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**Albinism**

Albinism (from Latin albus, "white"; see extended etymology, also called achromia, achromasia, or achromatosis) is a congenital disorder characterized by the complete or partial absence of pigment in the skin, hair and eyes due to absence or defect of tyrosinase, a copper-containing enzyme involved in the production of melanin. It is the opposite of melanism.



Albinism results from inheritance of recessive gene alleles and is known to affect all vertebrates, including humans. While an organism with complete absence of melanin is called an albino (UK /ælˈbiːnoʊ/,[1] or US /ælˈbaɪnoʊ/)[ an organism with only a diminished amount of melanin is described as albinoid.

Albinism is associated with a number of vision defects, such as photophobia, nystagmus and astigmatism. Lack of skin pigmentation makes for more susceptibility to sunburn and skin cancers. In rare cases such as Chédiak–Higashi syndrome, albinism may be associated with deficiencies in the transportation of melanin granules. This also affects essential granules present in immune cells leading to increased susceptibility to infection.

**Genetics**

Oculocutaneous albinism is generally the result of the biological inheritance of genetically recessive alleles (genes) passed from both parents of an individual for example OCA1 and OCA2. A mutation in the human TRP-1 gene may result in the deregulation of melanocyte tyrosinase enzymes, a change that is hypothesized to promote brown versus black melanin synthesis, resulting in a third oculocutaneous albinism (OCA) genotype, ″OCA3.″ Some rare forms are inherited from only one parent. There are other genetic mutations which are proven to be associated with albinism. All alterations, however, lead to changes in melanin production in the body. Some of these are associated with increased risk of skin cancer (see list of such genetic variations).



The chance of offspring with albinism resulting from the pairing of an organism with albinism and one without albinism is low. However, because organisms (including humans) can be carriers of genes for albinism without exhibiting any traits, albinistic offspring can be produced by two non-albinistic parents. Albinism usually occurs with equal frequency in both sexes. An exception to this is ocular albinism, which it is passed on to offspring through X-linked inheritance. Thus, ocular albinism occurs more frequently in males as they have a single X and Y chromosome, unlike females, whose genetics are characterized by two X chromosomes.

There are two different forms of albinism: a partial lack of the melanin is known as hypomelanism, or hypomelanosis, and the total absence of melanin is known as amelanism or amelanosis.

**Leucism**

Leucism /ˈljuːsɪzəm/ is a condition in animals characterized by reduced pigmentation. Unlike albinism, it is caused by a reduction in all types of skin pigment, not just melanin.



Leucism (occasionally spelled leukism) is a general term for the phenotype resulting from defects in pigment cell differentiation and/or migration from the neural crest to skin, hair, or feathers during development. This results in either the entire surface (if all pigment cells fail to develop) or patches of body surface (if only a subset are defective) having a lack of cells capable of making pigment.

Since all pigment cell-types differentiate from the same multipotent precursor cell-type, leucism can cause the reduction in all types of pigment. This is in contrast to albinism, for which leucism is often mistaken. Albinism results in the reduction of melanin production only, though the melanocyte (or melanophore) is still present. Thus in species that have other pigment cell-types, for example xanthophores, albinos are not entirely white, but instead display a pale yellow colour.

More common than a complete absence of pigment cells is localized or incomplete hypopigmentation, resulting in irregular patches of white on an animal that otherwise has normal colouring and patterning. This partial leucism is known as a "pied" or "piebald" effect; and the ratio of white to normal-coloured skin can vary considerably not only between generations, but between different offspring from the same parents, and even between members of the same litter. This is notable in horses, cows, cats, dogs, the urban crow and the ball python but is also found in many other species.

A further difference between albinism and leucism is in eye colour. Due to the lack of melanin production in both the retinal pigmented epithelium (RPE) and iris, albinos typically have red eyes due to the underlying blood vessels showing through. In contrast, most leucistic animals have normally coloured eyes. This is because the melanocytes of the RPE are not derived from the neural crest, instead an outpouching of the neural tube generates the optic cup which, in turn, forms the retina. As these cells are from an independent developmental origin, they are typically unaffected by the genetic cause of leucism. Genes that, when mutated, can cause leucism include, c-kit, mitf and EDNRB.

References

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