

Types and symptoms



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Albinism is a genetic anomaly characterized by little or no pigment in their eyes, skin, or hair. Not specific to race or ethnicity, it can affect African-Americans, Hispanics, and Asians as much as it does whites. More exactly, albinism is an autosomal recessive trait where melanocytes have diminished or restricted ability to produce melanin—the substance responsible for skin, hair and eye color.

Phenotypically, albinism is relatively rare: one person in 17,000 in the U. S. A. is affected. In fact, most children with albinism are born to parents with normal hair and eye color. In some Scandinavian countries, however, it is difficult to tell if a child has albinism because fair hair and eyes are the norm. A common myth is that by definition people with albinism have red eyes. In fact there are different types of albinism, and the amount of pigment in the eyes varies.

The National Organization for Albinism and Hypopigmentation dispels many myths and legends in popular culture, “ Although some individuals with albinism have reddish or violet eyes, most have blue eyes. Some have hazel or brown eyes.

People with albinism always have problems with vision, and many have low vision. Many are "legally blind," but most use their vision for reading, and do not use braille. Some have vision good enough to drive a car. Vision problems in albinism result from abnormal development of the retina and abnormal patterns of nerve connections between the eye and the brain. It is the presence of these eye problems that defines the diagnosis of albinism.

Therefore the main test for albinism is simply an eye exam. While most people with albinism have very light skin and hair, not all do.

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Oculocutaneous albinism involves the eyes, hair, and skin. Ocular albinism involves primarily 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 red-tinged, and vision often corrects to 20/60.

Early descriptions of albinism called these main categories of albinism "complete" and "incomplete" albinism. Later researchers used a test that involved plucking a hair root, and seeing if it would make pigment in a test tube. This test separated "ty-neg" (no pigment) from "ty-pos" (some pigment). Further research showed that this test was inconsistent, and added little information to the clinical exam."