Melanin synthesis & Albinism

Sharad Gautam
Lecturer, Biochemistry

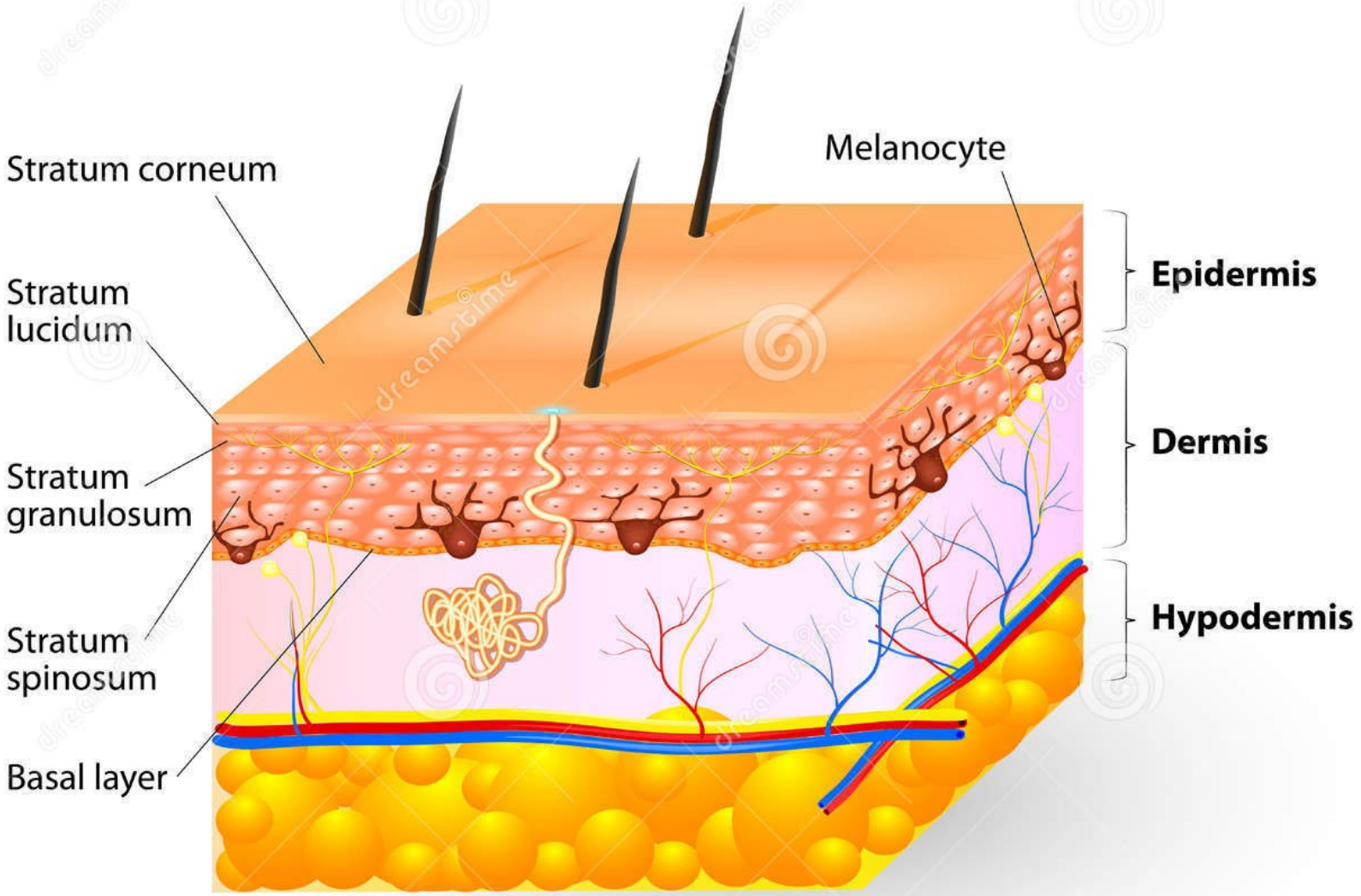
Melanin

- *Skin color is determined by the relative concentrations of black & red melanins*
- *This is dependent on genetic and environmental factors*
- *Moles: due to hyperpigmentation caused by the hyperactivity of melanocytes*
- *White patches: due to localized absence or degeneration of melanocytes from the skin.*

