

WHAT IS ALBINISM?

Albinism is a medical problem that people are born with and it runs in families. People with albinism have less pigment (melanin) which gives color to their skin, hair, and eyes [figure 1].



Fig. 1: People with albinism make less melanin (pigment). The result is little or no pigment in eyes, skin, and/or hair. See how the child in the photo has very light-colored hair, skin and eyes.

HOW COMMON IS ALBINISM?

About 1 in 17,000 people have albinism. Albinism affects people from all races. Most children with albinism are born to parents who have normal hair and eye color for their ethnic backgrounds. Some people with albinism have white hair and very light blue eyes; others have blonde hair and blue eyes, and some even have brown hair and brown eyes. The signs of albinism are not always noticeable and a person may not even know that they have albinism.

ARE THERE DIFFERENT TYPES OF ALBINISM?

There are two main types of albinism: Oculocutaneous (pronounced Ock-you-low-kew-TAIN-ee-us) Albinism (OCA) and Ocular Albinism (OA). OCA affects the color of the eyes, hair, and skin, and has 4 different types depending on the related gene problem. OA mainly affects the eyes, with normal or near-normal skin and hair color.

WHAT CAUSES ALBINISM?

Albinism is passed from parents to their children through genes. For most types of OCA, both parents must carry an albinism gene to have a child with albinism. Even if the parents look normal, they can still carry the gene. When both parents carry the gene but don't have albinism themselves, there is a 1 in 4 chance at each pregnancy that the baby will have albinism. This type of inheritance is called autosomal recessive inheritance.

For OA, the gene for albinism is on the X chromosome. Females have two X chromosomes, while males have one X chromosome and one Y chromosome. Mothers (who have the OA gene but don't have albinism themselves) pass the OA gene to their sons. These mothers have subtle signs in their eyes that an ophthalmologist could find, but they generally have normal vision and look normal. For each son born to a mother who carries the gene, there is a 50% chance of having ocular albinism.

WHAT ARE THE EYE FINDINGS OF ALBINISM?

People with albinism can have different eye problems, including:

- **Nystagmus**: They can have shaking eye movements.
- **Abnormal Head Position**: They might turn their head a certain way to lessen eye shaking and improve vision
- **Strabismus**: They might have eyes that drift in or outward.
- **Photophobia**: They might be sensitive to bright light.
- **Refractive Errors**: They might need glasses to see. They can be far-sighted (hyperopia), near-sighted (myopia) or have astigmatism .
- **Foveal hypoplasia**: They might have problems with the growth of the center part of the retina (the inside part of the back of eye). Vision is often blurry from this problem.
- **Optic nerve misrouting**: The nerve signals from the eye to the brain might not follow the usual pathway.
- **Iris transillumination**: The iris (the colored area in the center of the eye) can have little to no color. This allows extra light in the eye and the iris appears to glow (figure 2).

- **Blurred vision:** Vision can be normal for those mildly affected but as bad as legal blindness (vision worse than 20/200) or worse for those with more severe forms of albinism. Near vision is often better than distance vision. Generally, those who have the least amount of color in their eyes have the poorest vision.

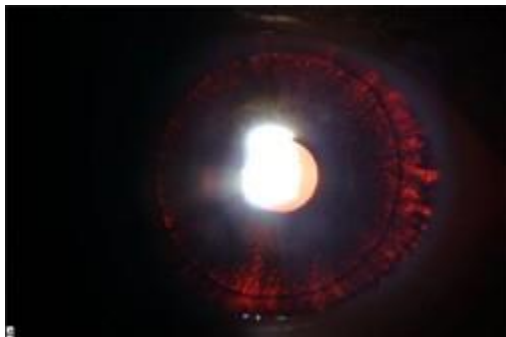


Fig. 2: Photograph showing iris transillumination due to little to no eye color.

HOW IS ALBINISM TREATED?

Albinism is a lifelong medical problem that stays the same with time. Unfortunately, there is no way to get the body to make more color (pigment) and vision is often blurry even with best treatments. However, it is very important to have regular eye care.

Ophthalmologists can give glasses to help with vision and light sensitivity. Magnifying glasses for reading and special aids for distance vision can also be helpful. Eye movement problems can be treated with glasses, eye patching or surgery. Abnormal head turns and nystagmus can get better with glasses or surgery.

For people and families with albinism, genetic counseling is recommended. Counselors can explain the problem in detail, including the chances of future children being affected. Speak with your primary doctor or ophthalmologist about genetic counseling. The American College of Medical Genetics and the



National Society of Genetic Counselors has a list of specialists who can also help: <https://www.nsgc.org/>

WHAT LIFESTYLE CHANGES DO PEOPLE WITH ALBINISM USUALLY NEED?

People with albinism can face challenges due to their blurry vision. It may be hard to see things far away and those with albinism may need special tools to get the most out of their vision. Blurry vision can limit driving and certain job choices. Students, parents, and teachers should work as a team to consider seating, lighting, vision aids, as well as social and emotional growth as a child with albinism learns and grows.

Albinism does not typically affect how smart someone is; children with albinism usually have normal intelligence.

Importantly, people with albinism are more likely to get sunburn and skin cancer. Sunscreen, large hats, sunglasses and/or prescription glasses with tint are strongly encouraged.

WHAT VISION AIDS ARE USEFUL IN ALBINISM?

The use of special tools to help with vision depends on the person. Some people with albinism may do well with ordinary glasses, while others do well with bifocals which have an extra strong lens at the bottom for reading. Contact lenses may work better for some. Others may use hand-held magnifying lenses or special small telescopes. Some prefer to use a tool to make the computer screen larger. For older children and adults, glasses with small telescopes may help with both close-up and far-away vision. The right choice depends on the needs of the person.

WHAT NON-MEDICAL TREATMENTS CAN HELP A PATIENT WITH ALBINISM?

Being a part of support groups can really help people with albinism. These groups share information and help manage feelings about the condition. Both children and adults with albinism can benefit from support groups. Being a part



of a group can help the person to feel less alone, learn positive attitudes and coping skills from others, and gather useful resources and information.

ARE THERE ANY OTHER MEDICAL PROBLEMS IN PEOPLE WITH ALBINISM?

Some people with albinism can have rare medical problems that affect the rest of the body. There are two main medical problems that can be seen in people with albinism.

Hermansky Pudlak Syndrome (HPS) can cause bleeding problems and bruising. Some types of HPS also have lung and bowel/gut problems. HPS is a less common form of albinism but should be checked for if a person with albinism shows unusual bruising or bleeding.

Chédiak-Higashi syndrome affects parts of the body. The disease can cause higher risk of infections, [anemia](#) (low blood count), and [hepatomegaly](#) (enlarged liver).

While these problems are rare, they can happen in some people with albinism and may need special testing and care.

WHERE CAN I FIND MORE INFORMATION ABOUT ALBINISM?

- [NOAH - The National Organization for Albinism and Hypopigmentation](#) PO Box 959, East Hampstead, NH 03826-0959 Phone: 800 473-2310 (US and Canada) | Phone: 603 887-2310 | Fax: 800-648-2310
- [The National Society of Genetic Counselors](#) Maintains a referral list of genetic counselors Phone: 610-872-7608
- <https://eyewiki.org/Albinism> Scientific information on albinism.

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