



Information about Albinism

National Organization for Albinism and Hypopigmentation

Ocular Albinism

Overview

Ocular albinism is an inherited condition affecting pigment cells in the eyes, skin and hair. Ocular albinism causes vision problems and may result in fairer hair and skin than other family members', though hair and skin color are generally within "normal" range for ethnicity.

The following vision problems are common with ocular albinism:

- Reduced visual acuity generally ranging between 20/60 and 20/200, with some cases better or worse;
- Involuntary eye movement, called nystagmus, present from birth or shortly thereafter, and usually most apparent during infancy when the eyes may sweep back and forth in a pendular movement;
- Significant refractive error and astigmatism;
- Crossed eyes or "lazy" eye, called strabismus; and
- Sensitivity to bright light and glare, called photophobia.

Reduced visual acuity from ocular albinism generally results in difficulty reading small type typically used in

publishing and low contrast material, difficulty with depth perception and skills which require tracking small fast-moving objects, difficulty with recognizing people and seeing things at a distance, difficulty with glare, and difficulty with meeting many states' driver's license requirements. Many difficulties can be overcome with accommodation.

Symptoms

With ocular albinism, eye color is normal. However, when an eye doctor examines the inside of the eye, there is usually a blonde fundus, macular hypoplasia, and iris transillumination. The main problem with the eye in ocular albinism is in the fovea, the small area of the retina responsible for fine detail or acute vision. With ocular albinism, the fovea does not develop completely, the same as with oculocutaneous albinism, presumably because normal pigment is needed for the normal ocular development occurring before birth.

Another problem that may occur with ocular albinism is that nerves from the back of the eye to the brain may not follow the usual pattern of routing. From the normal eye, nerve fibers go to both sides of the brain, that is, the same side as the eye and the side opposite from the eye. From the eye with ocular albinism, more of the nerve

fibers may cross from the eye to the opposite side of the brain. A test called a visually evoked potential (VEP) or visually evoked response (VER), which is performed like an EEG or brain wave test, can show this difference. This increased crossover of nerve fibers is also common with oculocutaneous albinism.

Although not obvious to the naked eye, hair and skin cells of people with X-linked ocular albinism are also affected. Ocular albinism results in production of macromelanosomes, enlarged granules of pigment, which can be seen at a microscopic level. Because skin cells are affected in ocular albinism, skin may be more susceptible to sun and ultraviolet light damage.

Genetics

Ocular albinism is X-linked, which means the gene responsible for ocular albinism is located on the X chromosome. Ocular albinism therefore occurs almost exclusively in males. It is almost always passed from mothers who carry the gene to their sons. Each time a mother who carries the gene for ocular albinism gives birth to a son, there is a 1 in 2 chance that the son will have ocular albinism. Each time a mother who carries the gene gives birth to a daughter, there is a 1 in 2 chance that the daughter will be a carrier. Each time an affected male fathers a son, the son will NOT have ocular albinism. Each time an affected male fathers a daughter, the daughter WILL be a carrier. In very rare cases, ocular albinism can manifest in females, if they have a father with ocular albinism and a mother who carries the gene, or if they have a genetic anomaly allowing the ocular albinism X chromosome to penetrate or dominate.

Although ocular albinism is an X-linked condition, and the specific gene responsible for it has now been isolated and identified, the term “ocular albinism” has historically been used to describe or classify anyone with vision problems common to albinism who had normal or near-normal hair and skin color, or substantial pigment. If there was no X-linked inheritance pattern or mottling of the mother’s retina, such cases were sometimes classified as “autosomal recessive ocular albinism” or AROA. However, most experts now agree that several genetic types of oculocutaneous albinism result in substantial skin and hair pigmentation. So, cases classified as AROA may actually be cases of oculocutaneous albinism, either OCA1 or OCA2. (see also the NOAH information bulletin "What is Albinism?" at <http://www.albinism.org>).

Carrier Symptoms

Ocular albinism is one of more than a dozen X-linked genetic conditions in which a woman’s eyes may indicate that she is a carrier. Women who carry the ocular albinism gene may have mottled pigmentation in the back of their eyes and spotty iris transillumination, but do not have macular hypoplasia and the other symptoms of ocular albinism. An eye doctor may be able to identify a female carrier of ocular albinism by the mottling of the retina, though it does not always occur to a significant enough degree or in an area of the retina normally viewed during an eye examination. Mottling is not found in approximately 20 to 50% of carriers. Female carriers may also have depigmented patches or areas of skin.

