

Sharing General Information with Educators

Items in this section:

- What is Albinism?
- What Do You See?
- What Do You Call Me?
- Albinism Terminology

Purpose

- Use the following information to support conversations about albinism with the general education teacher prior to the first day of school.
- Encourage an open dialogue between parent and teacher/student about any physical, social, or emotional issues pertaining to the diagnosis.
- Refer to the documents if/when the general education teacher has questions.
- Provide a resource list for any inquiries that require more attention or in the event that educators want to seek additional information.

What Is Albinism?

Albinism is an inherited genetic condition that reduces the amount of melanin pigment formed in the **skin, hair and/or eyes**. Albinism occurs in all racial and ethnic groups throughout the world. In the U.S., approximately one in 18,000 to 20,000 people has some type of albinism. In other parts of the world, the occurrence can be as high as one in 3,000. Most children with albinism are born to parents who have normal hair and eye color for their ethnic backgrounds.

A common myth is that people with albinism have red eyes. Although lighting conditions can allow the blood vessels at the back of the eye to be seen, which can cause the eyes to look reddish or violet, most people with albinism have blue eyes, and some have hazel or brown eyes. There are different types of albinism and the amount of pigment in the eyes varies; however, vision problems are associated with albinism.

Vision Considerations

People with albinism have vision problems that are not correctable with eyeglasses, and many have low vision. It's the abnormal development of the retina and abnormal patterns of nerve connections between the eye and the brain that cause vision problems. The presence of these eye problems defines the diagnosis of albinism.

The degree of impairment varies with the different types of albinism. Although people with albinism may be considered "legally blind" with a corrected visual acuity of

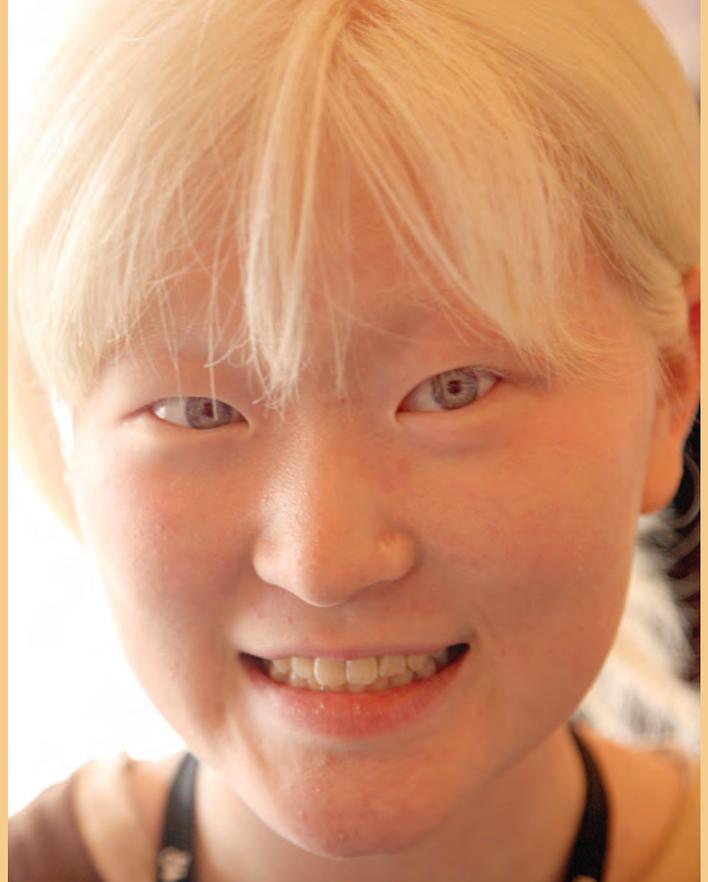


Photo courtesy of Positive Exposure, Rick Guidotti

20/200 or worse, most learn to use their vision in a variety of ways and are able to perform innumerable activities such as reading, riding a bike or fishing. Some have sufficient vision to drive a car.

Dermatological Considerations

Because most people with albinism have fair complexions, it's important to avoid sun damage to the skin and eyes by taking precautions such as wearing sunscreen or sunblock, hats, sunglasses and sun-protective clothing.

