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## What is albinism?

When someone has albinism, their body has trouble creating pigment. Pigment is the material in the body that produces color. Albinism is a hereditary condition, which means it's passed from parent to child. People with albinism have a reduced amount or absence of pigment in their eyes, hair and skin.

There are two basic types of albinism, both of which cause vision problems, including low vision. One type of albinism is called oculocutaneous albinism or OCA. OCA causes someone to have decreased pigment in the eyes, hair, and skin.

The second type of albinism is called ocular albinism. Ocular albinism mainly affects the eyes. The skin and hair are of normal or near-normal color. Because a child with ocular albinism does not have any outward difference in appearance, eye problems may be the first symptoms of albinism.

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## What causes albinism?

Albinism is caused by a genetic mutation that is usually passed from parents to child. The mutation disrupts the production of melanin, the pigment that protects the skin from UV rays. Melanin is also important for the proper development of the eye. Without melanin, the retina and the optic nerve may not develop properly. The retina is the light-sensitive tissue lining the back of the eye, and the optic nerve fibers, help relay images to the brain.



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## What are symptoms of albinism?

Albinism can cause a wide range of eye problems. Not all people with albinism have a noticeable lack of color in their skin or hair. So eye problems may be the first sign of albinism.

Some of the symptoms include:

- **Nystagmus** (when someone's eyes move rapidly and uncontrollably)
- **Strabismus** (misalignment of the eyes)
- **Sensitivity to bright light** (called photophobia)
- **Refractive errors**, including farsightedness (hyperopia), nearsightedness (myopia) and astigmatism
- **Monocular vision** (relying on vision in one eye only)

- **Foveal hypoplasia.** A condition in which the retina (the light-sensitive tissue at the back of the eye) does not develop normally before birth and during infancy
- **An optic nerve problem.** When nerve signals sent from the retina through the optic nerve to the brain do not follow the usual nerve routes; and
- **An iris problem.** When the colored area in the center of the eye does not have enough pigment to screen out stray light coming into the eye.

People with albinism can have vision that ranges from normal to severe vision impairment. Near vision is often better than distance vision. Generally, those who have the least amount of pigment have the poorest vision.

### Who is at risk for albinism?

Albinism is an inherited genetic disorder. Usually, both parents must carry the albinism gene to have a child with albinism. The albinism gene is a recessive gene, meaning that a child has to receive a copy from both parents to have the disorder. If the child gets a copy of the gene from just one parent, he or she will not have symptoms of albinism. If both parents carry the gene, there is a one-in-four chance with each pregnancy that the baby will be born with albinism.

One type of albinism, called X-linked ocular albinism, is usually inherited from the mother. In this case, the gene for albinism is located on an X chromosome. Females have two X chromosomes, while males have one X chromosome and one Y chromosome. X-linked ocular albinism appears almost exclusively in males. The gene for it is passed from mothers

(who carry it without developing the condition) to their sons. The mothers generally have normal vision. For each son born to a mother who carries the gene, there is a one-in-two chance of having X-linked ocular albinism.

Albinism occurs in about one in 17,000 births.

### How is albinism diagnosed?

To diagnose albinism, an ophthalmologist will give you a thorough eye examination. He or she will look for nystagmus, strabismus and photophobia. Any one of these conditions by itself is not necessarily a sign of albinism. An ophthalmologist will also look at the retina to see if it has developed as it should.

### How is albinism treated?

Albinism itself has no treatment. But some conditions that people with albinism have are treatable. Other conditions related to be albinism are manageable.

For example, strabismus can be treated with glasses or surgery. Glasses can also help improve vision and reduce light sensitivity. For children with low vision, low vision aids such as hand-held magnifiers can help. Glasses with small telescopes attached are helpful for older children and adults. These lenses can help with both close and distant vision.

Parents, students and teachers can work together to help a child with albinism. It's important to consider seating, lighting and optical aids in the classroom. These can make learning easier for a child with albinism.



Peer support groups can help children and adults with albinism. These groups can help the individual to:

- feel less isolated
- learn positive attitudes and coping skills from others with low vision; and
- gather valuable resource information.

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## Summary

When people have albinism their body has trouble creating color in their hair, skin or eyes. Albinism can cause many eye problems, such as nystagmus, strabismus, light sensitivity, and low vision. Albinism is a disease that is passed from parent to child. There is no treatment for albinism. But there are ways to treat many of the conditions related to albinism. Strabismus is treated with glasses and surgery. Glasses also can reduce light sensitivity. There are aids such as hand-held magnifiers that help with low vision.

Joining a peer support group can help those with albinism feel less isolated.

If you have any questions about your vision, speak with your ophthalmologist. He or she is committed to protecting your sight.

Get more information about albinism from EyeSmart—provided by the American Academy of Ophthalmology—at [aao.org/albinism-link](http://aao.org/albinism-link).

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