

In the name of GOD



# Congenital Pigmentation disorders

**Amirnia M. MD**

**Associated professor**

Sina. Hospital

# Typing

✦ Hypermelanosis

✦ Hypomelanosis

# Hypermelanosis

# Periorbital melanosis

- ✦ Darkening of the skin around the eyes
- ✦ Physiological
- ✦ A.D
- ✦ First noted: below the lower lids
- ✦ Start: puberty (usually)
- ✦ Wide variation in intensity and extent
- ✦ Treatment: Topical

# Multiple lentiginos synd (Leopard)

- ✦ At birth- early death
- ✦ A.D
- ✦ Lentiginos: Neck + Upper trunk
- ✦ ECG abnormality: Valvular + Conduction defect
- ✦ Ocular hyperteloniom
- ✦ Pulmonary stenosis
- ✦ Abnormal genital
- ✦ Retard in growth
- ✦ Deafness

# Incontinentia pigmenti

- ✦ Vesicular+ verrucous + pigmented cutaneous lesion + defect in eye + skeletal +CNS
- ✦ X- linked (lethal in male) ( 95% female)

# Clinical features

- \* Birth ( before end of first week)
- \* Bulla + papular and warty lesion  
+pigmentation



# Clinical features

## \* Bulla :

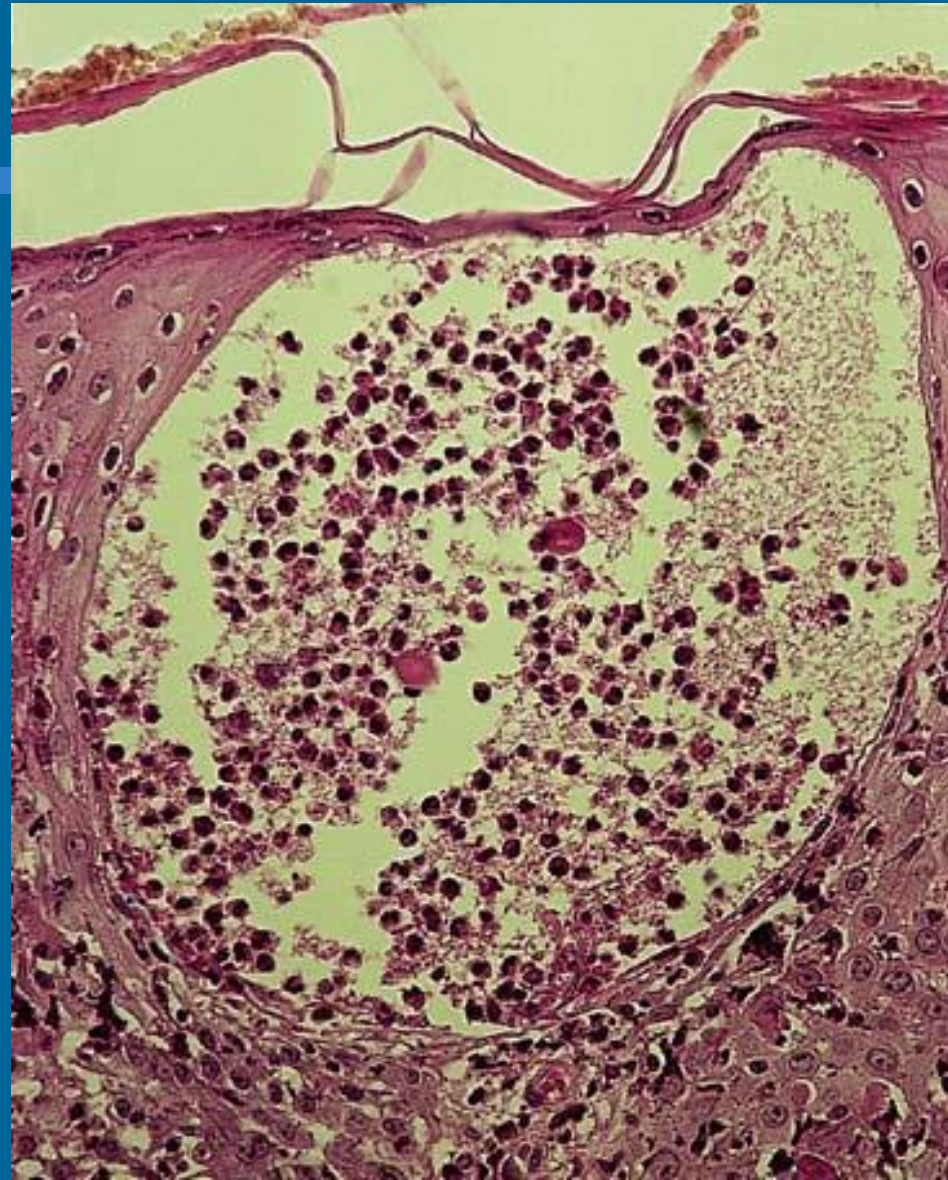
- \* clear tense, liner in limbs
- \* Days- month ( until 6 months)

