DISORDERS OF PIGMENTATION

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SKIN COLOR

- Determined by - melanin
  - haemoglobin
  - carotenoids
- Melanin - major determinant
- Melanin is synthesized by melanocytes within melanosomes and transferred to keratinocytes
- Constitutive skin colour - genetically determined
- Facultative skin colour - induced by sun and hormones

- Human beings come in a glorious spectrum of different colors: light, dark, plain or freckly skin; black, brunette, blond, auburn, and white hair; and eyes that are blue, hazel, green, amber and brown, to name just a few. It's amazing to realize that most of this color is attributed to a single class of pigments: **the melanins.**

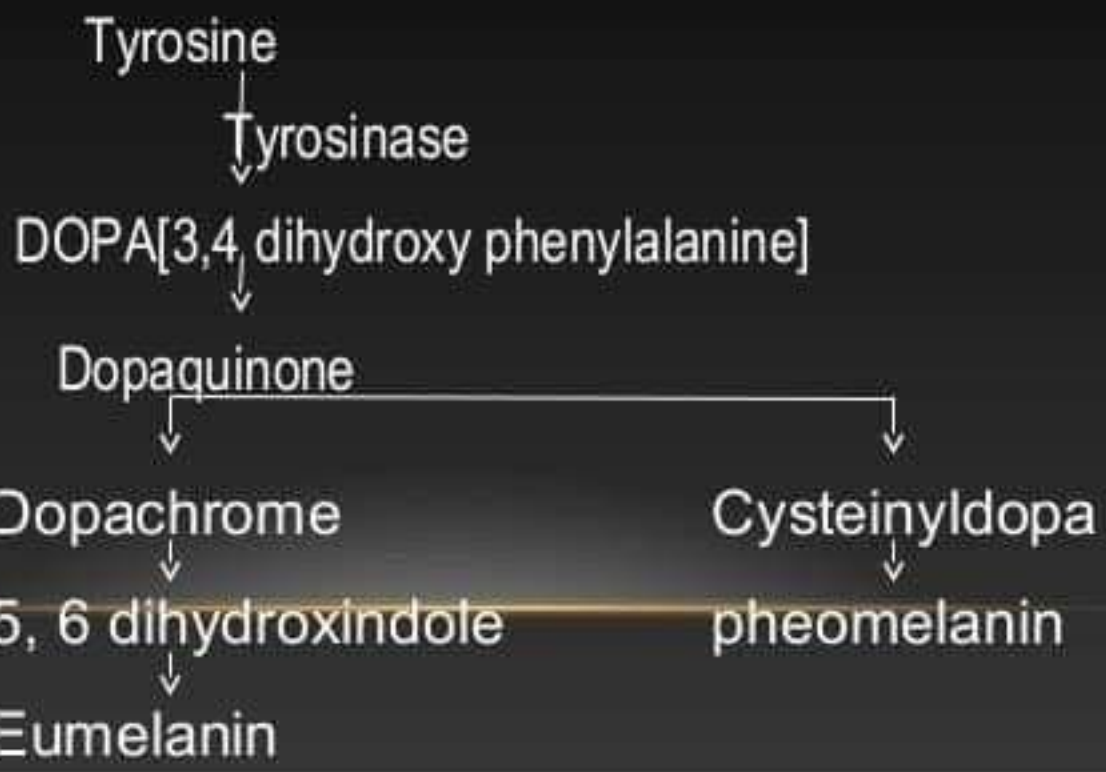
MELANIN

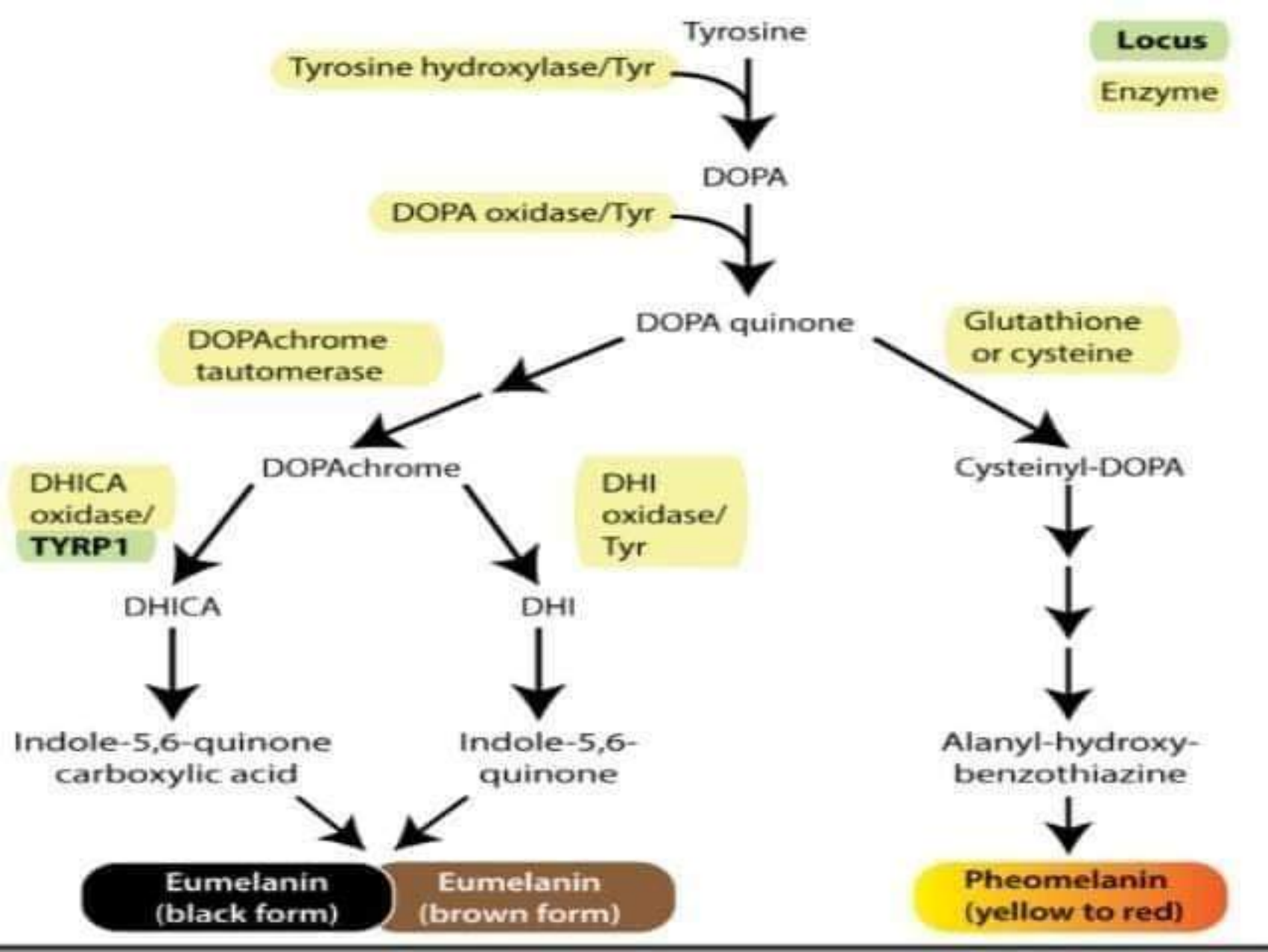
= primary pigment producing brown coloration

