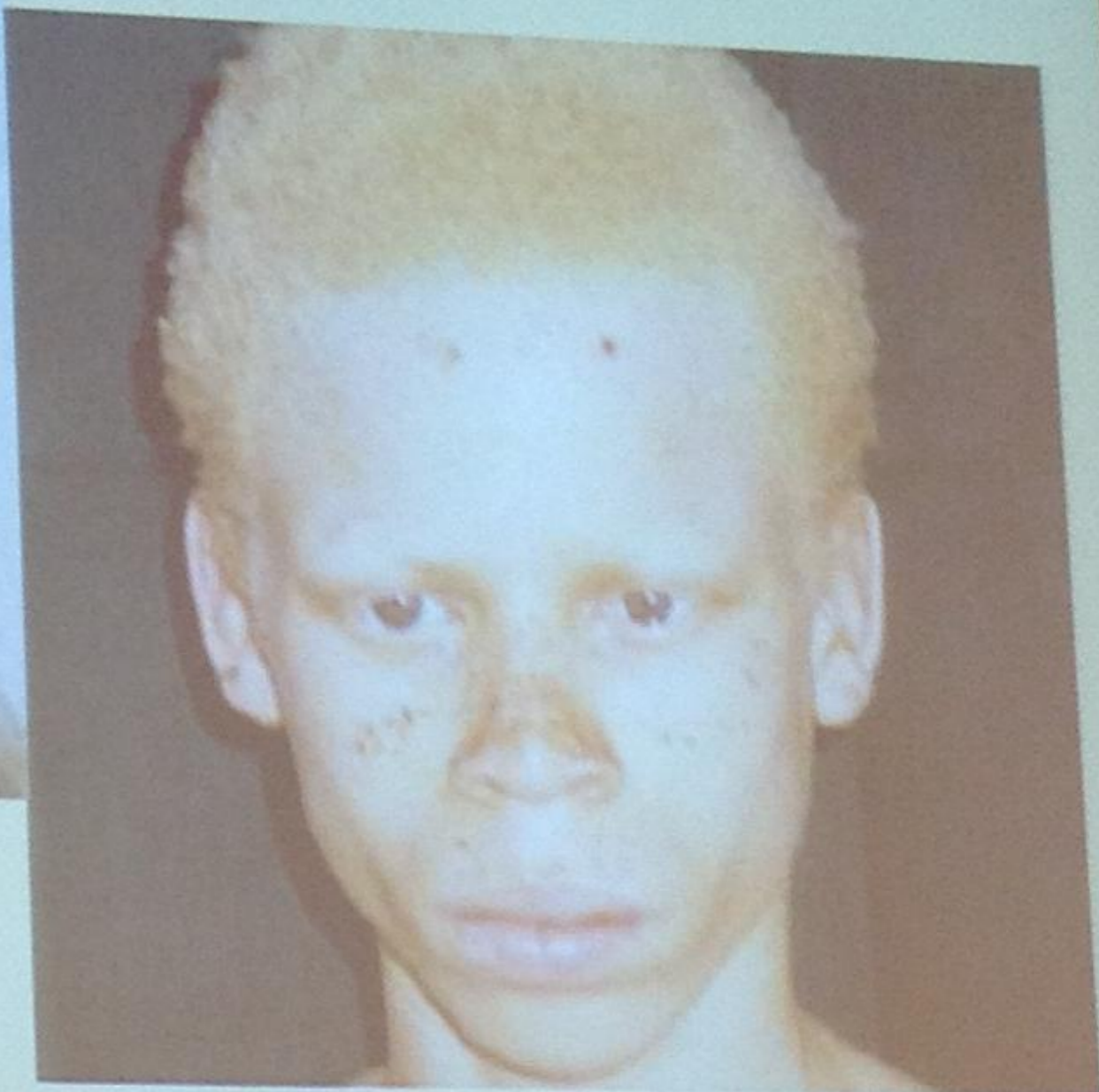


# Disorders Of Melanin

## Hypopigmentation

Generalised	Focal
Oculocutaneous albinism	Piebaldism
Chediak–Higashi syndrome	Localised albinism
Panhypopituitarism	Vitiligo
	Chemical Leukoderma

# Occulocutaneous albinism



# OCA

- A group of congenital inherited disorders (autosomal recessive) characterised by partial or complete absence of melanin in the skin, hair and eyes.
- 2 main types – tyrosinase negative and tyrosinase positive.

# OCA (TYROSINASE NEGATIVE)

- Lack pigment completely – lack of tyrosinase enzyme.
- Skin pink and pale throughout life.
- Hair white.
- Severe nystagmus.

# OCA (TYROSINASE POSITIVE)

- Defective tyrosinase
- Patients develop some pigmentation as they grow older
- Skin – yellowish to brownish
- Hair – yellow/reddish
- Freckles, solar lentigines
- Photophobia, poor visual acuity (poor sight), nystagmus and strabismus