Diagnosis

Ocular albinism may be identified in males by ophthalmologic or optometric examination of their eyes, their mothers' eyes and by a positive inheritance pattern. However, some mothers may not have any visible mottling in their retinas, and ocular albinism can skip many generations due to few male offspring, or failure of any males to inherit ocular albinism. A skin patch test may reveal macromelanosomes, but may also be inconclusive if no macromelanosomes are contained in the tested patch. A new genetic test for X-linked ocular albinism has been developed which can identify the condition in affected males and female carriers. The test had not been clinically approved in the United States at the time this bulletin went to press, but testing can be obtained through the Vision of Children organization and Baylor Medical Center in Houston, Texas (see www.visionofchildren.org). NOAH makes no representation concerning accuracy or utility of genetic testing, and takes no position on whether individuals should have genetic testing performed. A licensed genetic counselor can offer professional advice.

Effects On Family, Individual

Many parents of children with ocular albinism have more children. Parents report that children with ocular albinism appear to function normally in most situations, despite their visual difficulties, particularly if they receive early and continuing professional vision evaluation, accommodation, support and services. Children with ocular albinism can excel in regular daycare and classroom settings. Adults with ocular albinism can lead independent, productive and full lives, and

successfully pursue a variety of professions and occupations. Many are able to drive. Accommodation is nearly always required. However, normal vision is not fully achieved even with accommodation.

Attention to emotional and social adjustment is important. Parents and people with ocular albinism can feel anger and shame about the condition and try to deny it or pretend it does not exist. This denial can result in the person with ocular albinism experiencing low self-esteem, difficulties in relationships with others, and low functioning in school and work. The diagnosis of a child with ocular albinism can strain the parents' relationship and relationships with other family members. Diagnosis can also lead to anxiety and frustration, because it is difficult to accurately predict how well a child's vision will develop.

It is important for parents of a child with ocular albinism to learn as much as possible about the condition and about low vision. NOAH recommends parents be open and honest with the child, family members, friends, and others about the child's low vision and the reason for it. (see also the NOAH information bulletin "Social and Emotional Aspects of Albinism" at <http://www.albinism.org>)

Participation in support groups such as NOAH, NAPVI (National Association of Parents of Visually Impaired), ANN (American Nystagmus Network), and other groups and organizations can help in gathering information as well as in dealing with feelings about the condition. Children, parents, and adults with ocular albinism can benefit from participation in peer support groups. These groups can help the individual

and family to feel less isolated, to learn positive attitudes and coping skills from others, and to gather valuable resources and information.

NOAH can provide other information bulletins on visual aids, assisting students with albinism in the classroom, and resources for social and emotional support.

Treatment

Treatments for ocular albinism include routine eye examination, visual aids, environmental changes to protect, improve and facilitate vision, and therapies for stimulating visual development in infants and young children. Patching, eye drops and surgery for strabismus can avoid additional loss of vision and depth perception likely to result if strabismus is left untreated. Eye surgery may also have added benefits of improving appearance and self image. Correction of refractive error maximizes vision and can prevent permanent strabismus from developing in some cases.

The choice of optical aids for a child or adult is an individual one. Some children may do well with ordinary glasses. For older children and adults, glasses with small telescopes mounted on the lenses, or a hand held telescope, may help with both close and distant vision. Contact lenses sometimes provide additional correction that glasses cannot. New technology and innovation continues to provide new and improved aids and accommodations for treatment of low vision.

More technical information for health professionals on ocular albinism can be found in:

Lewen RM: Ocular albinism. Arch Ophthalmol 1988, 106:121-122. This is a photo essay about X-linked ocular albinism.

King RA, Hearing VJ, Creel DJ, Oetting WS: Albinism, 4353-4393, in Scriver, Charles R. et al, The Metabolic and Molecular Bases of Inherited Disease, 7th ed., McGraw Hill Inc. Health Professions Division, New York 1995. This textbook is available in most medical libraries.

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