- Tyrosine – tyrosinase –melanin- this occurs in the melanosomes of melanocytes
- Then the melanosomes are transferred from the melanocyte to a group of keratinocytes called the epidermal melanin unit
- Variations in skin color is related to the number of melanosomes, the degree of melanization, and the distribution of the epidermal melanin unit

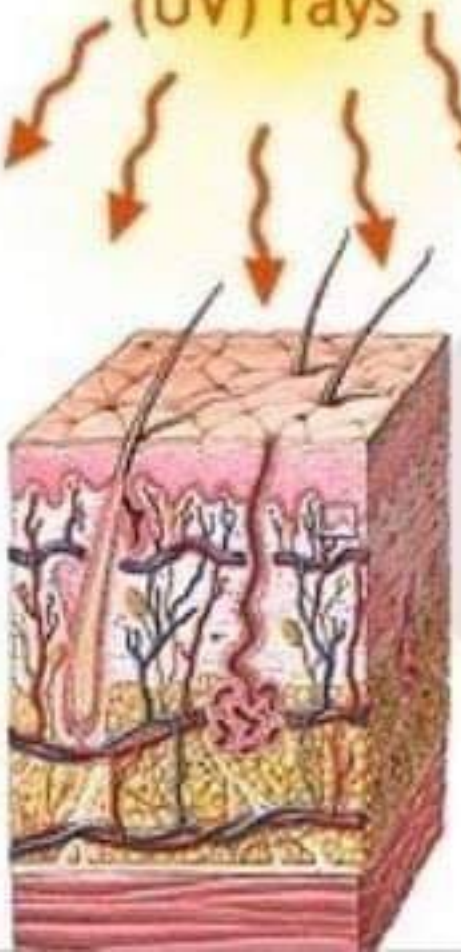
MELANIN

- Two types - eumelanin (black or brown)
  - pheomelanin (reddish)
- Derived from tyrosine

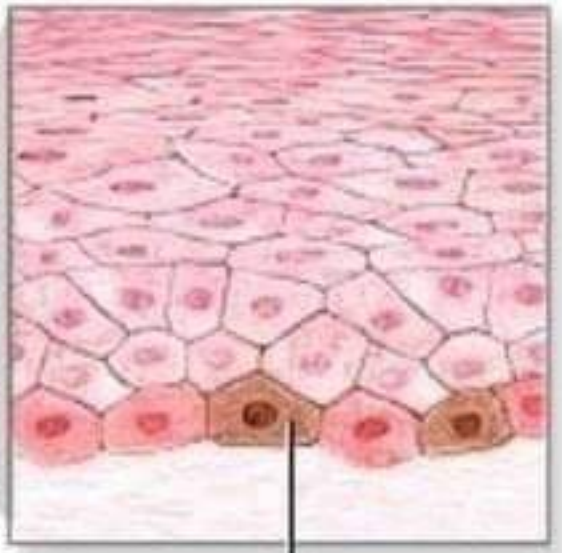



Locus
Enzyme
Tyrosine
Tyrosine hydroxylase/Tyr
DOPA
DOPA oxidase/Tyr
DOPA quinone
DOPAchrome tautomerase
Glutathione or cysteine
DOPAchrome
Cysteinyl-DOPA
DHICA oxidase/ TYRP1
DHI oxidase/ Tyr
DHICA
DHI
Indole-5,6-quinone carboxylic acid
Indole-5,6-quinone
Alanyl-hydroxy-benzothiazine
Eumelanin (black form)
Eumelanin (brown form)
Pheomelanin (yellow to red)