Melanin synthesis

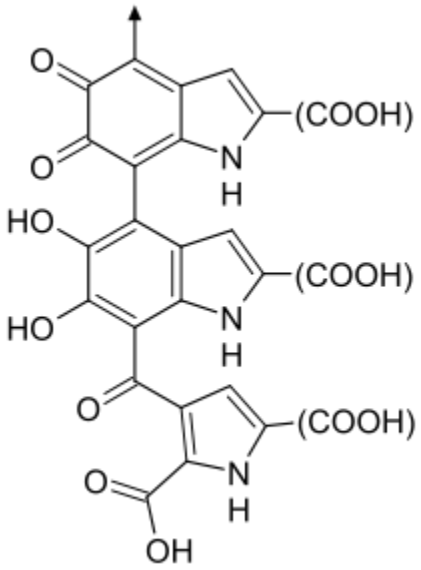
- *Melanin is black pigment of skin, hair & eyes*
- *Synthesis occurs in melanosomes present in melanocytes.*
- *Tyrosine is the precursor of melanin and tyrosinase is the enzyme involved in it's formation*
- *Melanochromes formed from tyrosine polymerizes to form melanin polymers*

THE LAYERS OF HUMAN SKIN



Melanin types

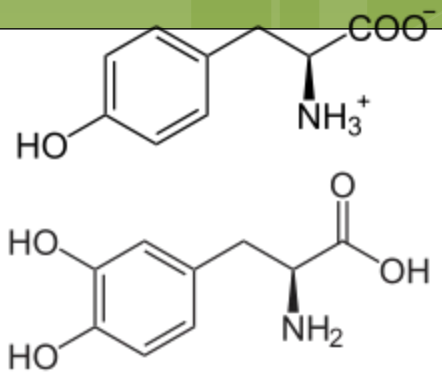
- *Eumelanin (brown & black),*
- *Pheomelanin, and*
- *Neuromelanin*



- *Tyrosinase is a copper-containing enzyme that is present in melanocytes*

Melanin types

- *Eumelanin:*
  - *black eumelanin in the absence of other pigments causes grey hair.*
  - *brown eumelanin in the absence of other pigments causes yellow (blond) color hair.*
- *Pheomelanin:*
  - *imparts a pink to red hue, depending upon its concentration.*
  - *concentrated in the lips, nipples, glans penis, and vagina.*
  - *When small amount of brown eumelanin in hair mixed with red pheomelanin, the result is red hair.*
- *Neuromelanin:*
  - *dark polymer pigment produced in specific populations of catecholaminergic neurons in the brain*



