

ALBINISM

Other Common Names

- Hypopigmentation

Causes/Etiology

- Most types of albinism are inherited when a child receives the albinism gene from both parents.
- There is an exception with one type of ocular albinism which is passed on from mothers to sons.
- Albinism occurs when there is a mutation in one of several genes.
- These genes provide chemically coded instructions for making proteins involved in the production of melanin, the pigment that gives skin, hair, and eyes their color.
- A mutation may result in no melanin production at all or a significant decline in the amount produced.

Incidence

- In the U.S., 1 person in 17,000 has some type of 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. Albinism can occur through autosomal recessive inheritance. When both parents are carriers with one defective gene each and neither parent has albinism they have a 1 in 4 chance of passing on both copies of the defective gene to the child, who will have albinism.

Characteristics

- Albinism is a group of genetic conditions that results in little or no production of the pigment melanin.
- Most individuals with albinism are very sensitive to sun exposure and are at increased risk of developing skin cancer.
- All forms of albinism cause problems with the development and function of the eyes, and visual impairments are common among all types of albinism.
- These impairments are caused by irregular development of the nerve pathways from the eye to the brain and from abnormal development of the retina.
- The system for classifying types of albinism is based primarily on which mutated gene caused the disorder.

Types of Albinism

- **Oculocutaneous albinism**—caused by a mutation in 1 of 4 genes. These mutations result in symptoms affecting vision, as well as skin, hair, and iris color.
 - **Type 1**— Caused by a mutation in a gene on chromosome 11. Some people with this disorder never experience changes in pigmentation, but others begin to produce melanin during childhood and adolescence. Their hair may become a golden blond or brown. Their skin usually doesn't change color, but it may tan somewhat. The irises may also change color and lose some of their translucence. This group is divided into several subtypes depending on associated characteristics.
 - **Type 2**— Caused by a mutation in a gene on chromosome 15. This form of albinism is more common among Sub-Saharan Africans, African-Americans and Native Americans. Their hair may be yellow, auburn, ginger or red, and their eyes can be blue-gray or tan. In people of African descent, the skin may be light brown, and for those of Asian or Northern European descent, the skin is usually white. Typically, skin color is close to the family's coloring, but a little bit lighter. If skin is exposed to the sun, freckles, moles or lentigines may develop over time.
 - Type 1 and Type 2 are the most common forms of this condition. Both types of albinism are usually associated with visual problems including nystagmus (abnormal shifting movements of the eyes) and decreased visual acuity, which is frequently not fully improved with glasses or contact lenses.
 - **Type 3**— Caused by a gene mutation on chromosome 9. People with this rare type of albinism have reddish brown skin, reddish hair and hazel or brown eyes. This form of albinism generally affects black South Africans.
 - **Type 4**— Caused by a gene mutation on chromosome 5. It is also a rare form of albinism. It is similar to Type 2. Most often affects people of East Asian descent.
- **X-Linked Ocular Albinism**— Caused by a gene mutation on the X chromosome. Occurs almost exclusively in males. Vision problems occur without changes in skin or hair.
- **Hermansky-Pudlak Syndrome**— Rare form of albinism caused by a mutation in 1 of at least 8 genes associated with this syndrome. Along with having symptoms similar to oculocutaneous albinism, people also may have lung, bowel, and bleeding problems.
- **Chediak-Higashi Syndrome**— Rare form of albinism that is caused by a mutation in the LYST gene. People generally have symptoms similar to oculocutaneous

albinism. Their hair is usually brown or blond with a silvery sheen, and their skin is typically creamy white to grayish. People with this syndrome also have problems with their immune system because they have defects in their white blood cells. This leads people to be susceptible to infections.

IDEA Category

- A child with albinism may be diagnosed under the category of Visual Impairment. This indicates that the child has an impairment in vision that even with correction, adversely affects his/her educational performance. The term includes both partial sight and blindness

DSM-IV Category

- Albinism does not have a DSM-IV diagnosis, but can be classified by the ICD-10-CM.
- The ICD-10-CM classifies different types of albinism under the hierarchy code E70.3.
- Albinism is considered a metabolic disorder, more specifically, a disorder of aromatic amino-acid metabolism.

