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Short Communication

# Albinism and its Mechanism in Vision Impairment

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## DESCRIPTION

Albinism is a hereditary disorder that causes a decrease in melanin synthesis. A pigment that provides colour to skin and hair, resulting in fair skin, light eyes and hair, and a greater susceptibility to numerous skin and eye disorders. The pigment called melanin is what gives skin and hair colour [1]. It can be found in the epidermis, the skin's outer layer, the hair, and the eyes. Melanin protects skin from the sun's Ultraviolet (UV) radiation, which helps to prevent skin damage and cancer. Eumelanin comes in two varieties. Pheomelanin, a dark pigment that imparts brown to black colour, is the most prevalent kind of melanin. The relative quantities of these two types are determined by a person's genetic composition, which is handed down from their parents. Albinism is classified into two types: Oculo-Cutaneous Albinism (OCA), which affects the eyes, skin, and hair, and Ocular Albinism (OA), which only affects the eyes. Albinism can affect any ethnic group. This sickness is believed to impact one in every 20,000 Americans and one in every 17,000 Australians. Albinism is more common in some places of the world, like as Zimbabwe, Africa, where one in every 1,000 persons is affected.

Albinisms are mostly autosomal recessive genetic diseases. The majority of albinisms are autosomal recessive, meaning that unaffected parents pass the disease on to their children [2]. Because chromosomes are paired, all genes on non-sex (autosomal) chromosomes exist in two copies, or alleles. A mutation (a change in the DNA sequence) in one allele of a gene that causes albinism is required, but both parents must also have the normal allele. In carriers of albinism, who have one faulty allele but no symptoms, two copies of the defective gene (one from each parent) are necessary to produce symptoms [3]. Several genes involved in pigmentation and albinism produce or secrete melanin. To understand how these genes can cause albinism, let's look at how the proteins generated by these genes, known as their products, help melanin-producing cells (also known as melanocytes) make pigment. Enzymes are a type of protein that aids in the occurrence of chemical processes. Enzymes convert tyrosine, the starting component, into melanin. The distinction between eumelanin and pheomelanin is related to how tyrosine is altered by enzymes. A mutant version of one of these enzymes may be unable to execute its intended role, affecting the melanocyte's ability to create different types of melanin [4].The ratio of eumelanin (brown-black pigment) to pheomelanin (yellow-red pigment) causes differences in skin colour in normal pigmentation. In albinism, melanocytes are no longer able to produce enough eumelanin, leading in the development of more or exclusively pheomelanin. Distinct genes produce seven different forms of OCA. The degree of OCA can range from complete lack of pigmentation in the skin, hair, and eyes in OCA type 1A to light brown pigmentation in OCA type 2 or reddish-brown pigmentation in OCA type 3. Due to their greater UV sensitivity, people with albinism are more likely to have skin cancer and sunburns. The likelihood of developing skin cancer increases with the amount of UV radiation damage. By producing molecules known as free radicals, which break DNA in the skin, UV radiation helps to trigger skin cancer. By absorbing free radicals, eumelanin often offers protection against UV-induced DNA damage. In fact, pheomelanin increases the production of free radicals. Therefore, both decreased sun protection and increased free radical generation contribute to skin cancer development in people with albinism. Risks of skin damage and skin cancer vary among various forms of albinism. For instance, skin cancer is the primary killer of several ethnic groups, such African albinos. By wearing clothes that covers the skin, using sunscreen, and getting regular skin checks, skin cancer can be avoided. The need of sun protection and regular skin checks comes from the fact that skin cancers that are discovered later in their progression are more challenging to cure.

### Mechanisms of vision impairment in albinism

The mechanisms of visual impairment in albinism are multifaceted and unquestionably linked to pigmentation. Infoveal hypoplasia, which occurs in albinism, causes the central cones to be spread apart, resulting in lower central visual acuity while maintaining normal peripheral visual acuity [5]. Refractive errors, particularly astigmatism, are more prevalent. As a result,

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some amblyopia may be present. It is highlighted that many newborns' refractive error is caused by inadequate visual acuity rather than its course. The albino is predisposed to strabismus. Stereopsis is lacking as a result of optic pathway misrouting. The retinal picture will also be degraded by intraocular light scatter.

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