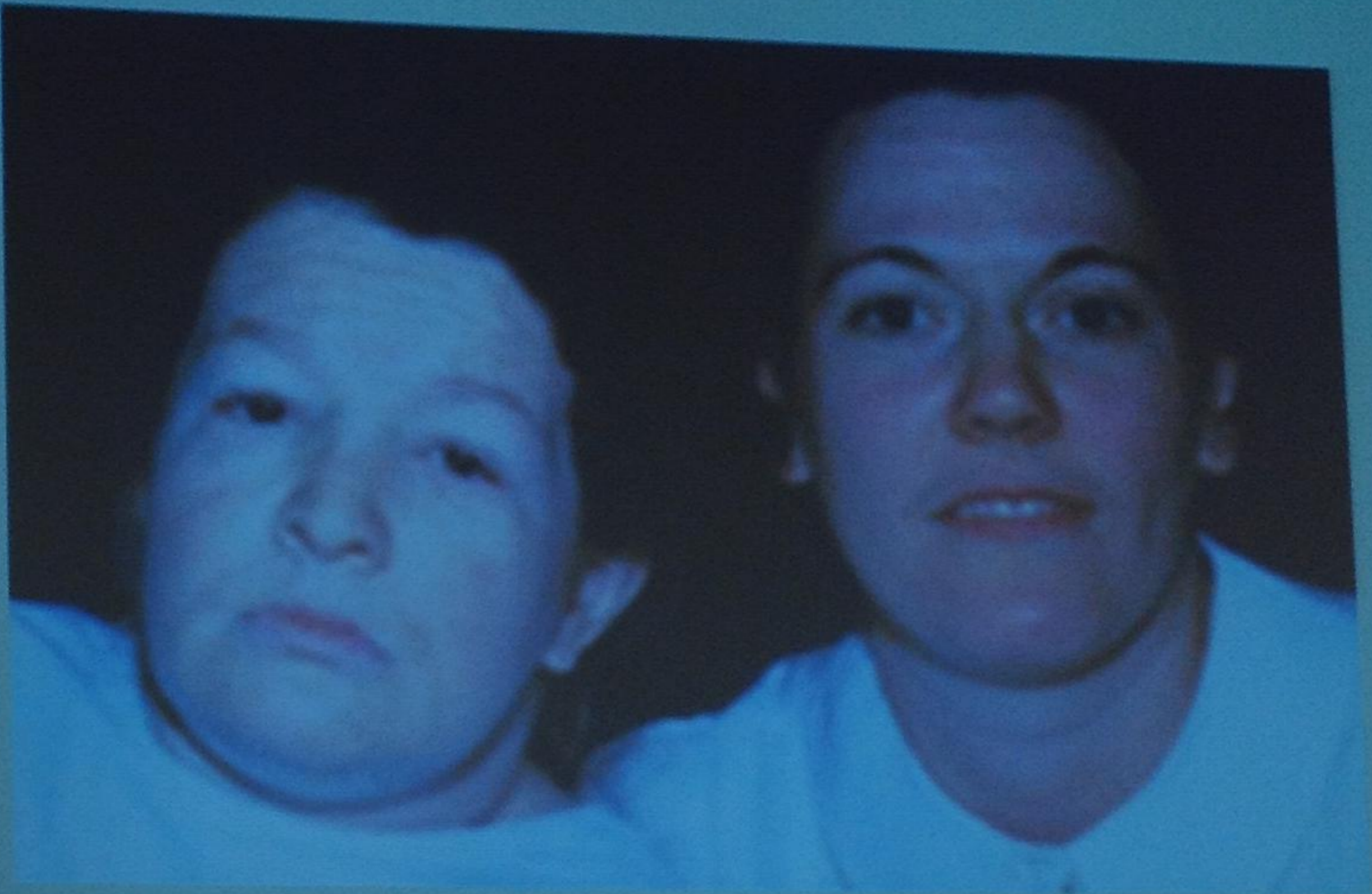
# COMPLICATIONS

- Skin and eyes photosensitive
- Frequent and early solar keratoses.
- Early and frequent squamous cell carcinomas
- Melanomas more common and often amelanotic

# MANAGEMENT

- Avoidance – sun exposure
- Photoprotection – clothing, hats, high SPF(UVB/UVA) sunscreens
- Regular follow up – screening suspicious lesions - early detection and treatment of malignancies