Ultraviolet (UV) rays

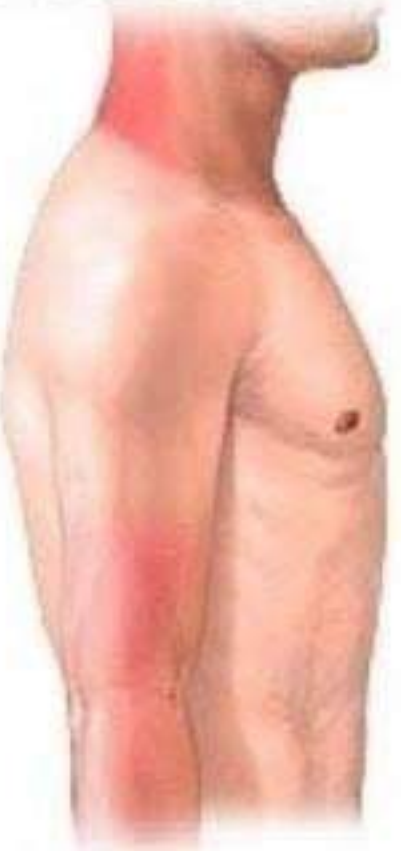


Melanocytes produce melanin to protect the skin from UV rays



Melanocyte

If UV rays exceed what can be blocked by your level of melanin, sunburn results



NORMAL PIGMENTATION

- Normal skin pigmentation is influenced by:
  -the degree of vascularity
  -the amount & location of melanin
  -the presence of carotene
  -the thickness of the horny layer

MELANIN PRODUCTION

- The amount produced is dependent on:

-genetics

-the amount and the wavelengths of ultraviolet light received

-the amount of melanocyte-stimulating hormone (MSH) secreted

- the effect of melanoccyte stimulatingg chemicals like furocoumarins (psoralens)

APPROACH TO A PATIENT

History

- Onset : birth, infancy or later
- Cause: sun exposure, drugs, occupation
- Systemic complaints
- Family history: neurofibromatosis, tuberous sclerosis, vitiligo

Examination:

- Type of lesion: brown, blue, hypopigmented (check sensation), depigmented
- Shape: Ash leaf macules (tuberous slerosis)

  Koebner phenomenon(vitiligo)
- Distribution pattern : linear/segmental (nevus depigmentosus), symmetric (vitiligo), specific sites (melasma, Addison's disease)

Examination aids:

- Hand lens
- Oblique lighting for elevation or depression
- Dermatoscopy
- Wood's lamp - 360 nm. Epidermal pigmentary anomalies made more prominent
- Histology- H and E for presence or absence of melanin