Types of Albinism

While most people with albinism have very light skin and hair, levels of pigmentation can vary depending on one's type of albinism. Oculocutaneous (pronounced ock-you-low-kew-TAIN-ee-us) albinism (OCA) involves the eyes, hair and skin.



Photo courtesy of Positive Exposure, Rick Guidotti

Ocular albinism (OA), which is much less common, involves only the eyes, while skin and hair may appear similar or slightly lighter than that of other family members.

Over the years, researchers have used various systems for classifying oculocutaneous albinism. In general, these systems contrasted types of albinism having almost no pigmentation with types having slight pigmentation. In less pigmented types of albinism, hair and skin are cream-colored and vision is often in the range of 20/200. In types with slight pigmentation, hair appears more yellow or has a reddish tinge and vision may be better.



Photo courtesy of Positive Exposure, Rick Guidotti

Recent research has used analysis of DNA, the chemical that encodes genetic information, to arrive at a more precise classification system for albinism. **Seven forms of oculocutaneous albinism are now recognized – OCA1, OCA2, OCA3, OCA4, OCA5, OCA6 and OCA7.** Some are further divided into subtypes.

- **OCA1**, or tyrosinase-related albinism, results from a genetic defect in an enzyme called tyrosinase. This enzyme helps the body to change the amino acid, tyrosine, into pigment. (An amino acid is a “building block” of protein.) There are two subtypes of OCA1. In OCA1A, the enzyme is inactive and no melanin is produced, leading to white hair and very light skin. In OCA1B, the enzyme is minimally active and a small amount of melanin is produced, leading to hair that may darken to blond, yellow/orange or even light brown, as well as slightly more pigment in the skin.

- **OCA2**, or P gene albinism, results from a genetic defect in the P protein that helps the tyrosinase enzyme to function. People with OCA2 make a minimal amount of melanin pigment and can have hair color ranging from very light blond to brown.
- **OCA3** is rarely described and results from a genetic defect in TYRP1, a protein related to tyrosinase. People with OCA3 can have substantial pigment.
- **OCA4** results from a genetic defect in the SLC45A2 protein that helps the tyrosinase enzyme to function. People with OCA4 make a minimal amount of melanin pigment similar to people with OCA2.
- **OCA5–7** were recognized in humans in 2012 and 2013. They have reported mutations on three additional causative genes. As gene testing becomes available, and more people with these types of albinism are identified, the complete range of physical manifestations will be recognized, and may overlap with other known types of OCA. Currently, these types of albinism are considered to be uncommon.

Researchers have also identified several other genes that result in albinism with other features. One group includes at least nine genes leading to **Hermansky-Pudlak Syndrome** (HPS). In addition to albinism, HPS is associated with bleeding problems and bruising. Some forms are also associated with lung and bowel disease. HPS is a less common form of albinism but should be suspected if a person with albinism shows unusual bruising or bleeding or if

a genetic test for a type of OCA produces inconclusive results.

Other albinism-related syndromes include **Chediak-Higashi Syndrome** and **Griscelli Syndrome**.



Photo courtesy of Positive Exposure, Rick Guidotti

Genetics of Albinism

The genes for OCA are located on “autosomal” chromosomes. Autosomes are the 22 pairs of chromosomes that contain genes for our general body characteristics, compared to the one pair of sex chromosomes. We normally have two copies of these chromosomes and the many genes on them – one inherited from our father, the other inherited from our mother. For a recessive trait (like most types of albinism) to occur, both of the person’s chromosomes must carry that trait. That means that most

types of albinism result from inheriting an albinism trait from both the mother and the father who often have normal pigmentation. In this case, the mother and father are considered to be carriers of the albinism trait because they each carry a recessive gene for the condition but do not manifest the condition themselves. When both parents carry the albinism 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.



Photo courtesy of Positive Exposure, Rick Guidotti

with OA1 are males. Parents should be suspicious if a female child is said to have ocular albinism. While possible if the mother is a carrier of ocular albinism and the father has ocular albinism, it is extremely rare.