# Panhypopituitarism



**Generalised pallor**



# Disorders Of Melanin

## Hypopigmentation

### Generalised

### Focal

Oculocutaneous albinism

Piebaldism

Chediak–Higashi syndrome

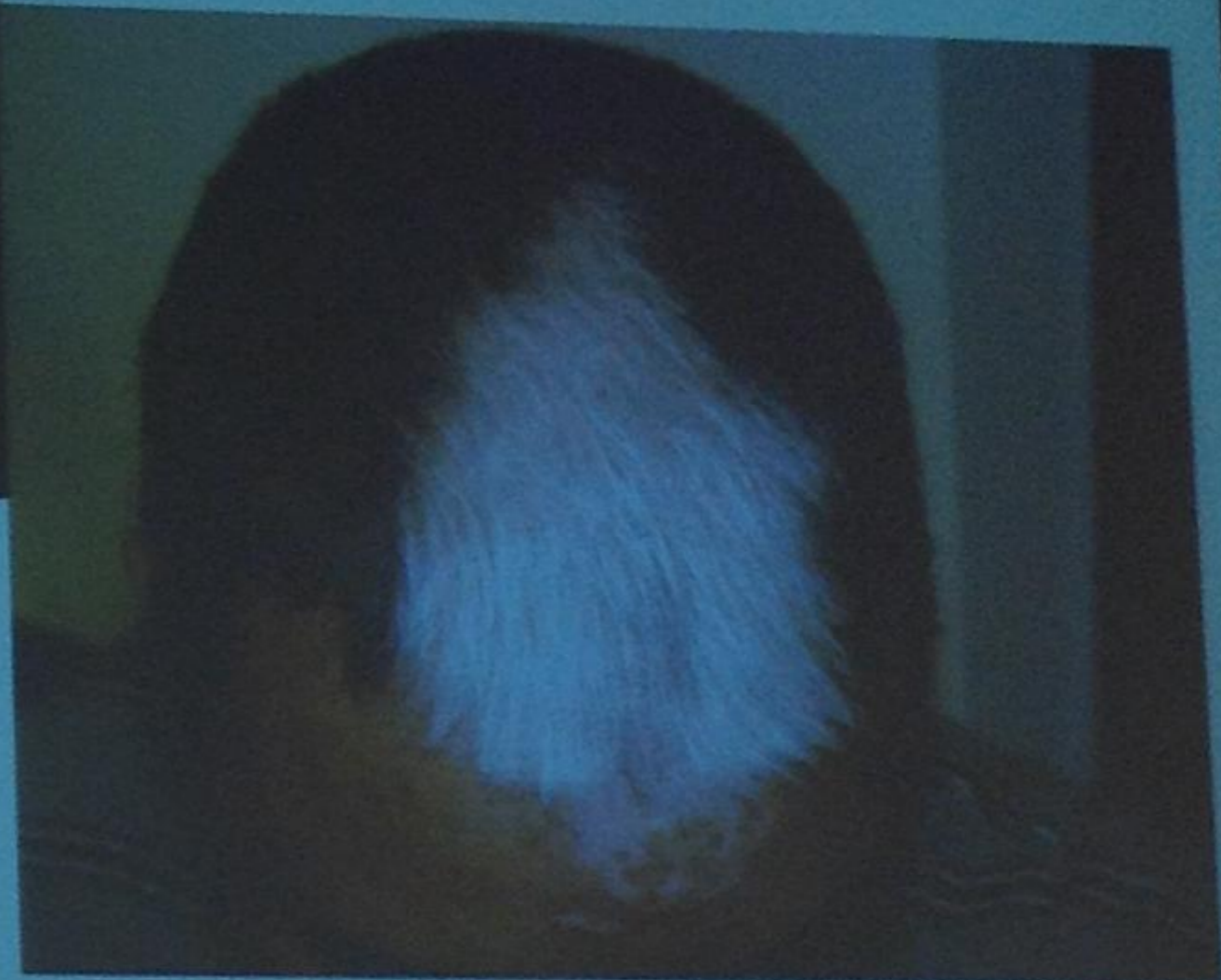
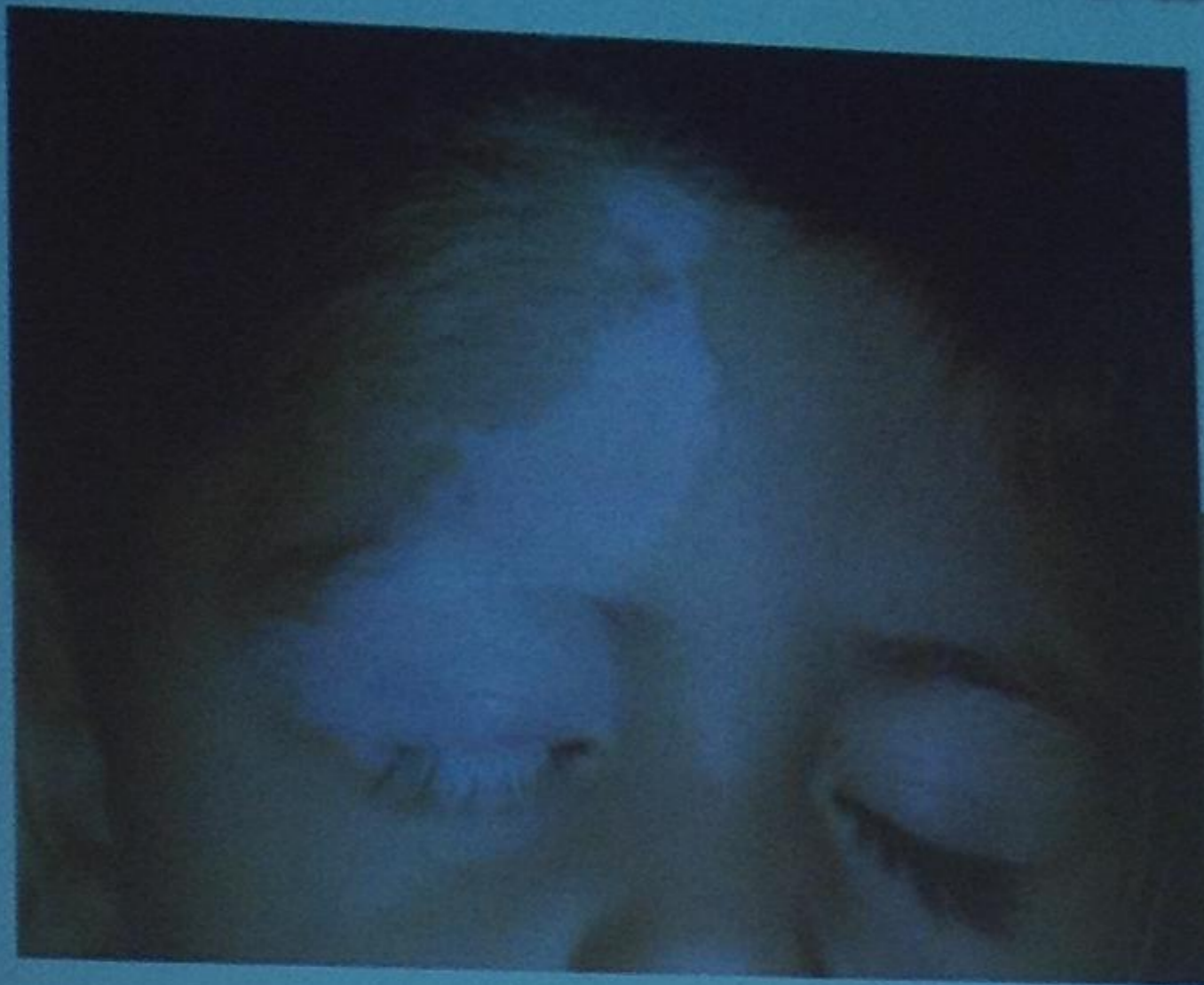
Localised albinism