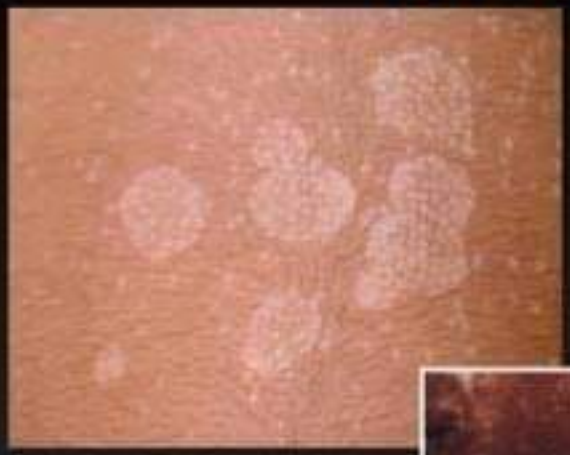
Dopa reaction - melanocytes stain dark

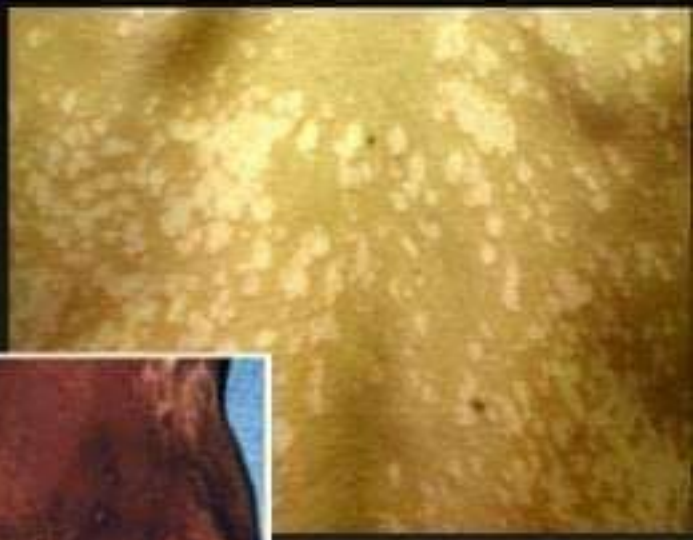
Silver stains - melanin stains black

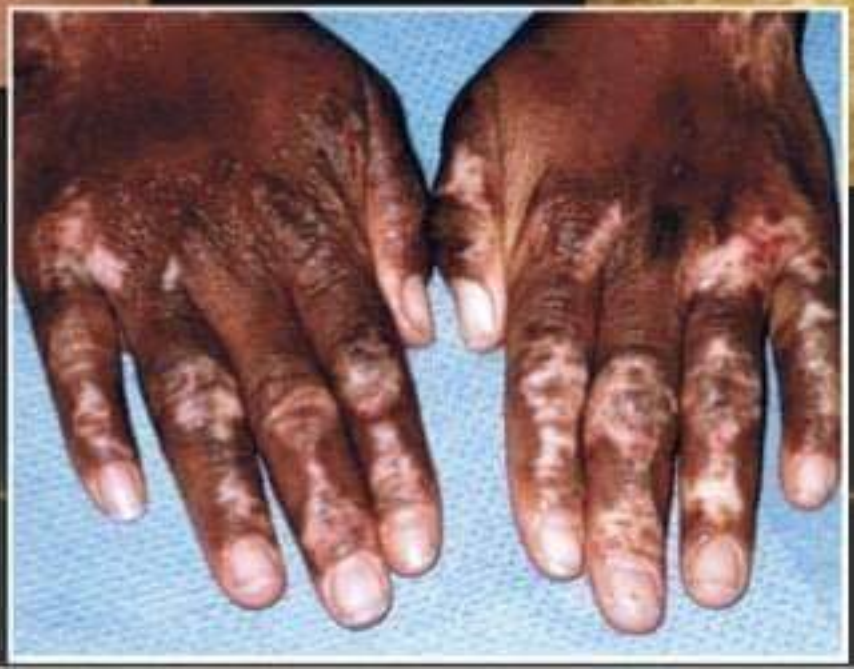
CLASSIFICATION

- GENERALIZED
- LOCALISED
- HYPERPIGMENTATION
- HYPOPIGMENTATION
- DEPIGMENTATION
- CONGENITAL
- ACQUIRED

HYPOPIGMENTATION







CLASSIFICATION

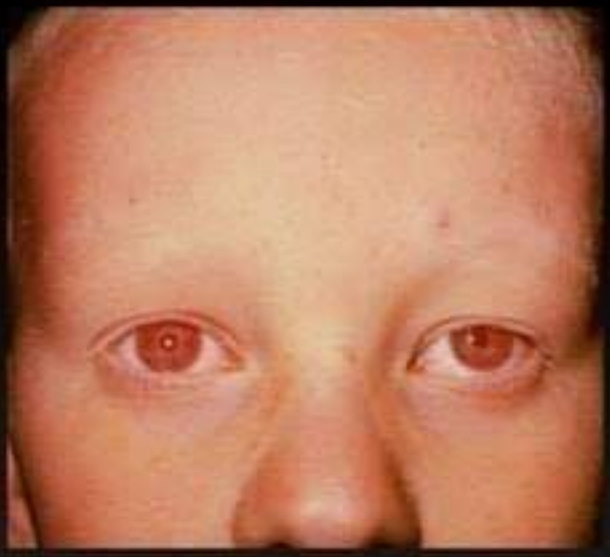
- Genetic and Developmental:
  - *Albinism*, Nevus depigmentosus, Nevus anaemicus, Halo nevus, *Tuberous sclerosis* (ash leaf macule)
- Endocrine:
  - Addison's disease, Hypothyroidism, Hypopituitarism
- Nutritional:
  - Vit.B12 deficiency, *Kwashiorkor*, Malabsorption

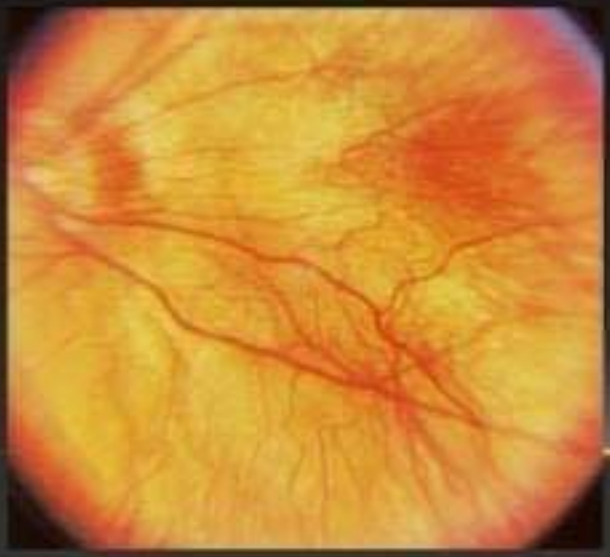
- Post-inflammatory:
  - *Pityriasis alba*, Eczema, Psoriasis, Pityriasis rosea, Lupus erythematosus, Morphea, Scleroderma, Bullous dermatoses
- Infection:
  - *Leprosy, Tinea versicolor*, Candidiasis, Post kala azar dermal leishmaniasis
- Chemicals and Drugs:

- Physical:
  - Burns, Trauma, Post dermabrasion, Post laser
- Miscellaneous:
  - *Idiopathic guttate hypomelanosis*, *Vitiligo*, Mycosis fungoides

ALBINISM

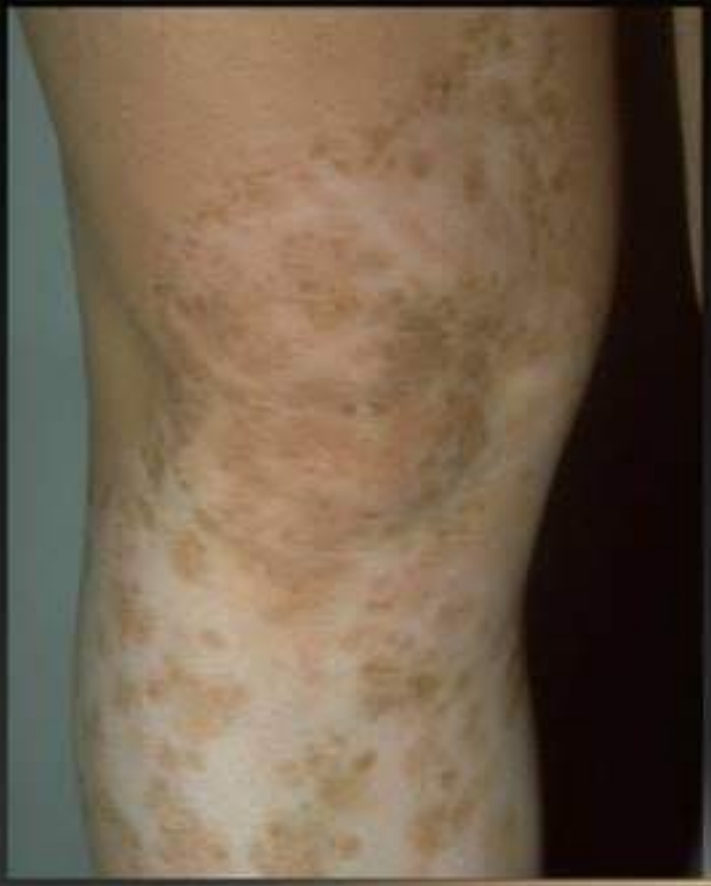
- Oculocutaneous albinism involves skin, hair and eyes or the eyes alone (ocular albinism)
- Mostly autosomal recessive
- Absence of pigmentation from birth
- Photophobia, reduced visual activity, nystagmus, pale irides that transilluminate, hypopigmented fundi, hypoplastic foveae, and lack of stereopsis
- Sunburns, skin cancers common
- Protection of eyes and skin by sunglasses, sunscreens SPF > 20, clothing



