Parents should be suspicious if a female child is said to have ocular albinism. While possible if the mother is a carrier of ocular albinism and the father has ocular albinism, it is extremely rare.

For couples who have not had a child with albinism, there is no simple test to determine whether a person carries a gene for albinism. Researchers have analyzed the DNA of many people with albinism and found the changes that cause albinism, but these changes are not always in exactly the same place, even for a given type of albinism. Moreover, many of the tests do not find all possible changes. Therefore, the tests for the albinism gene may be inconclusive. If parents have had a child with albinism previously, and if that affected child has had a confirmed diagnosis by DNA analysis, there is a way to test in subsequent pregnancies to see if the fetus has albinism. The test uses either amniocentesis (placing a needle into the uterus to draw off fluid) or chorionic villous sampling (CVS). Cells in the fluid are examined to see if they have an albinism gene from each parent.

For specific information and genetic testing, seek the advice of a qualified geneticist or genetic counselor. The American College of Medical Genetics and the National Society of Genetic Counselors maintain a referral list. Those considering prenatal testing should be made aware that people with albinism usually adapt quite well to their disabilities and lead very fulfilling lives.

Vision Rehabilitation

Eye problems in albinism result from the abnormal development of the eye because of a lack of pigment and often include:

- **Nystagmus:** Regular horizontal back and forth movement of the eyes
- **Strabismus:** Muscle imbalance of the eyes, “crossed eyes” (esotropia), “lazy eye” or an eye that deviates out (exotropia)
- **Photophobia:** Sensitivity to bright light and glare
- **Refractive Error:** People with albinism may be either farsighted or nearsighted and usually have astigmatism
- **Foveal hypoplasia:** The retina, the surface inside the eye that receives light, does not develop normally before birth and in infancy
- **Optic nerve misrouting:** The nerve signals from the retina to the brain do not follow the usual nerve routes

The iris, the colored area in the center of the eye, has very little or no pigment to screen out stray light coming into the eye. Light normally enters the eye only through the pupil, the dark opening in

the center of the iris, but in albinism light can pass through the iris as well.

For the most part, treatment consists of visual rehabilitation. Surgery to correct strabismus may improve the appearance of the eyes. However, since surgery will not correct the misrouting of nerves from the eyes to the brain, surgery will not improve eyesight or fine binocular vision. In the case of esotropia or “crossed eyes,” surgery may help vision by expanding the visual field (the area that the eyes can see while looking at one point). Surgical intervention is also available to minimize nystagmus.

People with albinism are sensitive to glare, but they do not prefer to be in the dark, and they need light to see just like anyone else. Sunglasses or tinted contact



Photo courtesy of Positive Exposure, Rick Guidotti

lenses may help outdoors. Indoors, it is important to place lights for reading or close work over a shoulder rather than in front.

Various optical aids are helpful to people with albinism, and the choice of an optical aid depends on how a person uses his or her eyes in jobs, hobbies or other usual activities. Some people do well using bifocals which have a strong



Photo courtesy of Positive Exposure, Rick Guidotti

reading lens, prescription reading glasses or contact lenses. Others use handheld magnifiers or special small telescopes, and some prefer to use screen magnification products on computers.

Some people with albinism use bioptics, glasses which have small telescopes mounted on, in or behind their regular lenses so that one can look through either the regular lens or the telescope. Some states allow the use of bioptic telescopes for driving.

Optometrists or ophthalmologists who are experienced in working with low vision patients can recommend various

optical aids. Clinics should provide instruction in their use. The American Foundation for the Blind maintains a directory of low vision clinics. In Canada, support is available from the Canadian National Institute for the Blind.

Medical Problems

In the United States, most people with albinism live normal life spans and have the same types of general medical problems as the rest of the population. The lives of people with Hermansky-Pudlak Syndrome can be shortened by lung disease or other medical problems.

In tropical countries, people with albinism who do not have access to adequate skin protection may develop life-threatening skin cancers. If they use appropriate skin protection, such as



Photo courtesy of Positive Exposure, Rick Guidotti



Photo courtesy of Positive Exposure, Rick Guidotti

sunscreens rated 20 SPF or higher and opaque clothing, people with albinism can enjoy outdoor activities even in summer.

Social Considerations

People with albinism are at risk of isolation because the condition is often misunderstood. Social stigmatization can occur, especially within communities of color, where the race or paternity of a person with albinism may be questioned. Families and schools must make an effort to include children with albinism in group activities. Contact with others with albinism or who have albinism in their families or communities is most helpful. NOAH can provide the names of contacts in many regions of the country.

This publication may be reproduced as long as no changes are made, the copyright notice is kept intact and the author and photographer is given credit.



Photo courtesy of Positive Exposure, Rick Guidotti



Photos courtesy of Positive Exposure, Rick Guidotti

NOAH envisions a world where people with albinism are empowered to be fully functioning members of society, where barriers and the stigma of difference no longer exist, and where people with albinism have a quality of life that is rewarding, dignified and fulfilling. The organization sponsors national and regional conferences and a biennial family summer camp. In

addition, local chapters meet in many areas of the U.S. and Canada. NOAH can provide a list of chapters and contact persons.

Visit **www.albinism.org** for more information about albinism including bulletins, events, ways to connect with the albinism community and to subscribe to NOAH's large-print quarterly magazine, *Albinism InSight*.



**National Organization for
Albinism and Hypopigmentation**

PO Box 959, East Hampstead, NH 03826-0959 USA

Phone: 800-473-2310 • 603-887-2310

Fax: 603-887-6049

www.albinism.org • Email: info@albinism.org