Panhypopituitarism

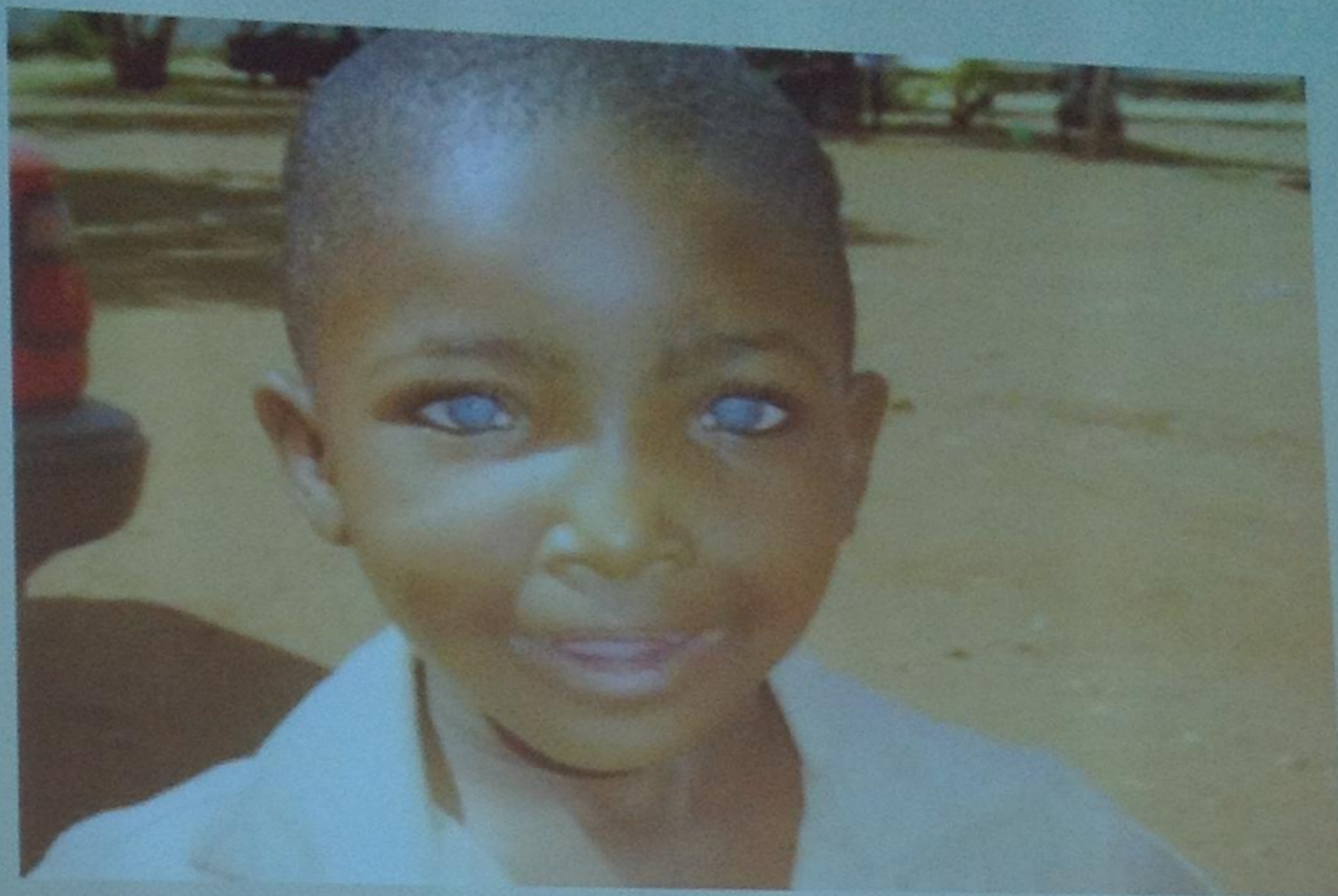
Vitiligo

Chemical Leukoderma

# Piebaldism

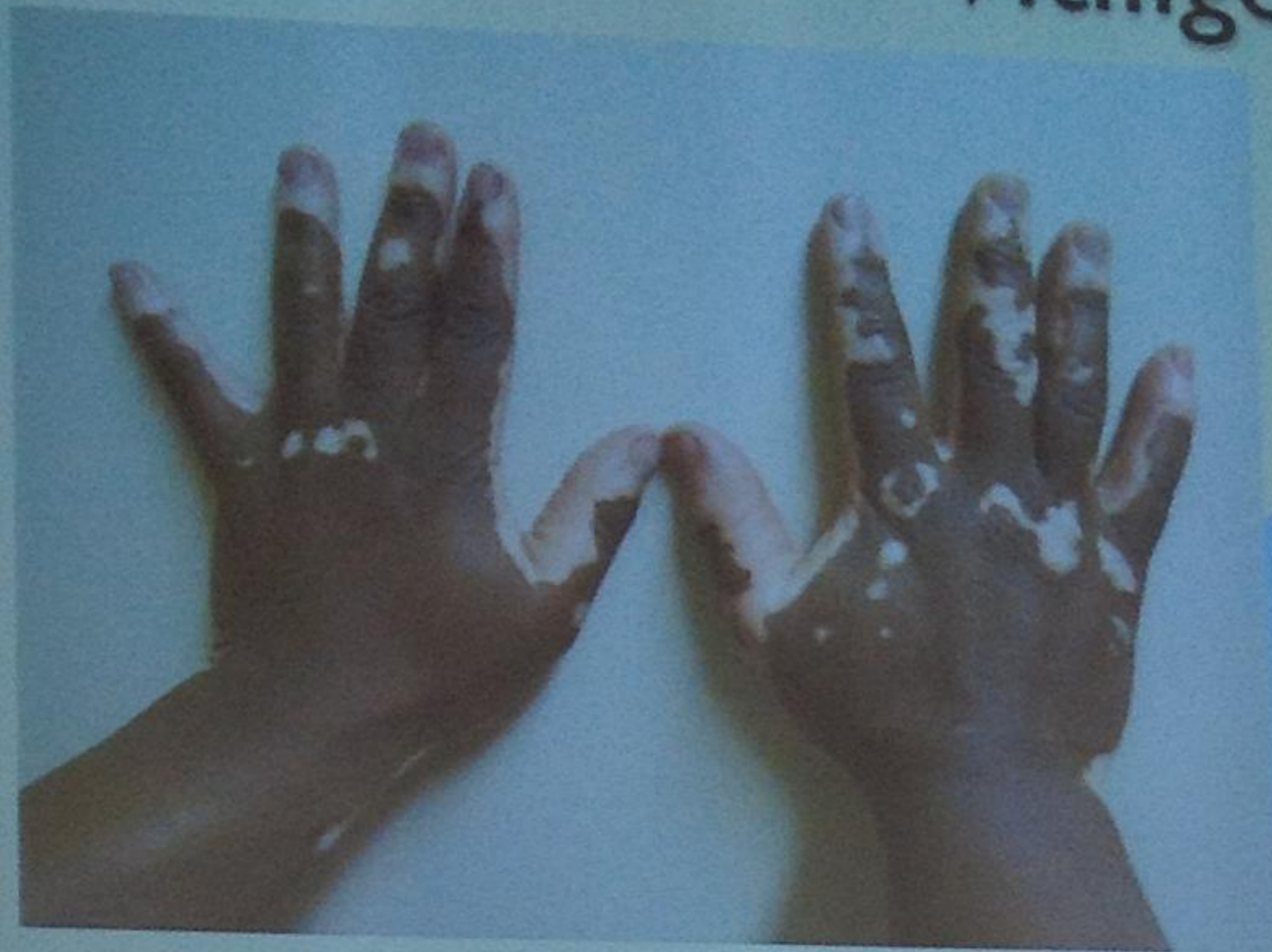


# Localised albinism



Waardenburg Syndrome

# Vitiligo



# VITILIGO

- An acquired, slowly progressive skin disorder, characterized by circumscribed areas of depigmentation due to destruction of melanocytes

