MAYO CLINIC

Diseases and Conditions

Albinism

By Mayo Clinic Staff

Albinism includes a group of inherited disorders that are characterized by little or no production of the pigment melanin. The type and amount of melanin your body produces determines the color of your skin, hair and eyes. Most people with albinism are sensitive to sun exposure and are at increased risk of developing skin cancer.

Although there's no cure for albinism, people with the disorder can take steps to protect their skin and maximize their vision. Some people with albinism may feel socially isolated or experience discrimination.

Signs of albinism are usually, but not always, apparent in a person's skin, hair and eye color. However, all people with the disorder experience vision problems.

Skin

Although the most recognizable form of albinism results in white hair and pinkish skin, skin coloring (pigmentation) can range from white to brown, and may be nearly the same as that of parents or siblings without albinism.

For some people with albinism, skin pigmentation never changes. For others, melanin production may begin or increase during childhood and the teen years, resulting in slight changes in pigmentation. With exposure to the sun, some people may develop:

- Freckles
- Moles, with or without pigment — moles without pigment are generally pink-colored
- Large freckle-like spots (lentigines)
- The ability to tan

Hair

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, reddish or brown. Hair color may also darken by early adulthood.

Eye color

Eye color can range from very light blue to brown and may change with age.

The lack of pigment in the colored part of the eyes (irises) makes them somewhat translucent. This means that the irises can't completely block light from entering the eye. Because of this, very light-colored eyes may appear red in some lighting. This occurs because you're seeing light reflected off the back of the eye and passing back out through the iris again — similar to the red-eye that occurs in a flash photo.

Vision

Signs and symptoms of albinism related to eye function include:

- Rapid, involuntary back-and-forth movement of the eyes (nystagmus)
- Inability of both eyes to stay directed at the same point or to move in unison (strabismus)
- Extreme nearsightedness or farsightedness
- Sensitivity to light (photophobia)
- Abnormal curvature of the front surface of your eye or the lens inside your eye (astigmatism), which causes blurred vision

When to see a doctor

If your child lacks pigment in his or her hair or skin at birth that affects the eyelashes and eyebrows — as is often the case in infants with albinism — your doctor will likely order an eye exam and closely follow any changes in your child's pigmentation.

For some infants, the first sign of albinism is poor visual tracking. This may be followed at 3 to 4 months of age by rapid back-and-forth shifting (nystagmus) in the eyes. If you observe these signs in your baby, talk to your doctor.

Contact your doctor if your child with albinism experiences frequent nosebleeds, easy bruising or chronic infections. These signs and symptoms may indicate the presence of Hermansky-Pudlak or Chediak-Higashi syndromes, which are rare but serious genetic disorders.

Albinism is caused by a mutation in one of several genes. Each of these genes provides instructions for making one of several proteins involved in the production of melanin. Melanin is produced by cells called melanocytes, which are found in your skin and eyes. A mutation may result in no melanin at all or a significant decline in the amount of melanin.

In some types of albinism, a person must inherit two copies of a mutated gene — one from

each parent — in order to have albinism (recessive inheritance).

Impact on eye development

Regardless of which gene mutation is present, vision impairment is a key feature of all types of albinism. These impairments are caused by irregular development of the optic nerve pathways from the eye to the brain and from abnormal development of the retina.

Types of albinism

Types of albinism, based mainly on which mutated gene caused the disorder, include:

- **Oculocutaneous albinism.** Oculocutaneous (ok-u-low-ku-TAY-nee-us) albinism is caused by a mutation in one of four genes. People with oculocutaneous albinism (OCA) type 1 have milky white skin, white hair and blue eyes at birth. Some people with OCA type 1 never experience an increase in pigmentation, but others begin to produce melanin during early childhood. Their hair may become a golden blond, brown or red, and their irises may change color and lose some translucence.

  OCA type 2 is most common in sub-Saharan Africans, African-Americans and Native Americans. The hair may be yellow, auburn, ginger or red, the eyes can be blue-gray or tan, and the skin is white at birth. With sun exposure, the skin may, over time, develop freckles, moles or lentigines.

  People with OCA type 3, mainly found in black South Africans, usually have reddish-brown skin, ginger or reddish hair, and hazel or brown eyes. OCA type 4 looks similar to type 2 and is most often found in people of East Asian descent.

- **X-linked ocular albinism.** The cause of X-linked ocular albinism, which occurs almost exclusively in males, is a gene mutation on the X chromosome. People who have ocular albinism have vision problems, but their skin, hair and eye color are generally in the normal range or slightly lighter than that of others in the family.
- **Hermansky-Pudlak syndrome.** Hermansky-Pudlak syndrome is a rare albinism disorder caused by a mutation in one of at least eight different genes. The disorder is much more common in Puerto Rico. People with this disorder have signs and symptoms similar to people with oculocutaneous albinism, but they may also develop lung and bowel diseases, or a bleeding disorder.
- **Chediak-Higashi syndrome.** Chediak-Higashi syndrome is a rare form of albinism associated with a mutation in the LYST gene. With signs and symptoms similar to oculocutaneous albinism, the hair is usually brown or blond with a silvery sheen, and the skin is usually creamy white to grayish. People with this syndrome have a defect in white blood cells that increases their risk of infections.