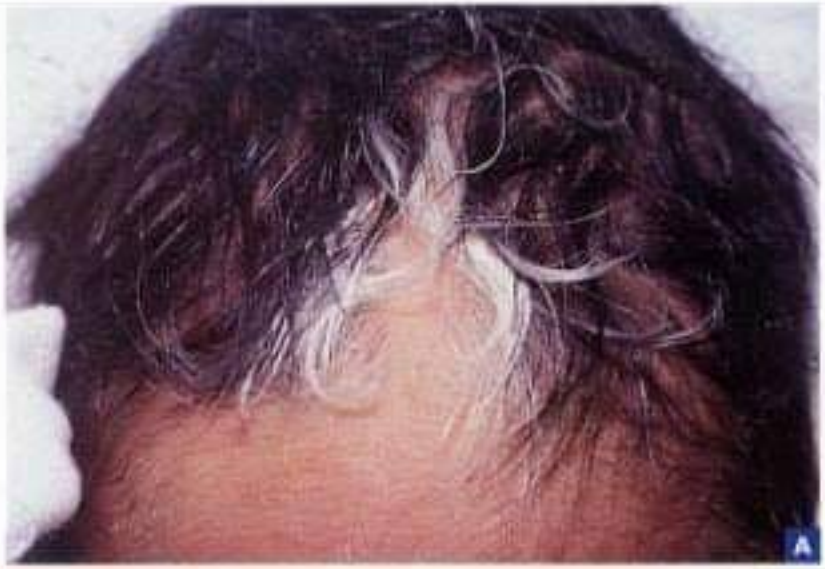


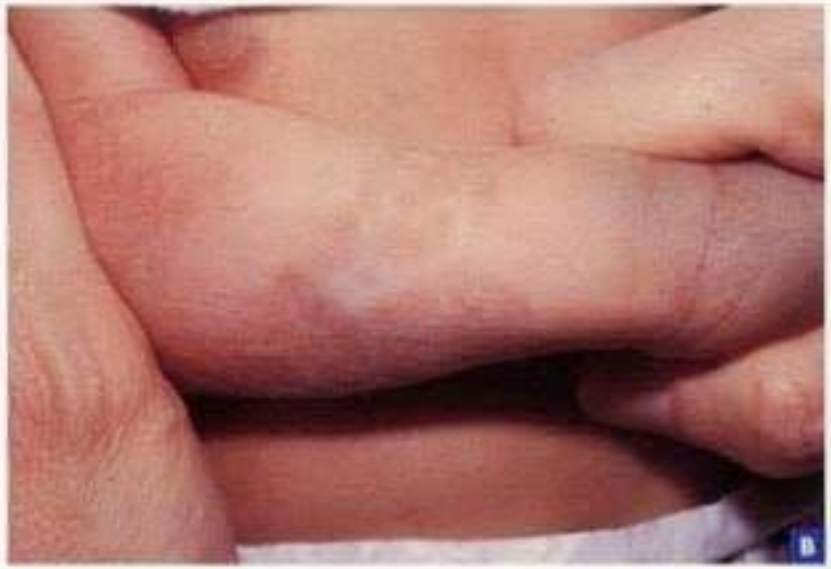
- **Top:** albinism with white hair, pale skin, and translucent irides
- **Bottom:** ophthalmoscopic view of a patient with albinism demonstrates a pale fundus, poor macular development, and prominent choroidal vasculature