# VITILIGO

- Autoimmunity the most accepted theory  
– common association with other autoimmune disorders, viz; Hashimoto's thyroiditis, Grave's disease, alopecia areata, pernicious anaemia, insulin dependent diabetes mellitus and Addison's disease

# CLINICAL PRESENTATION

- Depigmented macules without inflammation and scaling - often symmetrical.
- Main presentations :
  - localized/acral/periorificial, segmental, generalized and universal
  - Periorificial - around eyes, mouth, nose, ears, areolar, genitalia, anus and umbilicus
  - Acral -Hands, feet and also elbows/knees (friction areas)
  - Generalized - widespread involvement
  - Universal - entire body involved

# CLINICAL PRESENTATION

- Cosmetic appearance may cause great deal of distress and psychosocial problems
- Vitiligo lesions sunburn easily (no melanin)
- Repigmentation starts around hair follicles – spotted pigmentation. Spots slowly coalesce, cover lesions



# DIFFERENTIAL DIAGNOSIS

- Post inflammatory hypopigmentation.
- Pityriasis alba
- Pityriasis versicolor
- Leprosy
- Sarcoidosis
- Naevi – anaemicus and depigmentosa.
- Burn scar

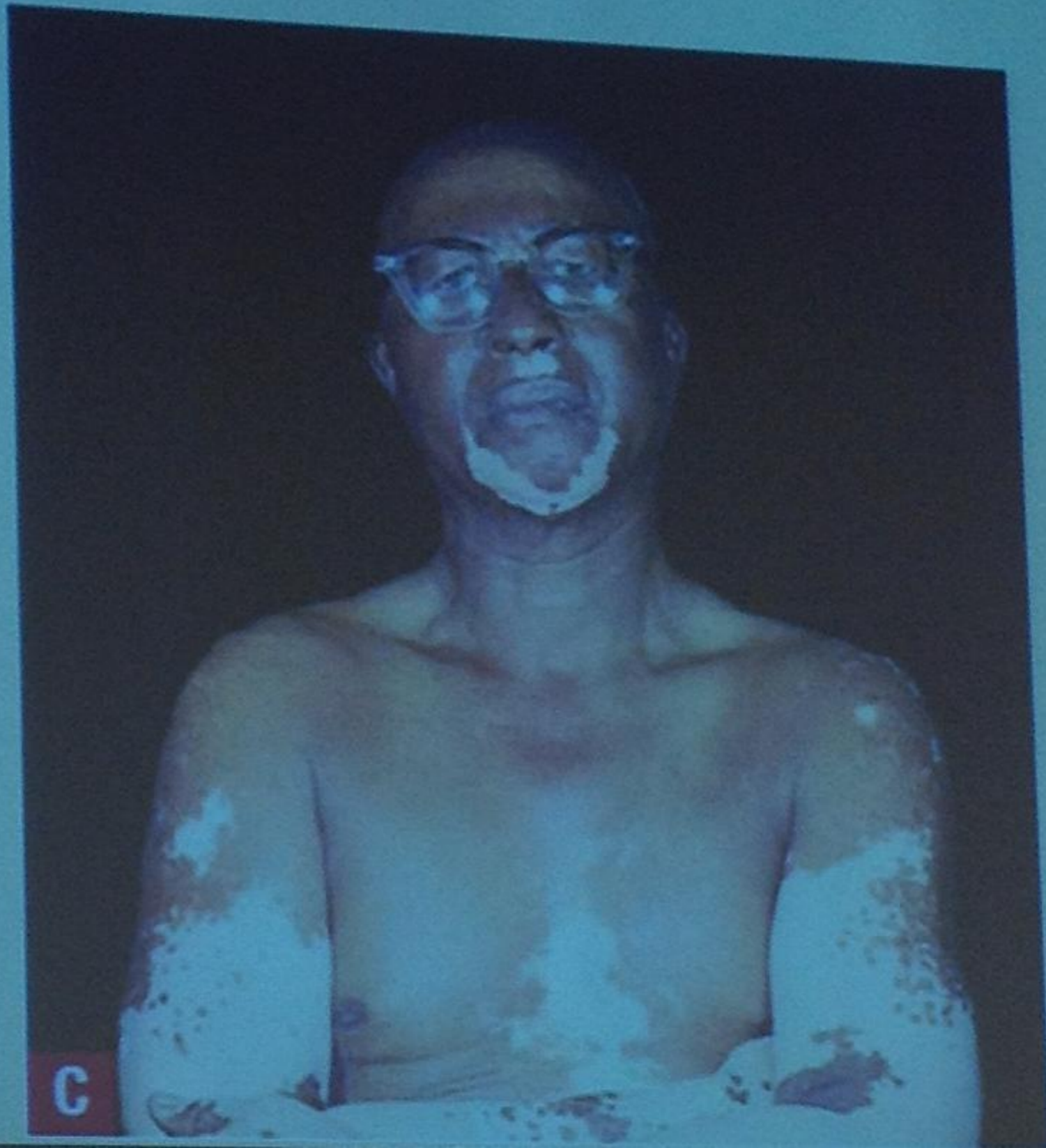
# MANAGEMENT

- Treatment generally unsatisfactory.
- Sun protection
- Cosmetic camouflage - coverderm, dermablend make-up
- Topical - potent corticosteroids, vit D analogue, calcineurin inhibitors
- PUVA, narrow band UVB, excimer laser.
- Surgical - melanocyte grafting

# PROGNOSIS

- Course unpredictable, with spontaneous repigmentation in sun-exposed areas +/- 10 - 20%
- Response to different treatment modalities and between individuals differs