For couples who have not had a child with albinism, there is no simple test to determine whether a person carries a gene for albinism. Researchers have analyzed the DNA of many people with albinism and found the changes that cause albinism, but these changes are not always in exactly the same place, even for a given type of albinism. Moreover, many of the tests do not find all possible changes. Therefore, the tests for the albinism gene may be inconclusive. If parents have had a child with albinism previously, and if that affected child has had a confirmed diagnosis by DNA analysis, there is a way to test in subsequent pregnancies to see if the fetus has albinism. The test uses either amniocentesis (placing a needle into the uterus to draw off fluid) or chorionic villous sampling (CVS). Cells in the fluid are examined to see if they have an albinism gene from each parent.



Photo courtesy of Positive Exposure, Rick Guidotti

Ocular albinism (OA1) is caused by a change in the GPR143 gene that plays a signaling role that is especially important to pigmentation in the eye. OA1 follows a simpler pattern of inheritance because the gene for OA1 is on the X chromosome. Females have two copies of the X chromosome while males have only one copy (and a Y chromosome that makes them male). To have ocular albinism, a male only needs to inherit one changed copy of the gene for ocular albinism from his carrier mother. Therefore almost all of the people

For specific information and genetic testing, seek the advice of a qualified geneticist or genetic counselor. The American College of Medical Genetics and the National Society of Genetic Counselors maintain a referral list. Those considering prenatal testing should be made aware that people with albinism usually adapt quite well to their disabilities and lead very fulfilling lives.

Vision Rehabilitation

Eye problems in albinism result from the abnormal development of the eye because of a lack of pigment and often include:

- **Nystagmus:** Regular horizontal back and forth movement of the eyes
- **Strabismus:** Muscle imbalance of the eyes, “crossed eyes” (esotropia), “lazy eye” or an eye that deviates out (exotropia)
- **Photophobia:** Sensitivity to bright light and glare
- **Refractive Error:** People with albinism may be either farsighted or nearsighted and usually have astigmatism
- **Foveal hypoplasia:** The retina, the surface inside the eye that receives light, does not develop normally before birth and in infancy
- **Optic nerve misrouting:** The nerve signals from the retina to the brain do not follow the usual nerve routes

The iris, the colored area in the center of the eye, has very little or no pigment to screen out stray light coming into the eye. Light normally enters the eye only through the pupil, the dark opening in

the center of the iris, but in albinism light can pass through the iris as well.

For the most part, treatment consists of visual rehabilitation. Surgery to correct strabismus may improve the appearance of the eyes. However, since surgery will not correct the misrouting of nerves from the eyes to the brain, surgery will not improve eyesight or fine binocular vision. In the case of esotropia or “crossed eyes,” surgery may help vision by expanding the visual field (the area that the eyes can see while looking at one point). Surgical intervention is also available to minimize nystagmus.

People with albinism are sensitive to glare, but they do not prefer to be in the dark, and they need light to see just like anyone else. Sunglasses or tinted contact



Photo courtesy of Positive Exposure, Rick Guidotti

lenses may help outdoors. Indoors, it is important to place lights for reading or close work over a shoulder rather than in front.

Various optical aids are helpful to people with albinism, and the choice of an optical aid depends on how a person uses his or her eyes in jobs, hobbies or other usual activities. Some people do well using bifocals which have a strong



Photo courtesy of Positive Exposure, Rick Guidotti

reading lens, prescription reading glasses or contact lenses. Others use handheld magnifiers or special small telescopes, and some prefer to use screen magnification products on computers.

Some people with albinism use bioptics, glasses which have small telescopes mounted on, in or behind their regular lenses so that one can look through either the regular lens or the telescope. Some states allow the use of bioptic telescopes for driving.

Optometrists or ophthalmologists who are experienced in working with low vision patients can recommend various

optical aids. Clinics should provide instruction in their use. The American Foundation for the Blind maintains a directory of low vision clinics. In Canada, support is available from the Canadian National Institute for the Blind.

Medical Problems

In the United States, most people with albinism live normal life spans and have the same types of general medical problems as the rest of the population. The lives of people with Hermansky-Pudlak Syndrome can be shortened by lung disease or other medical problems.