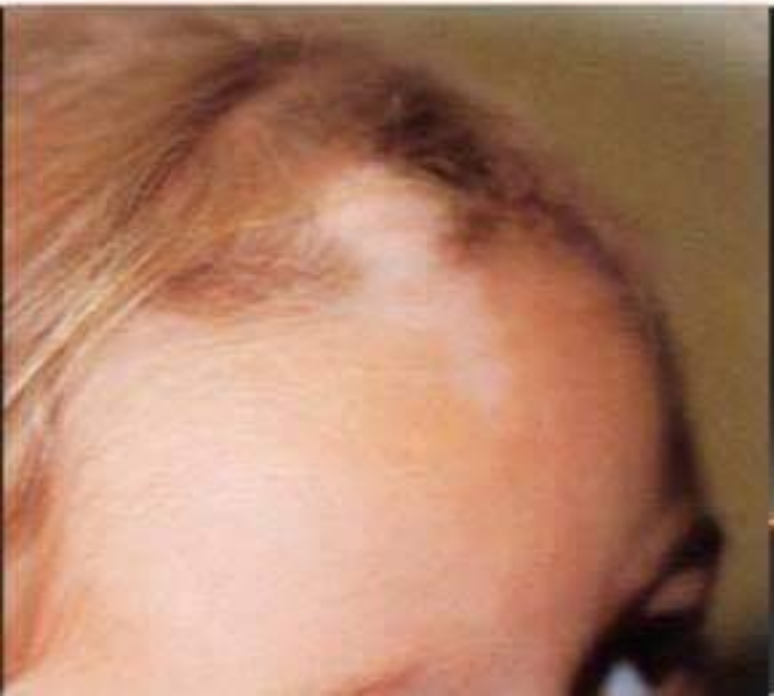
PIEBALDISM



- Rare, AD with variable phenotype, *presenting at birth*
- White forelock, patchy absence of skin pigmentation
- Depigmented lesions are static and occur on the anterior and posterior trunk, mid upper arm to wrist, mid-thigh to mid-calf, and shins
- A characteristic feature is the presence of *hyper pigmented macules within the areas of lack of pigmentation and on normal skin*







- White forelock and patch of unpigmented skin in a young girl with piebaldism

WAARDENBURG'S SYNDROME

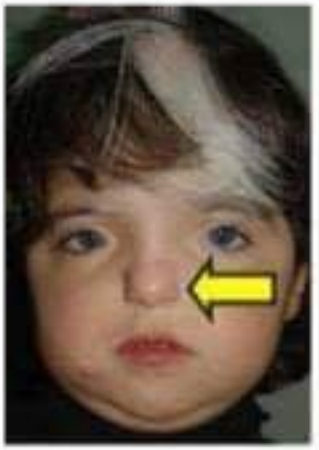
- Rare, autosomal dominant disorder
- White forelock
- Hypertelorism
- Congenital deafness
- Hypomelanotic macules
- Heterochromic irides
- Incomplete forms may occur

Waardenburg Syndrome

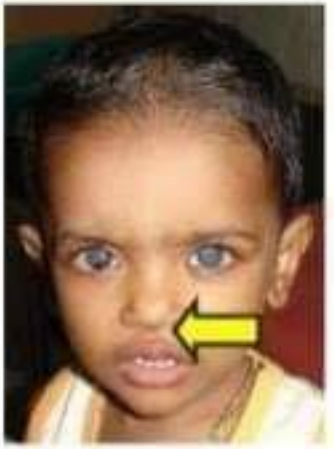
Features

(courtesy of Dr. Hutchinson)