# Chemical Leukoderma



# Pigments Related To Melanin

## ■ Ochronosis

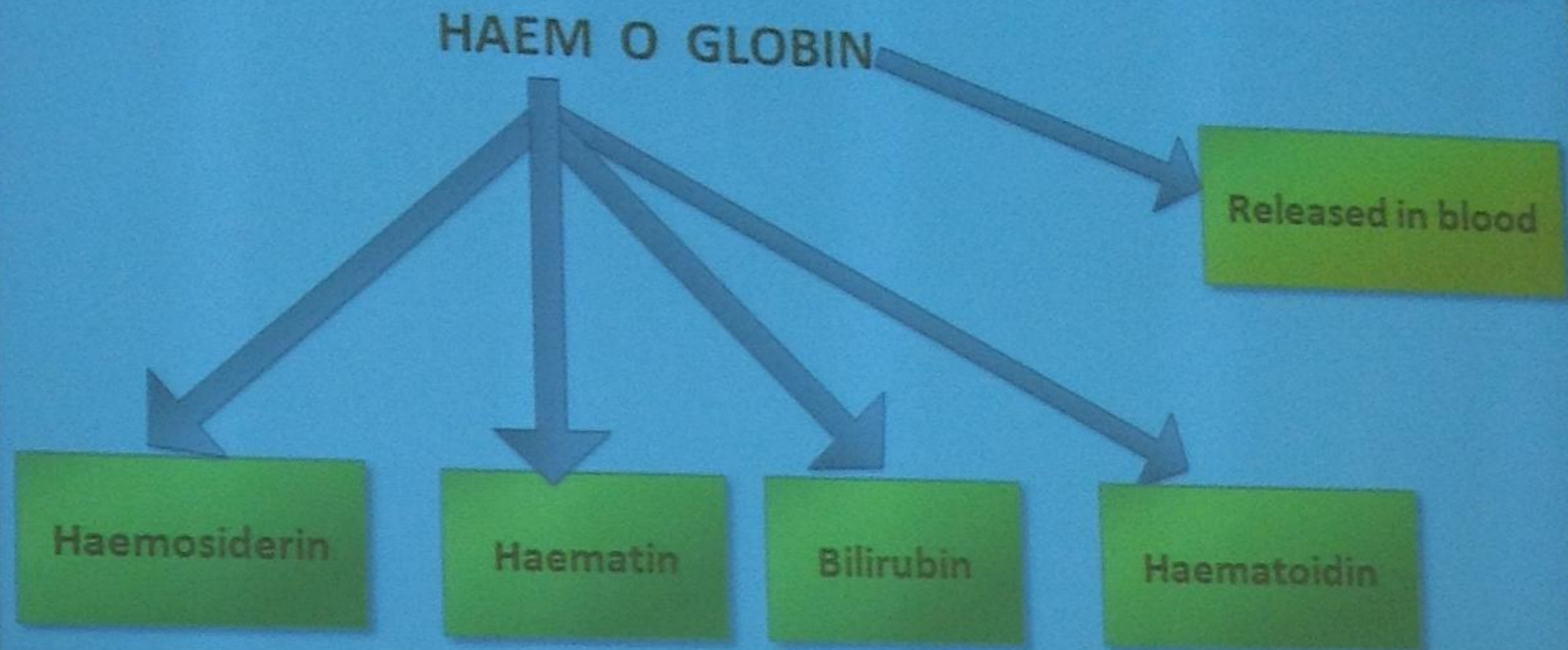
- Inborn error of metabolism resulting in accumulation of homogentisic acid
- Melanin-like pigment deposited in cartilage ear, nose, larynx, trachea, joints, tendons, ligaments

## ■ Melanosis coli

- brown or black pigment discolouration of the large bowel mucosa
- Origin not certain

# Pigments Derived From Haemoglobin

## Haemoglobin Breakdown



# Pigments Derived From Haemoglobin

## ■ Haemosiderin

- Excess iron stored in the cells, appears golden brown pigment
- Stored with a protein (apoferritin) – ferritin  
Insoluble granules give a Prussian blue reaction – ferric ferrocyanide complex

