

Albinism

What is albinism?

The word albinism refers to a large group of inherited conditions. People with albinism have little or no pigment in their eyes, skin, and/or hair. They have inherited genes that do not make the usual amounts of melanin, the major pigment giving color to our skin and eyes [See figure 1].

How common is albinism?

One person in 17,000 has some type of 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.



Fig. 1 People with albinism have inherited genes that code for hypoproduction of melanin. The result is little or no pigment in eyes, skin, and/or hair.

Are there different types of albinism?

These disorders are generally divided into two types.

Oculocutaneous (pronounced Ock-you-low-kew-TAIN-ee-us) albinism involves decreased pigment in the eyes, hair, and skin. Ocular albinism involves primarily the eyes, while the skin and hair show normal or near-normal coloration.

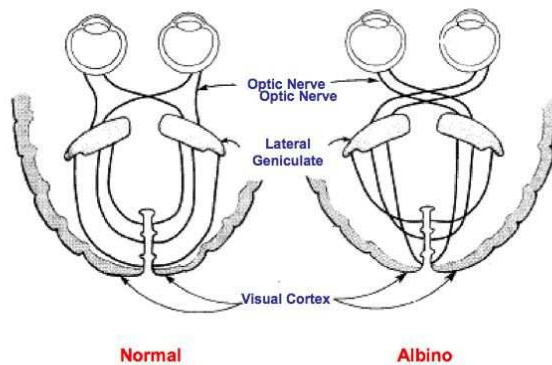
What causes albinism?

Albinism is passed from parents to their children through genes. For most types of albinism, both parents must carry an albinism gene to have a child with albinism. Parents may have normal pigmentation but still carry the gene. When both parents carry the 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.

What is X-linked ocular albinism?

In X-linked inheritance, 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 have subtle eye changes that an ophthalmologist could identify, but they 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.

What are the visual symptoms of albinism?



The symptoms of albinism begin in early childhood and may include decreased vision, sensitivity to bright lights, an unintentional jittery movement of the eyes (nystagmus) and misalignment of the eyes (strabismus). Vision can range from normal, for those minimally affected, to legal blindness 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 pigment have the poorest vision.

Fig. 2 Diagram showing miswiring of nerve fibers.

because of lack of pigment. The fovea, which is the area of the retina which receives the sharpest light images, does not develop normally before birth and in infancy. Therefore vision may not be sharp, even with corrective lenses. Also, there is a misrouting of the optic nerve fibers as they course from the retina to the brain.

These eye problems result from abnormal development of the eye

Finally, the iris, the colored area in the center of the eye, does not have enough pigment to screen out stray light coming into the eye as it normally does [See figure 3].

How is albinism treated?

Albinism is a lifelong condition. Although there is no way to treat poor or absent pigment production or correct early vision development, proper eye evaluation and management can be useful. Eye misalignment can be treated with glasses or surgery. Glasses can be prescribed to improve vision and reduce light sensitivity. Magnifying glasses for reading and low vision aids for distance vision can also be helpful.

Genetic counseling of affected individuals and their families is recommended. Counselors can provide a detailed explanation of the disorder including the chances of future children being affected.

What lifestyle adjustments does albinism usually require?

The reduced visual acuity associated with albinism may result in difficulty in school, such as the inability to read what is written on a chalkboard except when very close, and difficulty with ball sports. It may also result in the inability to drive, and place limitations on vocational choice.

Student, parents, and teachers should work as a team to consider seating, lighting, optical aids, and social and emotional growth.

What optical aids are useful in albinism?

The use of optical aids depends on the individual. Some children may do well with ordinary glasses. For older children and adults, glasses with small telescopes mounted on the lenses may help with both close and distant vision. Contact lenses work satisfactorily for some with albinism.

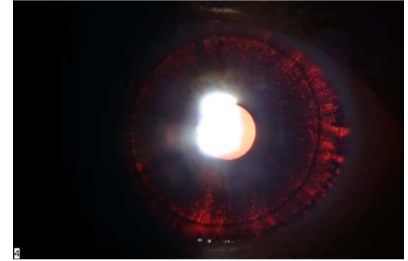


Fig. 3 Slit lamp photograph showing transillumination due to decreased iris pigmentation

What non-medical approaches can help a patient with albinism?

Participation in support groups can help in gathering information, as well as in dealing with feelings of anger or shame about the condition. Children and adults with albinism can benefit from participation in peer support groups. These groups can help the individual to feel less isolated, to learn positive attitudes and coping skills from others with low vision, and to gather valuable resource information.

Where can I find more information about albinism?

For more information visit the NOAH website (National Organization for Albinism and Hypopigmentation). www.albinism.org

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