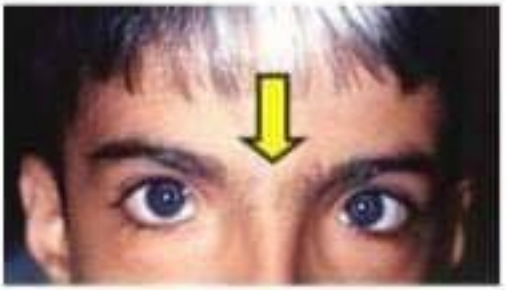
White forelock



Hypoplastic alae



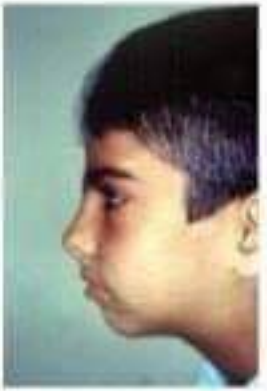
Short philtrum



Synophyrs

Heterochromia irides

Dystopia canthorum
Isohypochromia irides

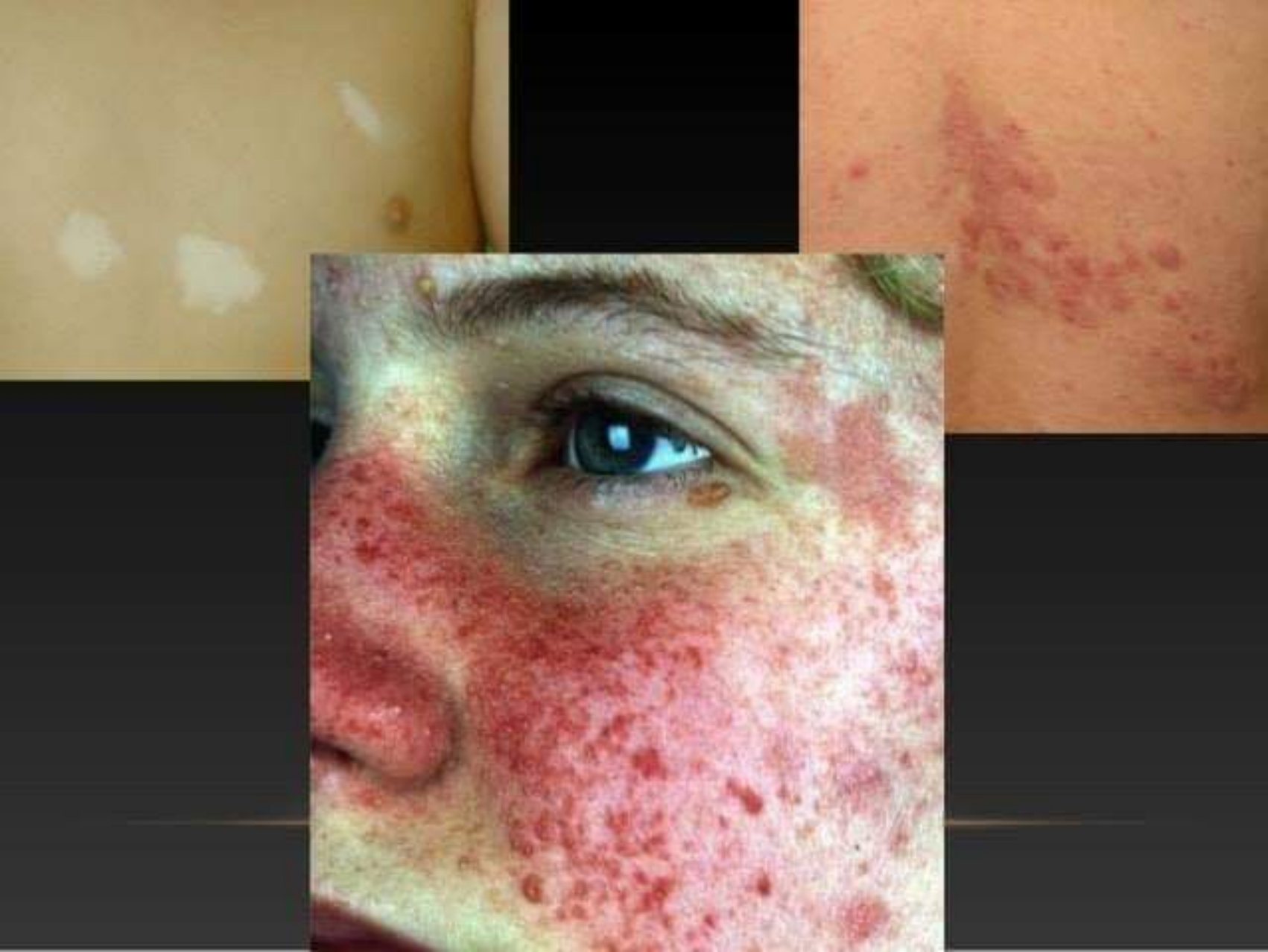


Flat nasal root

(Schwartz, 2010)

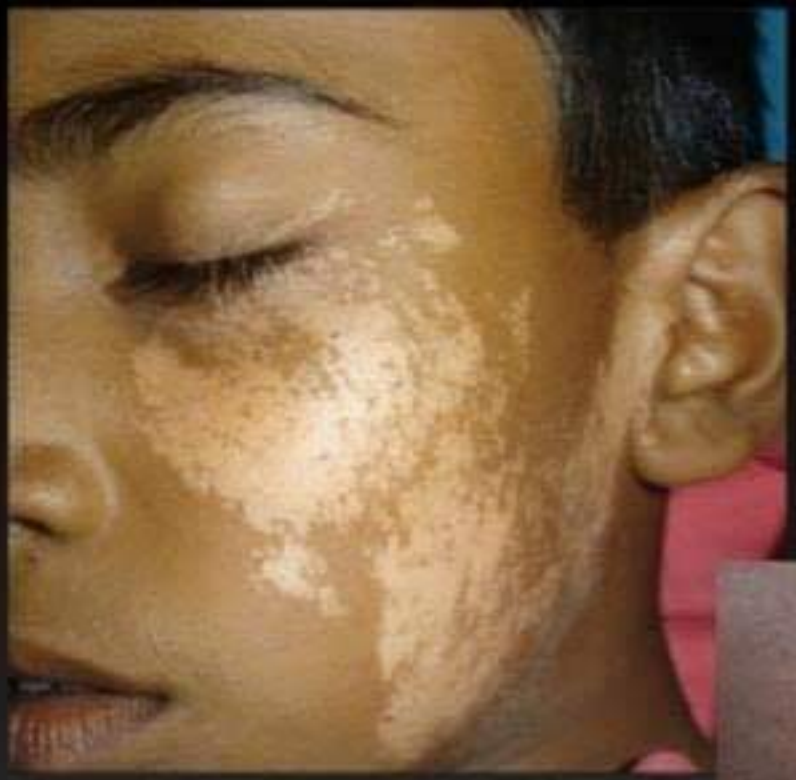
TUBEROUS SCLEROSIS

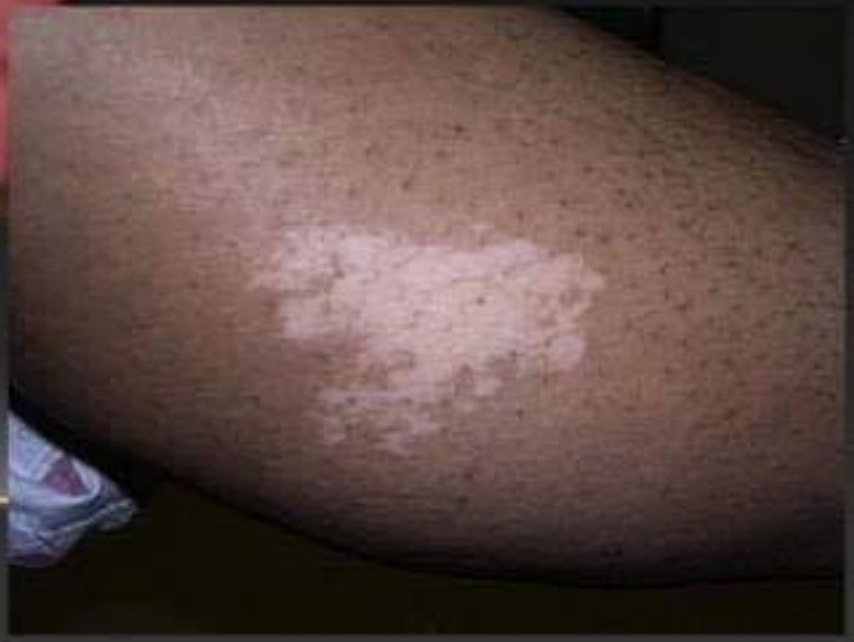
- Autosominal dominant, neurocutaneous syndrome with skin lesions, mental retardation and epilepsy
- Skin lesions are ash-leaf macules, angiofibromas and shagreen patches
- Ash-leaf macules - present at birth in > 90% cases, so important in early diagnosis
  Oval or ash-leaf shaped, hypopigmented macules, made prominent in Wood's lamp
- Long axis is axial on limbs and transverse on trunk