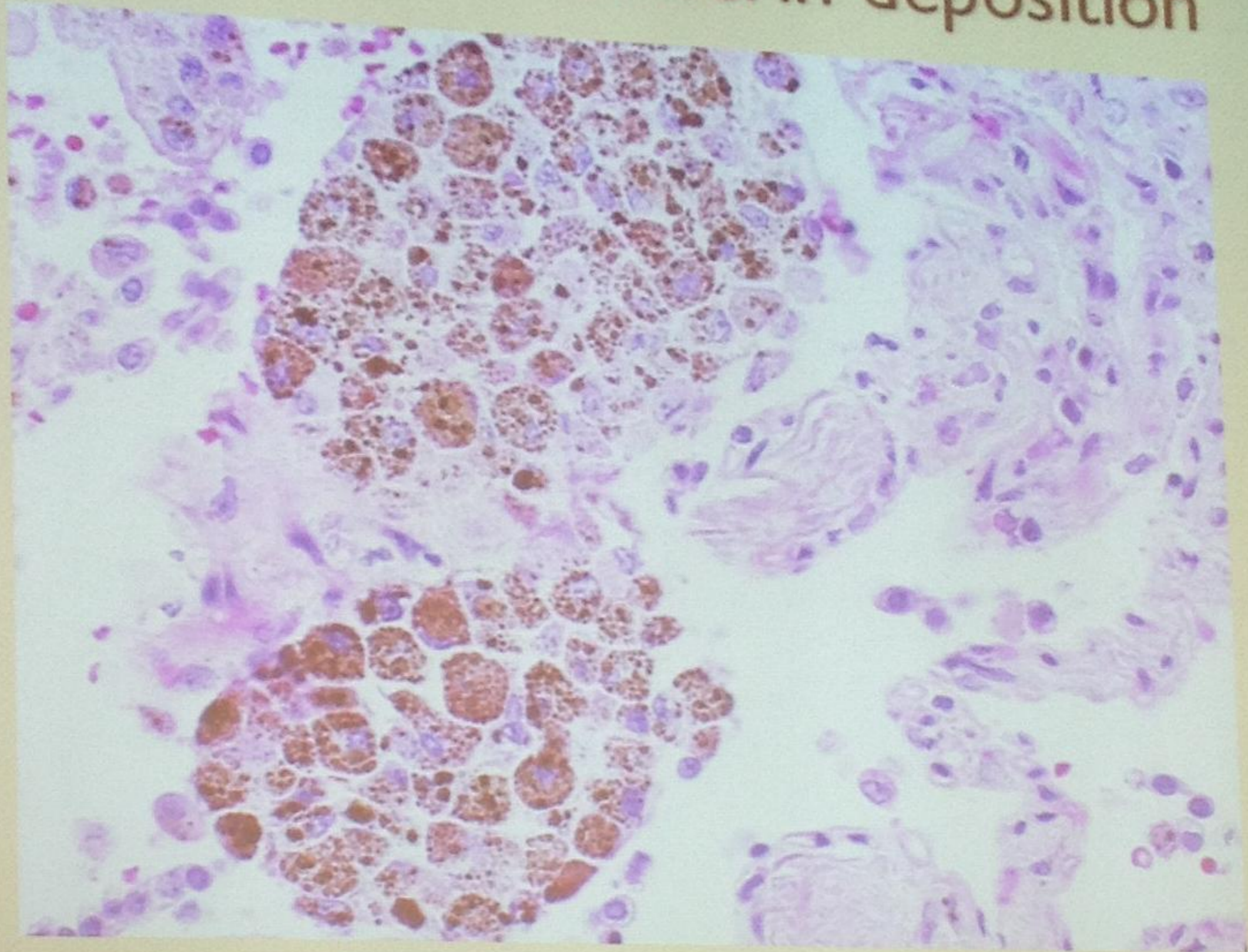
## ■ May be localised or generalised

# Pigments Derived From Haemoglobin

- ▣ Localised haemosiderin deposition:
  - Areas of haemorrhage – tissue injury (bruise) or haemorrhagic infarcts
  - Hb breakdown can be seen as different colour changes in a bruise
    - Red/blue – RBC in tissues, green-biliverdin, yellow-bilirubin, brown/black-haemosiderin
- ▣ May be seen in lungs – small haemorrhages in congestive conditions like MS and LVF



# Intra-alveolar haemosiderin deposition



# Pigments Derived From Haemoglobin

- Generalised haemosiderosis :
  - Occurs when there is overload of iron in the body.
    - Increase in RBC – increased iron absorption
    - Excess iron in diet or therapeutic
    - Following multiple blood transfusion
  
- Excess iron is stored in the liver, pancreas, heart and endocrine organs

# Pigments Derived From Haemoglobin

■ Haemochromatosis/Bronze diabetes is characterised by a triad:

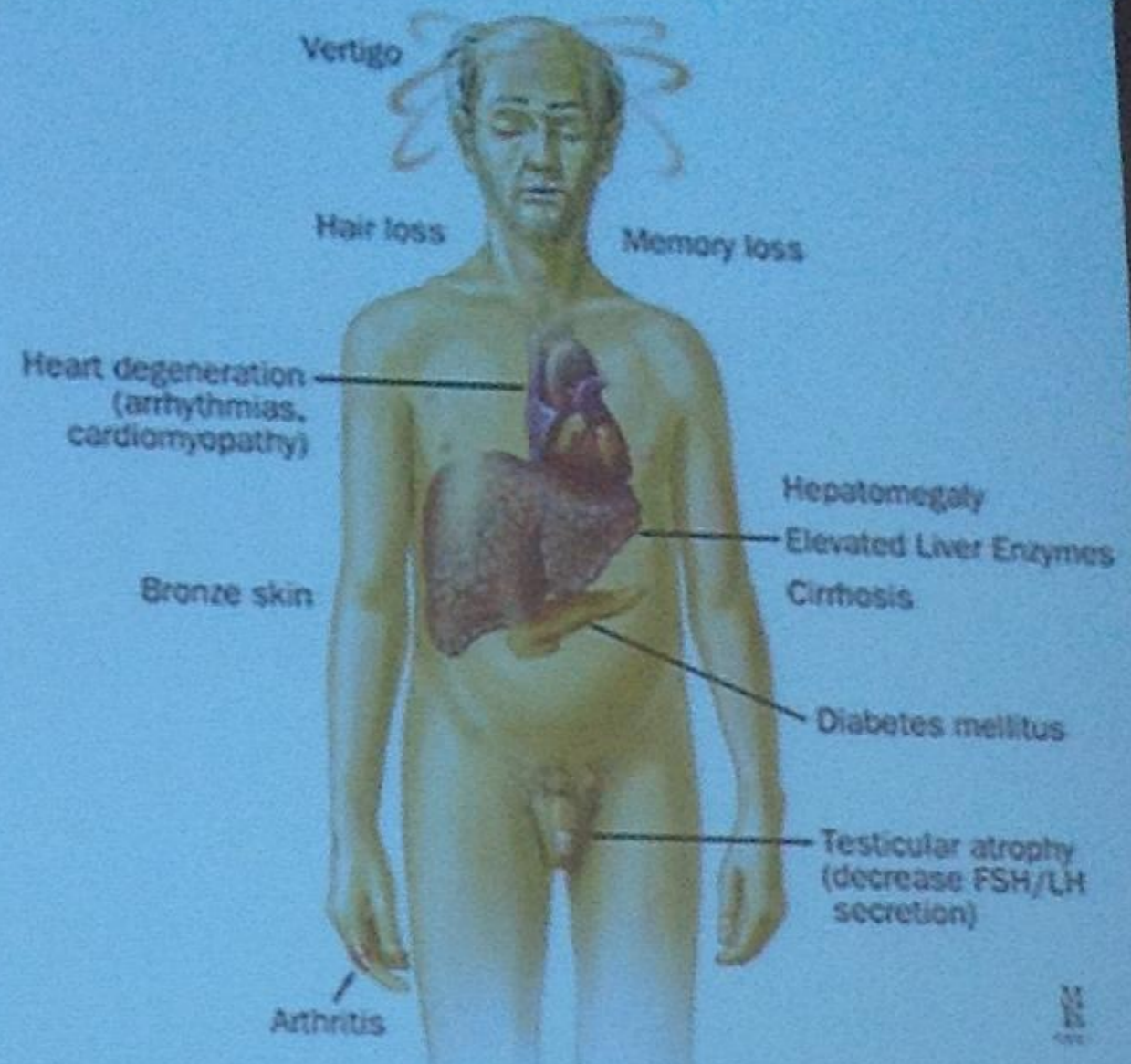
■ Hyperpigmentation

■ Diabetes mellitus

■ Liver cirrhosis

■ Males > females

■ Increased iron deposition in the internal organs



# Pigments Derived From Haemoglobin

- ▣ **African siderosis** : high dietary intake of iron found in home brewed beer prepared in iron containers



