Albinism occurs congenitally due to an error in the production of melanin despite normal numbers of melanocytes. Melanin is directly responsible for the pigmentation of the skin, hair and eyes. The error in production is a genetic defect of the enzyme tyrosinase that causes the body to be unable to make or transport melanin to its desired location. Most commonly albinism is inherited from previous generations of family with each parent having an albinism gene to give to their child. An estimated 1 in 17000 suffer from one form of albinism. People with albinism are commonly referred to as albinos.

Albinism occurs in two main forms. The more severe and most recognizable form is oculocutaneous albinism which affects the eyes, skin and hair. The lack of melanin, which produces the brown, black and yellow skin colors, results in individuals that appear white or very pale. These skin findings may be generalized over the entire body or in patchy areas. This is important as melanin is responsible for protecting against ultraviolet radiation. Albinism’s lack of skin pigmentation therefore makes sunburns and skin cancers much more common and problematic. A variety of the oculocutaneous form is more common in Sub-Saharan Africans, African-Americans, and Native Americans. The second main type is ocular albinism which only affects the eyes. In this form, the person’s skin and hair usually have colors similar to variety seen in other individuals.

Both forms are associated with a number of vision effects including light sensitivity, rapid eye movements (nystagmus), inability for the eyes to move in unison (strabismus), and astigmatism. These impairments result from abnormal nerve development pathways from the eye to brain and abnormal retinal development. The lack of coloration also explains the unusual eye iris coloring. The eye normally produces enough pigment to color the eye blue, but it is often noted that the eyes appear a red color. This is explained by the red of retina being visible through the iris.

Syndromes that can be associated with albinism include Hermansky-Pudlak syndrome (HPS) and Chediak-Higashi syndrome. HPS is a form of albinism caused by a single gene often diagnosed through history of easy bruising, frequent nosebleeds, or bleeding following dental work or surgery. It can occur with a bleeding disorder, as well as with lung and bowel diseases. Chediak-Higashi syndrome may present with a history of frequent infections, silvery hair, and neurological issues.

Individuals afflicted with albinism are normally as healthy as any other individual. That is not to say that their life is easy. People with albinism often suffer a gamete of social and emotional challenges due to their difference in appearance. Young children especially may fall victim to social isolation, low self-esteem, and unwarranted stress. For this reason, a strong support network is recommended. Coping with this ailment may also be augmented by advanced programs such as the National Organization of Albinism and Hypopigmentation (NOAH) and the Albinism World Alliance (AWA). Skin problems are also possible. Although albinism itself does not directly lead to increased mortality, the lack of pigmentation predisposes individuals to skin cancer and other problems caused by their susceptibility to ultraviolet radiation. For this reason, protection from the sun is a key preventative measure. Vision aids such as glasses and magnifiers as well as UV-blocking sunglasses are highly encouraged. Albinos should always use sunscreen (SPF>30 with UVA and UVB protection) and wear protective clothing such as hats, long sleeves, and pants. Sun avoidance all together is also recommended.

The main doctors that need to be involved in the care of an individual with albinism include, but are not limited to a primary care physician, a dermatologist, and an ophthalmologist. It is essential for preventative care to be given by the primary doctor and dermatologist, new skin lesions to be evaluated by the dermatologist, and regular eye exams including electroretinogram to be performed by the ophthalmologist. Eye surgery may also be recommended to correct many of the early visual impairments. In many cases, treatment will have to begin immediately after birth. Overall, however, with the right guidance and preventative measures, an individual with albinism may lead a full and satisfactory life.