Tyrosine

Tyrosine hydroxylase

DOPA

Tyrosinase

Dopaquinone

Leucodopachrome

Cysteine

5,6-Dihydroxyindole

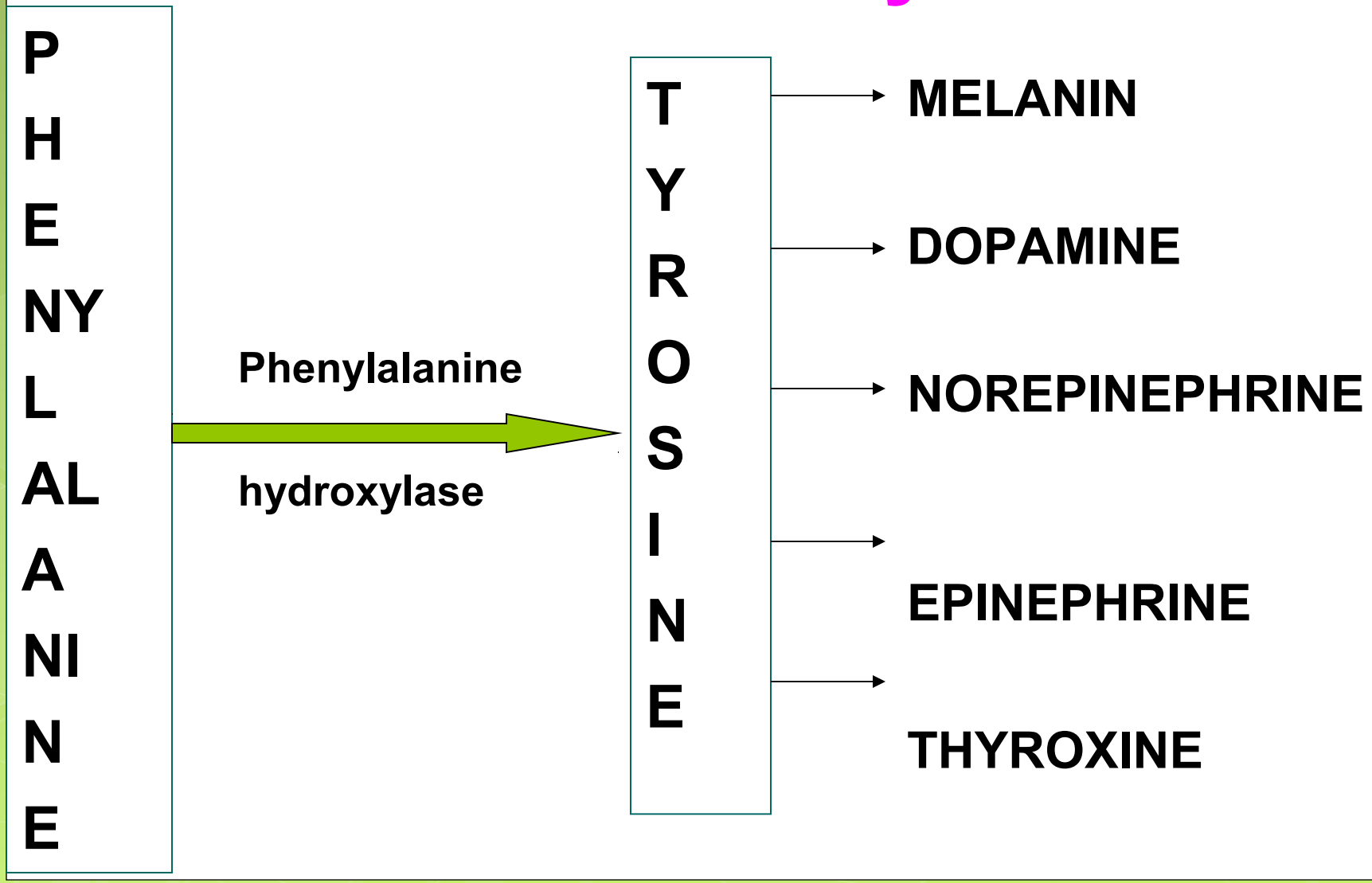
Melanin red polymers