Deficits/Complications

- Vision problems-visual impairments/blindness
- Medical problems- High risk of developing skin cancer. In addition to albinism, could have Hermansky-Pudlak Syndrome or Chediak-Higashi Syndrome.
- Social and emotional problems- reactions of other students may have a negative impact on children with albinism and put them at risk for developing low self esteem, social isolation, and stress.
- Educational problems-Albinism doesn't limit intellectual development, but students with this condition may have trouble functioning in the classroom due to their visual impairments.

Long-Term Developmental Outcomes

- 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 problems.
- People with albinism are at higher risk for developing skin cancer.

Assessment Approaches

- A complete diagnostic work-up for albinism includes:
 - Physical examination
 - Description of changes in pigmentation

- Thorough examination of the eyes
- Comparison of child's pigmentation to that of other family members
- An ophthalmologist (medical doctor specializing in vision and eye disorders) conducts the child's eye exam.
- The exam will include an assessment of potential nystagmus (involuntary eye movement), strabismus (misalignment of eyes) and photophobia (abnormal intolerance to perception of light).
- The doctor will also use a device to visually inspect the retina and determine if there are any signs of abnormal development.
- A test called an electroretinogram, which measures brain waves produced when light is shined into the eyes, can indicate the presence of misrouted optical nerves.

Intervention and Treatments

- There is no cure for albinism.
- Because albinism is a genetic disorder, treatment is limited.
- Most treatments involve visual rehabilitation.
- Many children will need to wear prescription lenses, which can provide improvements in their vision.
- Children should receive annual examinations by an ophthalmologist.
- Surgery is rarely part of treatment for albinism, however in some cases, an ophthalmologist may recommend surgery on optical muscles that minimize nystagmus.
- Surgery can also be done to correct for strabismus. While this may make the condition less noticeable, it will not improve vision.
- Children should receive annual skin assessments to screen for skin cancer or lesions that could lead to cancer.
- Adults with albinism need annual eye and skin exams throughout their lives.
- People with Hermansky-Pudlak and Chediak-Higashi syndromes usually require regular specialized care to prevent complications.
- Parents can help their children learn self-care practices that they will continue using into adulthood:
 - Use low vision aids, such as a hand-held magnifying glass, a monocular or a magnifier that attaches to glasses.
 - Apply sunscreens with a sun protection factor (SPF) of at least 30 that protects against both UVA and UVB light.
 - Avoid high-risk sun exposure, such as being outside in the middle of the day, at high altitudes and on sunny days with thin cloud cover.
 - Wear protective clothing, including long-sleeved shirts, long pants and broad-rimmed hats.
 - Protect your eyes by wearing dark, UV-blocking sunglasses.

- Teachers can make adjustments to the classroom environment to assist children with albinism:
 - A seat near the front of the classroom
 - Handouts of the content written on boards or overhead screens
 - High-contrast printed documents, such as black type on white paper rather than colored print or paper
 - Large-print textbooks

Role of the School Psychologist

- Interventions and services that school psychologists can provide to children with albinism will vary depending on the type and severity of the condition.
- School psychologists can work with parents to help them understand how albinism can affect their child's development. They are also able to provide parents with community resources such as medical or other mental health professionals.
- Children with albinism may feel socially isolated from their peers. School psychologists can work with these children on social skills and help them to feel more connected to school.
- School psychologists can educate students and school personnel about the characteristics of albinism.
- School psychologists can collaborate with school personnel to help create a positive and safe school environment where children with albinism feel welcome and appreciated.
- Children with albinism may have difficulties functioning in class due to their visual impairments. School psychologists can work with teachers to design classroom strategies to help these children be successful in the classroom.

Support Groups

- National Organization for Albinism and Hypopigmentation -- www.albinism.org
- International Albinism Center -- <http://albinismdb.med.umn.edu>
- Hermansky-Pudlak Syndrome Network -- www.hpsnetwork.org

Resources

Kids Health: http://kidshealth.org/kid/health_problems/skin/albinism.html#

Mayo Clinic: <http://www.mayoclinic.com/health/albinism/DS00941/TAB=resources>

National Dissemination Center for Children with Disabilities:
<http://nichcy.org/disability/categories>

The National Organization for Albinism and Hypopigmentation:
<http://www.albinism.org/faq/children.html>