Complications of albinism include skin disorders as well as social and emotional challenges.

Skin disorders

One of the most serious complications associated with albinism is the risk of sunburn and skin cancer.

Social and emotional factors

The reactions of other people to those with albinism can often have a negative impact on people with the condition.

- Children with albinism may experience name-calling, teasing or questions regarding their appearance, eyewear or visual aid devices.
- Many people with albinism find the word "albino" hurtful because they're being labeled simply on the basis of looks rather than being thought of as individuals.
- People with albinism usually look very different from members of their own families or ethnic groups, so they may feel like outsiders or be treated like outsiders.

All of these factors may contribute to social isolation, poor self-esteem and stress.

A complete diagnostic work-up for albinism includes a:

- Physical exam
- Description of changes in pigmentation
- Thorough exam of the eyes
- Comparison of your child's pigmentation to that of other family members

A medical doctor specializing in vision and eye disorders (ophthalmologist) should conduct your child's eye exam. The exam will include an assessment of potential nystagmus, strabismus and photophobia. The doctor will also use a device to visually inspect the retina and determine if there are signs of abnormal development. A simple test can measure the brain waves produced when light or a reversing pattern is flashed into each eye. This can indicate the presence of misrouted optical nerves.

If your child has only one eye impairment, such as nystagmus, another condition may be the cause. Disorders other than albinism can affect skin pigmentation, but these don't cause all of the visual problems associated with albinism.

Because albinism is a genetic disorder, treatment is limited. But getting proper eye care and monitoring skin for signs of abnormalities are especially important to your child's health.

- Your child will most likely need to wear prescription lenses, and he or she should receive annual eye exams by an ophthalmologist. Although surgery is rarely part of treatment for albinism, your ophthalmologist may recommend surgery on optical muscles to minimize nystagmus. Surgery to correct strabismus may make the condition less noticeable, but it

won't improve vision.

- Your doctor should conduct an annual assessment of your child's skin to screen for skin cancer or lesions that can lead to cancer. Adults with albinism need annual eye and skin exams throughout their lives.

People with Hermansky-Pudlak and Chediak-Higashi syndromes usually require regular specialized care to prevent complications.

You can help your child learn self-care practices that should continue into adulthood:

- **Use low vision aids,** such as a hand-held magnifying glass, a monocular or a magnifier that attaches to glasses.
- **Apply sunscreen** with a sun protection factor (SPF) of at least 30 that protects against both UVA and UVB light.
- **Avoid high-risk sun exposure,** such as being outside in the middle of the day, at high altitudes, and on sunny days with thin cloud cover.
- **Wear protective clothing,** including long-sleeved shirts, long pants and broad-rimmed hats.
- **Protect eyes** by wearing dark, UV-blocking sunglasses or transition lenses that darken in bright light.

Coping with vision impairment

Many people with albinism develop coping skills to adjust to vision impairments. Tilting the head to one side may minimize the effect of nystagmus and improve vision. Holding a book very close can make it easier to read without causing any harm to the eyes.

Coping with educational challenges

Despite visual handicaps and abnormal optic nerve pathways, children with albinism are developmentally normal. If your child has albinism, begin early to work with teachers and school administrators to facilitate measures to help your child adapt. These may include sitting in the front of the class, using large-print books or a tablet computer for learning, avoiding bright light in the learning setting, or allowing more time for taking tests.

If necessary, start with educating the school professionals about what albinism is and how it affects your child. Also ask about services the school can provide to assess your child's needs.

Adjustments to the classroom environment that may help your child include:

- A seat near the front of the classroom
- A tablet computer that can be synced to an interactive whiteboard (SMART board) at the

front of the room, allowing the child to sit farther back in the classroom

- Handouts of the content written on boards or overhead screens
- High-contrast printed documents, such as black type on white paper rather than colored print or paper
- Large-print textbooks
- Other options, such as showing the child how to enlarge font size on a computer screen

Coping with teasing and social isolation

Help your child develop skills to deal with other people's reactions to albinism:

- **Encourage your child to talk** to you about experiences and feelings.
- **Practice responses** to teasing or embarrassing questions.
- **Find a peer support group** or online community through agencies such as the National Organization for Albinism and Hypopigmentation (NOAH). You can reach NOAH at 800-473-2310.
- **Seek the services of a mental health professional,** who can help you and your child develop healthy communication and coping skills.

If a family member has albinism, a genetic counselor can help you understand your chances of having a future child with albinism. He or she can also explain the available tests and help you figure out the pros and cons of testing for your family member.

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