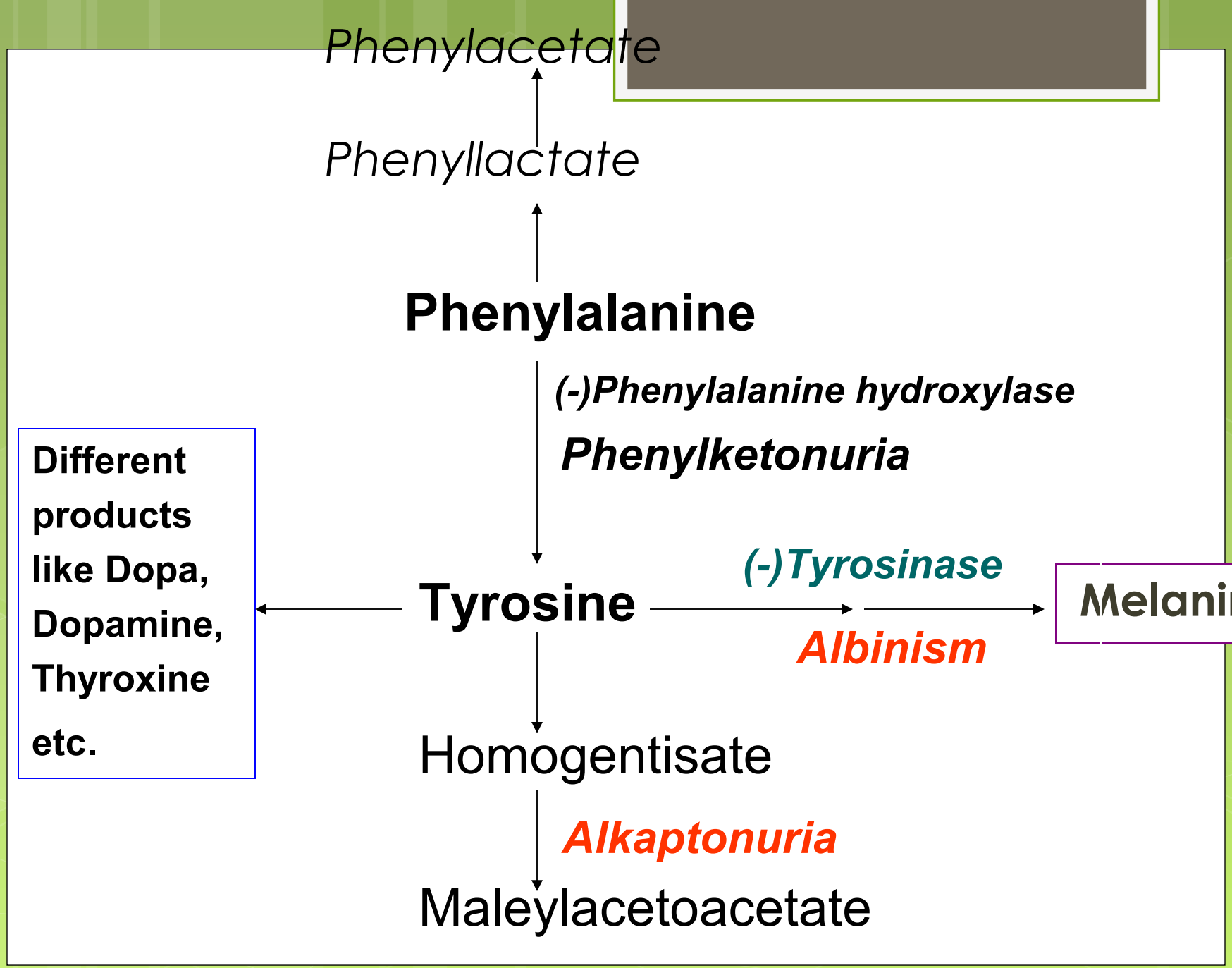
Tyrosinase

Indole 5-6-quinone

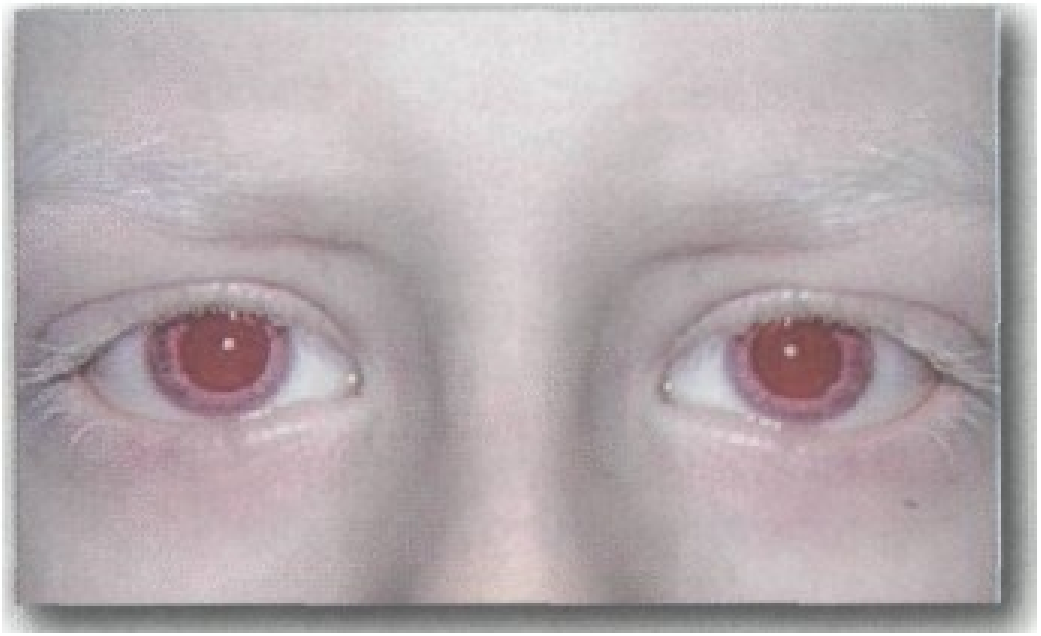
melanochrome → **Melanin black polymers**

