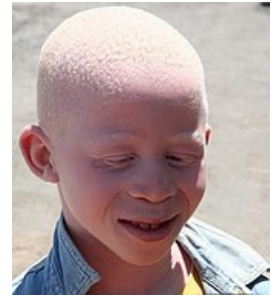


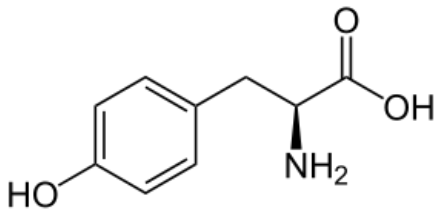
# Albinism

Albinism is a rare, hereditary (AR) metabolic disorder due to a defect in the enzyme *tyrosinase*. The disorder results in a partial or complete absence of **melanin**. It can occur in humans, animals and plants.

Albinism is phenotypically divided into tyrosinase-positive and tyrosinase-negative, genotypically into *OCA 1* to *OCA 3*.



## Biochemical essence



Melanin is a pigment synthesized in the melanosomes (derived from the Golgi apparatus) of melanocytes (epidermal cells) from the amino acid **tyrosine** (see figure). This enzyme has dual activity: tyrosine hydroxylase and dopaoxidase. Under normal circumstances, tyrosine is first hydroxylated by *tyrosinase* to **DOPA** – i.e. 3,4-dihydroxyphenylalanine, and then oxidized to dopaquinone. The next enzyme in the sequence catalyzes the tautomerization of the intermediate. The final product subsequently polymerizes into eumelanin.

If there is a defect in the region of the gene encoding tyrosinase (facial protein), the metabolic pathway cannot proceed and the pigment is not synthesized.

### OCA 1

Mutation of the structural gene for tyrosinase.

- *OCA 1A* (tyrosinase negative) – **complete absence of tyrosinase**
- *OCA 1B* (yellow albinism - xanthoalbinism) - weak enzyme activity in the hair

### OCA 2

Mutation of the structural gene for *P protein* (melanosomal transmembrane protein with a **transport** function – tyrosine transport), the most common type of albinism.

### OCA 3

The so-called brown albinism – mutation of the gene for *TRP-1*, is known only in blacks Albinism is also often accompanied by *phenylketonuria* (AR disease, caused by a defect in the enzyme *phenylalanine hydroxylase*).

## Clinical Picture

### Total albinism

Albinism is manifested by white skin and hair, red or light blue eyes. The skin is very sensitive to UV radiation and at the same time there is a higher risk of developing skin tumors. It also causes faster aging of the skin.

### Partial albinism

This is partial albinism manifested only locally, for example on areas of the skin and areas of hair. The eyes are not affected in this type.

## Links

## Related Articles

- [Pigment](#)

## External links

- [Albinismus \(Czech wikipedia\)](#)
- [Albinism \(English wikipedia\)](#)

## Sources

- ŠTORK, Jiří, et al. *Dermatovenerologie*. 2. vydání. Praha : Galén, 2013. 502 s. ISBN 978-80-7262-898-8.
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- MATOUŠ, Bohuslav. *Základy lékařské chemie a biochemie*. 1. vydání. Praha : Galén, 2010. 540 s.