In tropical countries, people with albinism who do not have access to adequate skin protection may develop life-threatening skin cancers. If they use appropriate skin protection, such as

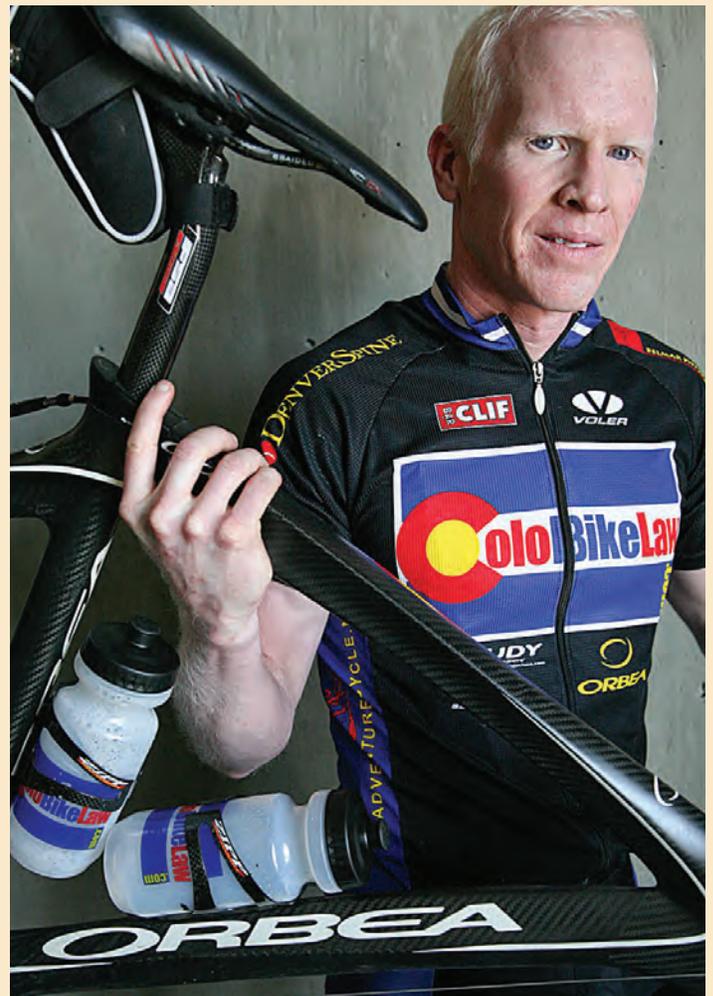


Photo courtesy of Positive Exposure, Rick Guidotti



Photo courtesy of Positive Exposure, Rick Guidotti

sunscreens rated 20 SPF or higher and opaque clothing, people with albinism can enjoy outdoor activities even in summer.

Social Considerations

People with albinism are at risk of isolation because the condition is often misunderstood. Social stigmatization can occur, especially within communities of color, where the race or paternity of a person with albinism may be questioned. Families and schools must make an effort to include children with albinism in group activities. Contact with others with albinism or who have albinism in their families or communities is most helpful. NOAH can provide the names of contacts in many regions of the country.

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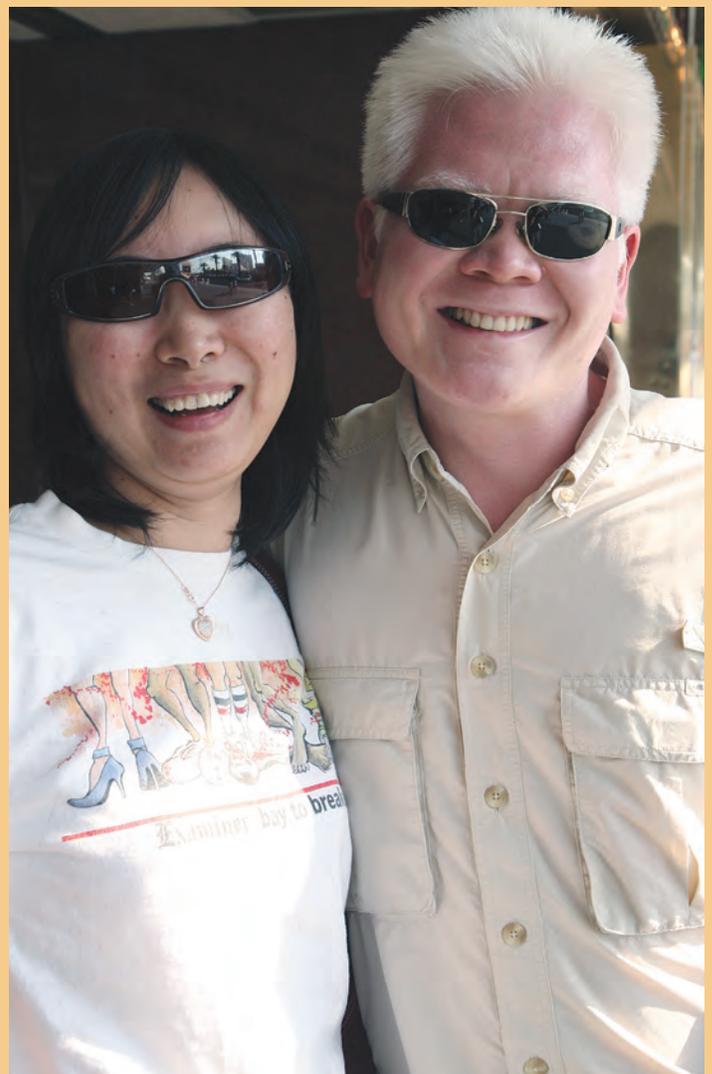