Metabolism of Tyrosine



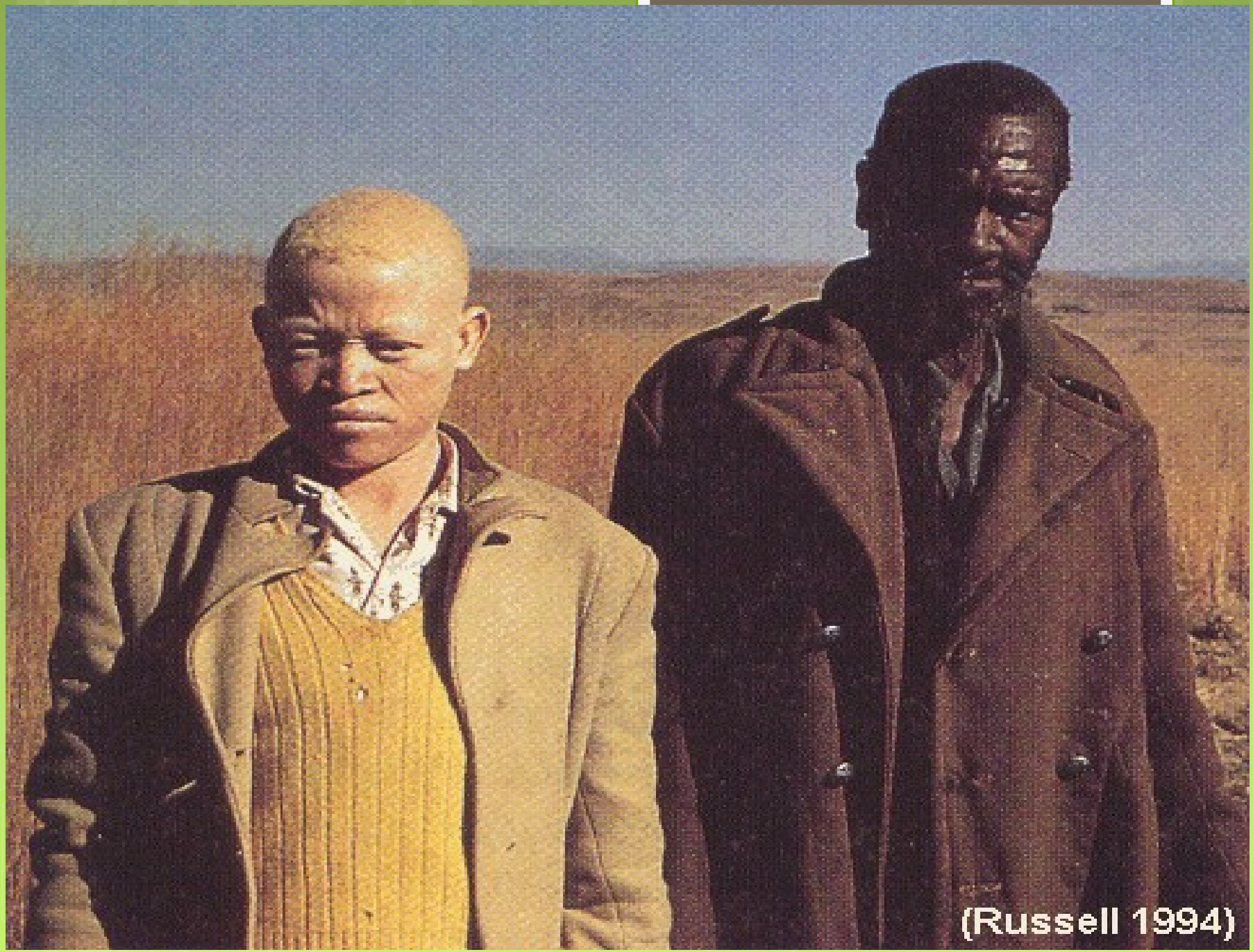

Phenylacetate
Phenyllactate
Phenylalanine
(-)Phenylalanine hydroxylase
Phenylketonuria
Different products like Dopa, Dopamine, Thyroxine etc.
Tyrosine
(-)Tyrosinase
Albinism
Melanin
Homogentisate
Alkaptonuria
Maleylacetoacetate

Albinism



History

Balthazar Telez coined the term “albino”, meaning white Negro, when he saw white African tribe members along the coast of West Africa. He and the other explorers thought they were seeing two different races of people


(Russell 1994)

What is albinism?