NEVUS DEPIGMENTOSUS

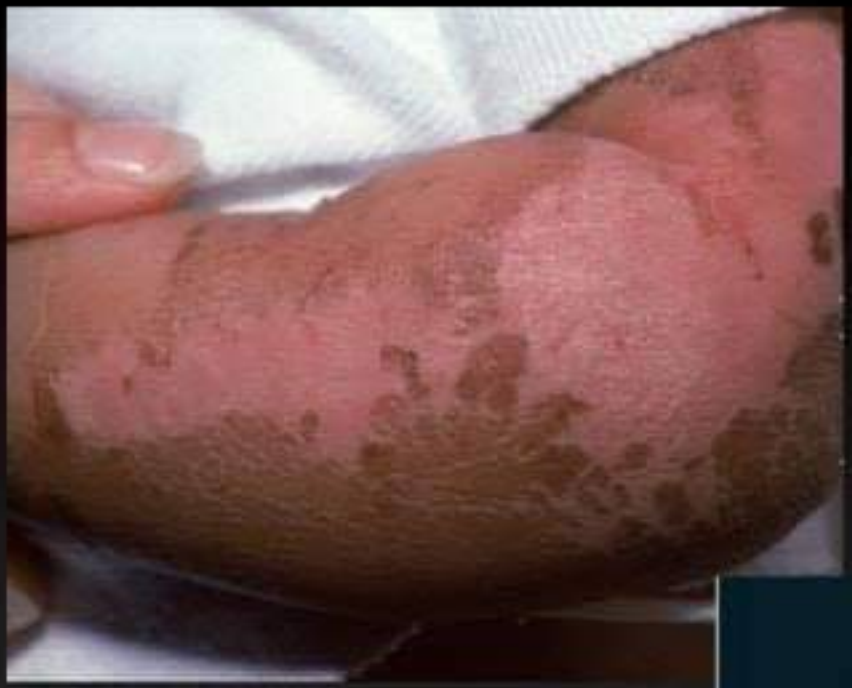
- A hypo pigmented birthmark which is congenital and stable
- Irregular, geographic margins and quasidermatomal distribution
- Block in transfer of melanosomes from melanocytes to keratinocytes
- Sporadic occurrence, no medical significance and no treatment required

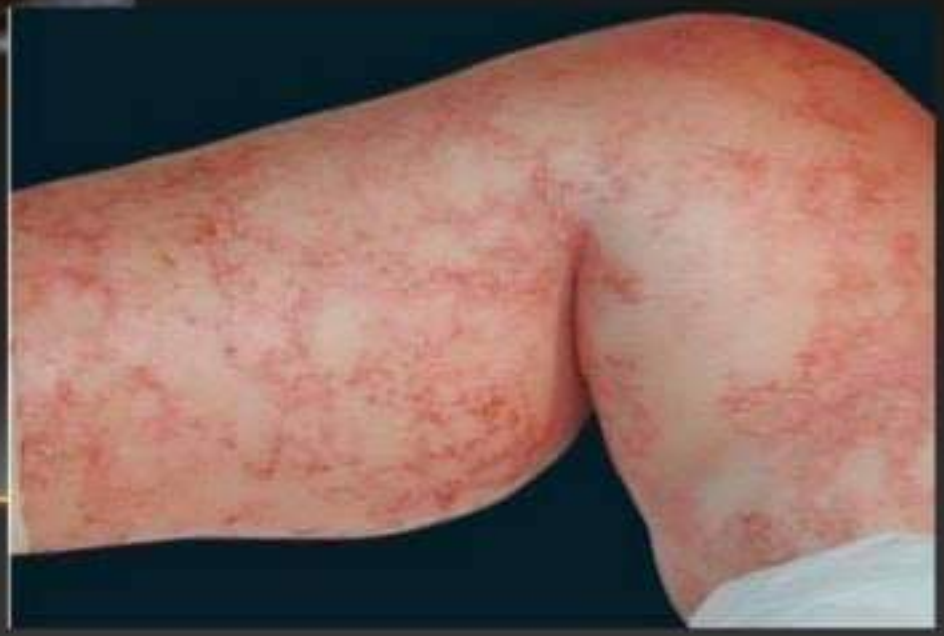




KWASHIORKOR

- Protein deficiency in post weaning years
- Reddish patches which turn into dark plaques which turn white after exfoliation (crazy pavement dermatosis)
- Disruption of melanogenesis is due to multiple deficiencies
- Pigment changes and dyschromic hair are reversible with proper diet





LEUKODERMA

- Postinflammatory leukoderma may result from inflammatory dermatoses ie:
  - Pityriasis rosea, psoriasis, herpes zoster, secondary syphilis, and morphea, sarcoidosis, tinea versicolor, mycosis fungoides, scleroderma, and pityriasis lichenoides chronica, and leprosy
  - Other causes: burns, scars, postdermabrasion, and intralesioal steroid injections