# Haematin Pigmentation

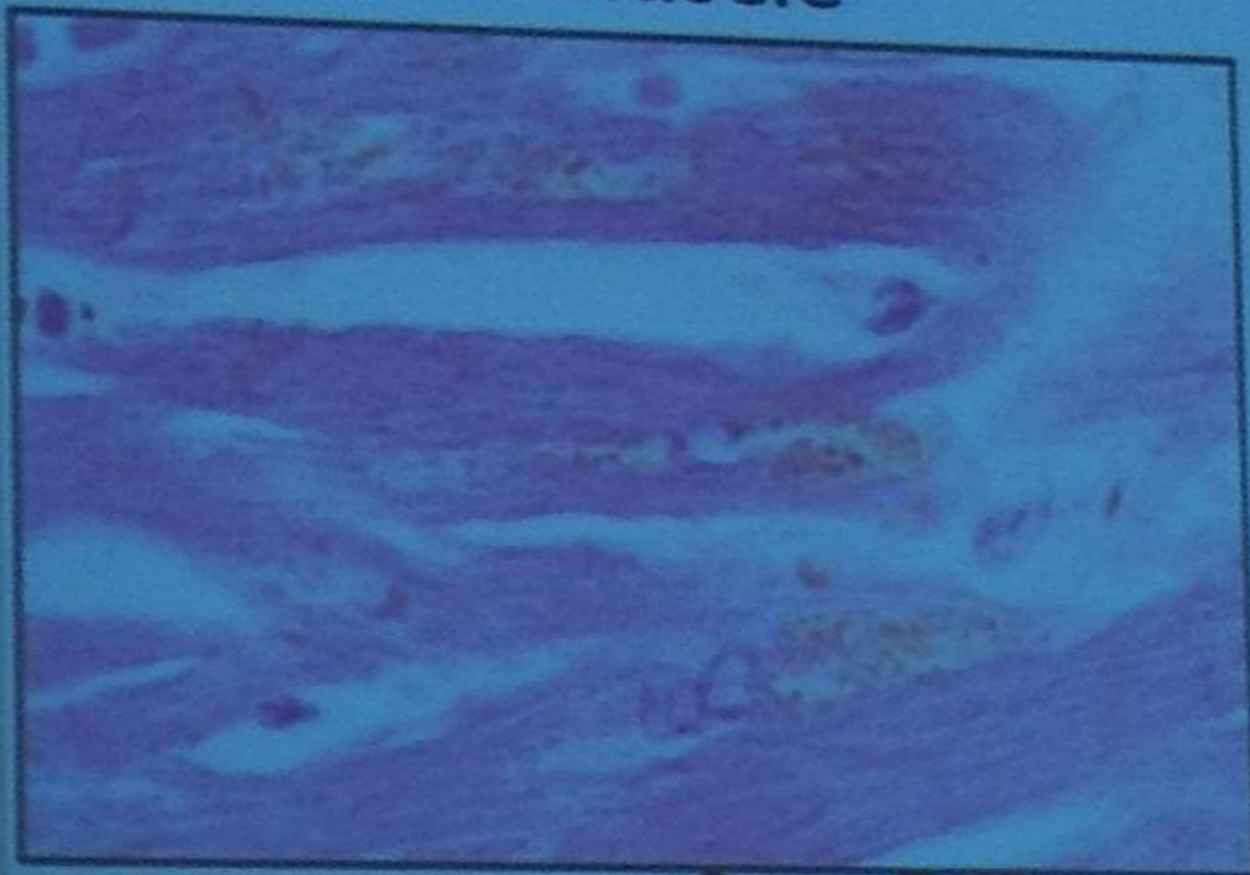
- ▣ Bluish black pigment derived from haem by oxidation of ferrous ion to ferric form
- ▣ Found particularly in macrophages of liver and spleen – malaria and schistosomiasis

# Pigments Associated With Fat

## ■ Lipofuscin

- Golden brown pigment thought to be 'wear and tear' resulting from cell injury or death in a tissue.

Cardiac muscle



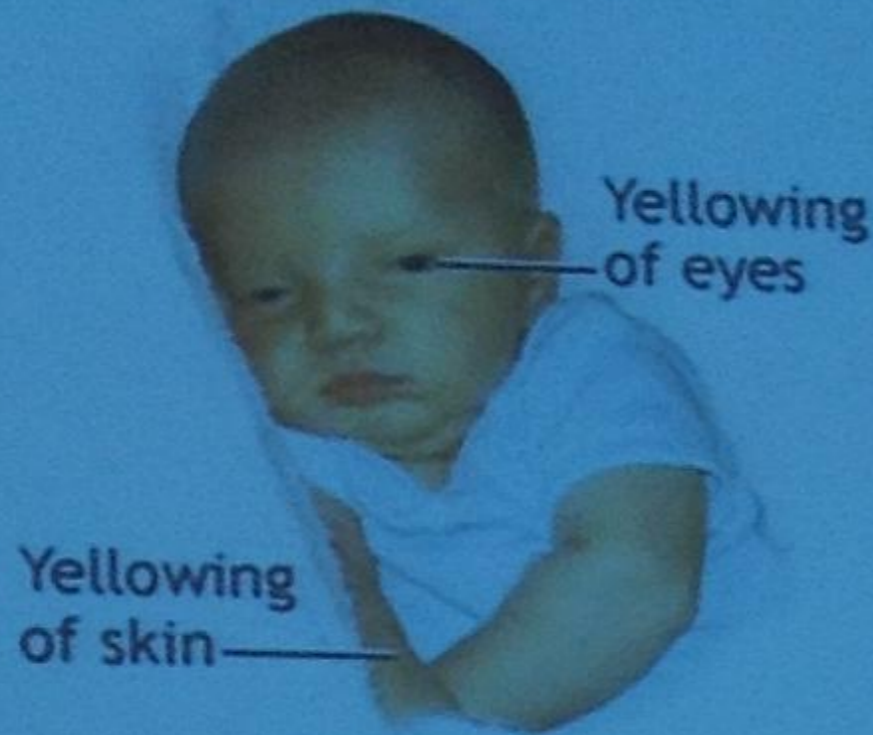
Liver



# Bilirubin

- Yellow pigment formed from the haem component of haemoglobin after removal of iron

Jaundice



Excess bilirubin in blood

Kernicterus



Bilirubin moves from bloodstream into brain tissue

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