Photo courtesy of Positive Exposure, Rick Guidotti



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What Do You See?

An Adult with Albinism Describes What He Sees

by Matt Bailey

Normally-sighted people often ask those of us with albinism, “What do you see?” Parents of children with albinism are especially interested in the answer. Here you’ll find a brief explanation of how a person with albinism “sees.”

Banishing the Blurry Misconception

There is often a misconception that our vision is blurry. Many people are baffled to learn that it’s not.

When normally-sighted people experience vision problems, it is because the lens in front of the eye doesn’t focus the image clearly onto the back of the eye causing blurriness. Near-sightedness, far-sightedness and astigmatism are “out of focus” problems. Glasses and contacts correct these problems by reshaping the light entering the eye so that images are focused correctly onto the back of the eye. This is similar to the way you would turn the focus knob to correct a blurry image when using binoculars.

Although blurriness isn’t associated with the major vision issue related to albinism, we may be affected by the same “out of focus” problems as normally-sighted people. If near-sightedness, far-sightedness and astigmatism aren’t addressed, our vision could become blurry. That’s why it can be important for young children and even babies with albinism to wear glasses. In many children, the use of their vision and their eyes develop more fully when images entering the eye are focused correctly.



Photo courtesy of Positive Exposure, Rick Guidotti

Clear, but not Hi-Def

Understanding the concept of resolution will help clarify the problems in the back of the eye that albinism causes but glasses cannot fix. All pictures are made up of dots, with each dot capturing a different color and brightness level. The pictures in magazines and newspapers, the pictures on TV and on social media, as well as the pictures taken by digital and film cameras are composed of a bunch of dots. You see more details in a picture that has more dots.