- Albinism is a group of genetic conditions that causes a lack of pigment- melanin.
- It can effect only the eyes or both the eyes and skin.
- Most types of albinism are inherited when an individual receives the albinism gene from both parents.
- The exception is one type of ocular albinism, which is passed on from mothers to their sons.

Description

- *Inherited by one of several modes:*
  - *autosomal recessive, autosomal dominant, or Complete albinism*
- *Affected people may appear to have white hair, skin & iris color.*
- *They may have vision defects and photophobia.*
- *Oculocutaneous albinism is most severe form resulting from a deficiency of tyrosinase activity, causing a total absence of pigment from the hair, eyes & skin*

Defects

- *Tyrosinase enzyme deficiency or reduced activity*
  - *Mutation in Tyrosinase related protein (TRP-1) gene*
  - *Recessive inheritance of OCA1 and OCA2*

Symptoms

- *Absence of pigment from the hair, skin, or iris of eyes*
- *Lighter than normal skin and hair or complete albinism*
- *Most forms of complete albinism have some of the following possible symptoms:*
- *Rapid eye movements*
- *Strabismus (eyes not tracking properly)*
- *Photophobia (avoidance of light because of discomfort)*
- *Decreased visual acuity*
- *Functional blindness*

Alternate names

- **Hypopigmentation**
- **Ocular Albinism**
- **Oculocutaneous Albinism**

Treatment

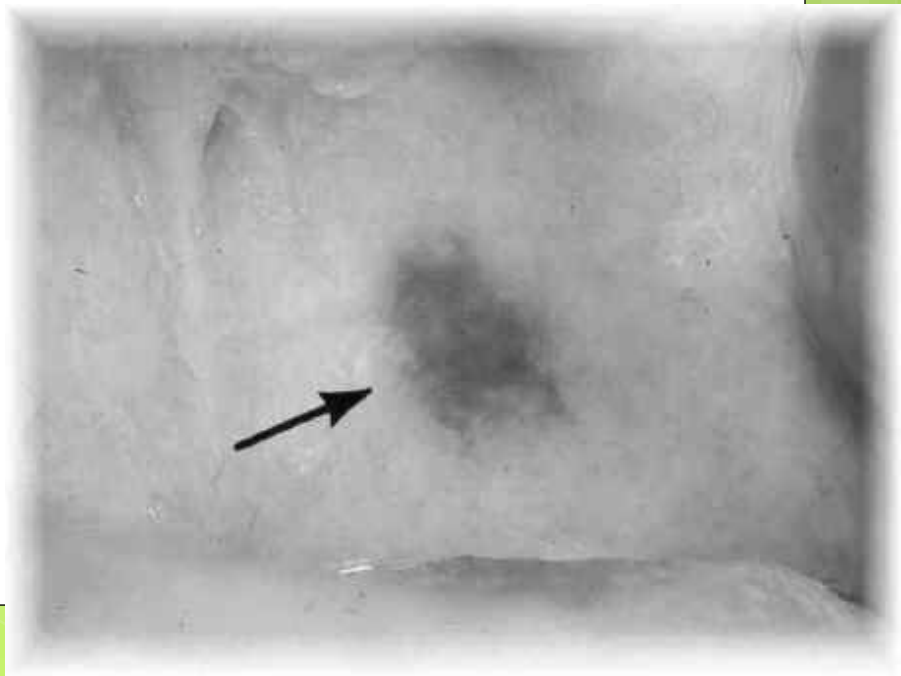
- *The skin and eyes must be protected from the sun. Sunglasses (UV protected) may relieve photophobia.*

Prognosis

- *Albinism does not affect the expected lifespan.*
- *Activities may be limited by intolerance to the sun.*

Melanotic macule

- *The melanotic macule is the mouth equivalent of a freckle or brown pigmented patch of the skin.*
- *One-third of lesions occur on the vermilion border of the lower lip, along with the buccal mucosa, gingiva and palate.*
- **One of every 1,000 adults**



Large brown macule of the soft palate

Causes of melanotic macule

- *Children: racial origin, no treatment required.*
- *Adults: may be smoke-induced, drug-induced, hormone-induced or spontaneous (without cause), and*
  - *Biopsied to rule out malignant melanoma.*
  - *Some inherited diseases show brown pigmentation of the oral membranes.*