Cross section of the human eye

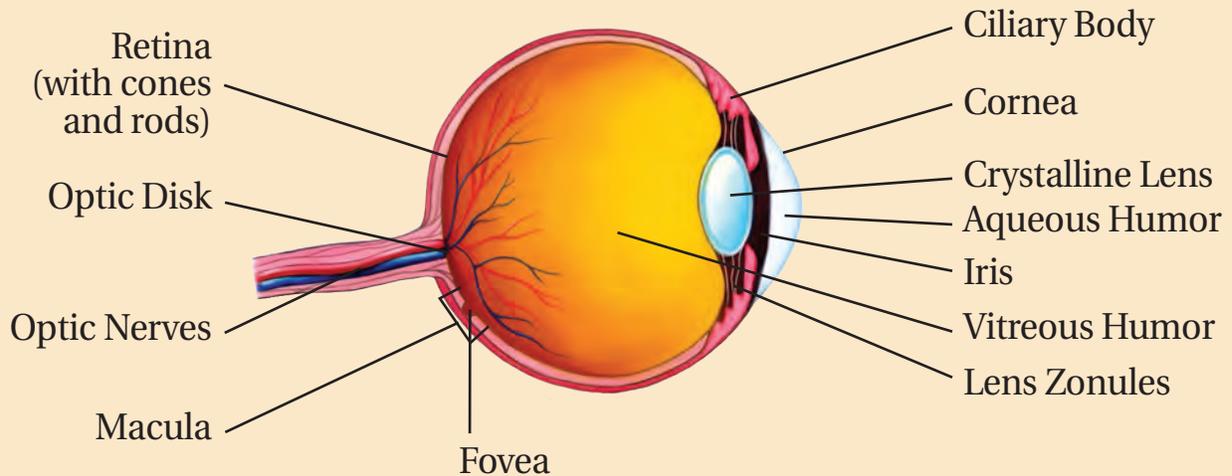


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The picture on the back of the human eye is also made up of dots, millions and millions of them in fact. They're the "cones" and "rods" on the retina in the back of the eye. The primary reason people with albinism can't see as well as normally-sighted people is because we have fewer cones in the fovea which is within the macula. In other words, we have fewer "dots" to make up the picture we see. The cones in particular are the "dots" that people use to see details, such as printed letters and numbers or the details of people's faces.

Nystagmus (the back and forth movement of the eyes) as well as the lack of pigment in the iris and the retina are also contributing factors to our reduced vision, although to a lesser degree.

The easiest way to understand how the lack of cones affects the vision of people with albinism is to turn on your television. If you have upgraded to a High Definition Television (HDTV), you have experienced an upgrade in the "visual acuity" of your television. That's because a high-definition



Photo courtesy of Positive Exposure, Rick Guidotti

Older TV (left) vs. High-Definition TV (right)



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television signal contains five times the visual information. The picture on an old-fashioned TV is made up of five times fewer dots therefore each dot covers a larger portion of the total picture so you can't see as many fine details. When TV manufacturers tout a television's resolution with terms such as "1080p," they're telling you how many "dots" the picture on their TV screen has.

To experience the difference in low visual acuity and normal visual acuity, watch a program in High Definition (HD) on your HDTV. Then, go to an old TV and watch the same program on the standard definition TV. (Alternatively, many cable and satellite TV companies offer both a HD and a Standard Definition (SD) version of the same channel. Simply switch between the HD and SD channel.) Notice how the standard definition image on the old TV isn't blurry. It simply lacks the fine detail of the image on the HDTV. You can still recognize faces, but you'll see more details in HD than on the old TV. You can still follow the action of sports on the old TV, but you won't see the

individual blades of grass on the field the way you can on a HDTV.

Another trick to see how reduced resolution does not make a picture blurry is to watch a video online with a service such as YouTube that allows you to select the quality of the video image. First, watch the video with the highest resolution or quality setting, such as 1080p. Then, switch to a lower resolution or quality setting designed for slower Internet connections, such as 480p. The difference between how those of us with albinism see and normally-sighted people see is a lot like the difference between the low resolution video versus the high resolution video: Neither is blurry, however, we can't quite make out some of the finer details that normally-sighted people see.

Just like that low resolution video or the program on your old-fashioned TV, we often don't need to see the details we're missing to fully understand and participate in the world we see.

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Photos courtesy of Positive Exposure, Rick Guidotti

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What Do You Call Me?

For many people the term “albino” brings to mind images of a person or animal with a pale complexion and pink eyes. The term “albinism,” by contrast, is less commonplace or recognizable to people who may not know a person with the condition. We all know that words can be powerful, so how do you say that someone is an albino without being disrespectful?

Words Can Hurt or Heal

In the albinism community, opinions vary on the use of the word albino. While some find it extraordinarily offensive, others feel the label carries neutral or even empowering connotations. Many people with albinism agree that their feelings depend on the context or intent in which the word is used. The term can be derogatory when said mockingly or with malice, or it can be used innocently by someone who means no offense or is just curious.

Some people with albinism grew up in families or communities that used the word albino often and learned at an early age that there was no shame or negativity in referring to themselves as such. Others may have found that they were only called albino when they were being teased. Some describe their negative association with the word as being as offensive as a racial slur.

Some children and adults with albinism may use the term albino proudly and may feel empowered by “taking back” a once offensive word. By showing the world that they are comfortable calling themselves a word they once found hurtful or derogatory, the word “albino” loses all the power and shock value that it once claimed. By taking personal ownership of one’s condition,



Photo courtesy of Positive Exposure, Rick Guidotti

many people with albinism find that words like albino can no longer hurt them. Of course this is a very personal decision, and not all people with albinism feel comfortable using or hearing the term.

Put the Person First

Although there are many people with albinism who are at peace with the term albino, when dealing with any condition, it is best to put the person first. For instance, say, “a person with albinism” rather than “an albino” or “an albino person.” The rationale for this person-centered language is to do just that: put the person ahead of the condition. When a person is referred to as an albino, he or she is essentially being reduced to and defined by nothing more than their condition. It’s as though the world looks at that person and sees only the condition of albinism. In this way, it can feel like a dehumanizing label.



Photo courtesy of Positive Exposure, Rick Guidotti

To most in the albinism community, the term “person with albinism” will always be a kinder, gentler, less shocking term. Regardless of the context, the word albino can sometimes be an ugly, jolting word to many, especially when heard unexpectedly. If you’re ever unsure, just ask. In a world of political correctness run rampant, you shouldn’t feel the need to tiptoe around the obvious. Every person with albinism will have personal experiences and opinions about the label. As one NOAH member said, “I may be an albino, but above all else, I am a person. I just happen to have the condition of albinism.”

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Albinism Terminology

Oculocutaneous Albinism (OCA): Oculocutaneous (pronounced ock-you-low-kew-TAIN-ee-us) Albinism is an inherited genetic condition characterized by the lack of or diminished pigment in the hair, skin, and eyes. Implications of this condition include eye and skin sensitivities to light and visual impairment.

Ocular Albinism (OA): Ocular Albinism is an inherited genetic condition, diagnosed predominantly in males, characterized by the lack of pigment in the eyes. Implications of this condition include eye sensitivities to light and visual impairment.

Hermansky Pudlak Syndrome (HPS): Hermansky-Pudlak Syndrome is a type of albinism which includes a bleeding tendency and lung disease. HPS may also include inflammatory bowel disease or kidney disease. The severity of these problems varies much from person to person, and the condition can be difficult to diagnose with traditional blood tests

Chediak Higashi Syndrome: Chediak Higashi Syndrome is a type of albinism in which the immune system is affected. Illnesses and infections are common from infancy and can be severe. Issues also arise with blood clotting and severe bleeding.

Melanin: Melanin is pigment found in a group of cells called melanocytes in most organisms. In albinism, the production of melanin is impaired or completely lacking.

Nystagmus: Nystagmus is an involuntary movement of the eyes in either a vertical, horizontal, pendular, or circular pattern caused by a problem with the visual pathway from the eye to the brain. As a result, both eyes are unable to hold steady on objects being viewed. Nystagmus may be accompanied by unusual head positions and head nodding in an attempt to compensate for the condition. Nystagmus appears more prominent as students focus visually on a target.

Null Point: A null point refers to the point in which a student has positioned his/her head to greatly reduce or eliminate nystagmus.

Strabismus: Strabismus is a misalignment of the eyes caused by a muscle imbalance. The eyes deviate upward, downward, inward, or outward simultaneously or independently of each other.

Photophobia: Photophobia is an abnormal discomfort or sensitivity to light and glare. Individuals with albinism experience photophobia because the lack of pigment in the iris does not allow the eyes to filter out light.

Refractive Error: Refractive errors are vision problems that happen when the shape of the eye keeps you from focusing well. The cause could be the length of the eyeball

(longer or shorter), changes in the shape of the cornea, or aging of the lens. This could manifest in a student being nearsighted or farsighted. Students with albinism sometimes experience refractive errors in addition to the impaired vision associated with albinism, but this is not always the case, thus, not all students with albinism will be prescribed or benefit from glasses.

Foveal Hypoplasia: The retina, the surface inside the eye that receives light, does not develop normally before birth and in infancy.

Optic Nerve Misrouting: The nerve signals from the retina to the brain do not follow the usual nerve routes.

Optical Devices: An optical device is a tool used by a student with low vision to improve access to a given near or distance tasks. These devices are typically prescribed by a Low Vision Specialist, and some examples include hand-held magnifiers, monoculars (telescopes), and/or video magnifiers.

Scotoma: A scotoma refers to a partial loss of vision in an area of the eye where otherwise normal vision exists. It is also known as a blindspot.