## \* Linear warty:

- \* Back of hands and foots( until 6 month)

## \* Pigmentation:

- \* From normal or lesional skin
- \* Maybe only abnormality
- \* Blue- grey or brown
- \* Bizarre splashed (diagnostic)









# Others ...

- ✦ Dental defects (absence of teeth upper incisor + premolar)
- ✦ Occular defects
- ✦ C.N.S disorders: 25% ( MR- epilepsy)

# Albright's syndrome

- ✦ Unknown cause
- ✦ 4 months- 2years (occasionally in birth)
- ✦ Extensive light-brown patches (irregular margin)
- ✦ Trunk- buttock- thighs
- ✦ Asymmetrical
- ✦ Bone lesions: first decade + pathological fractures+ deformity

Continue...



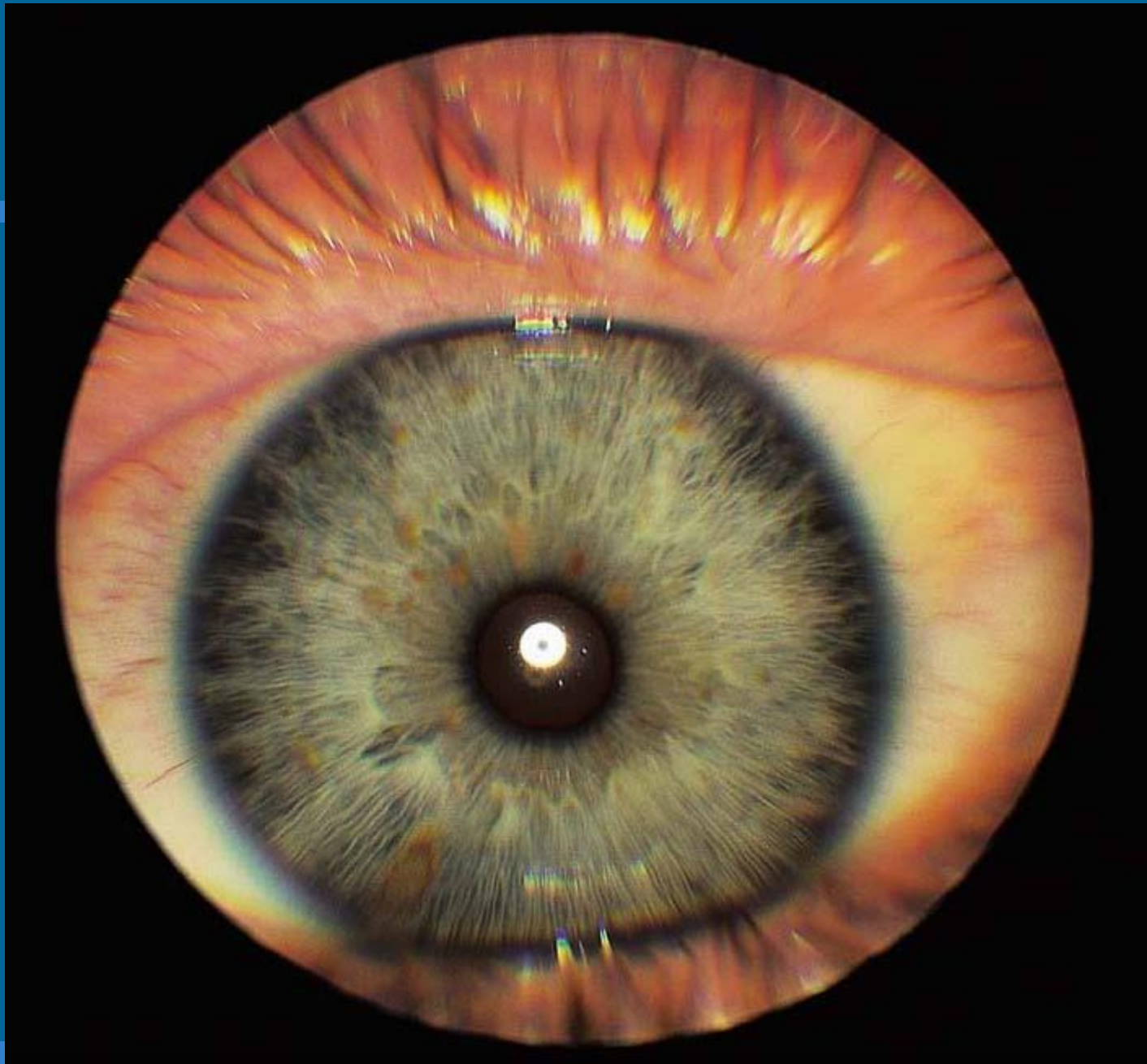


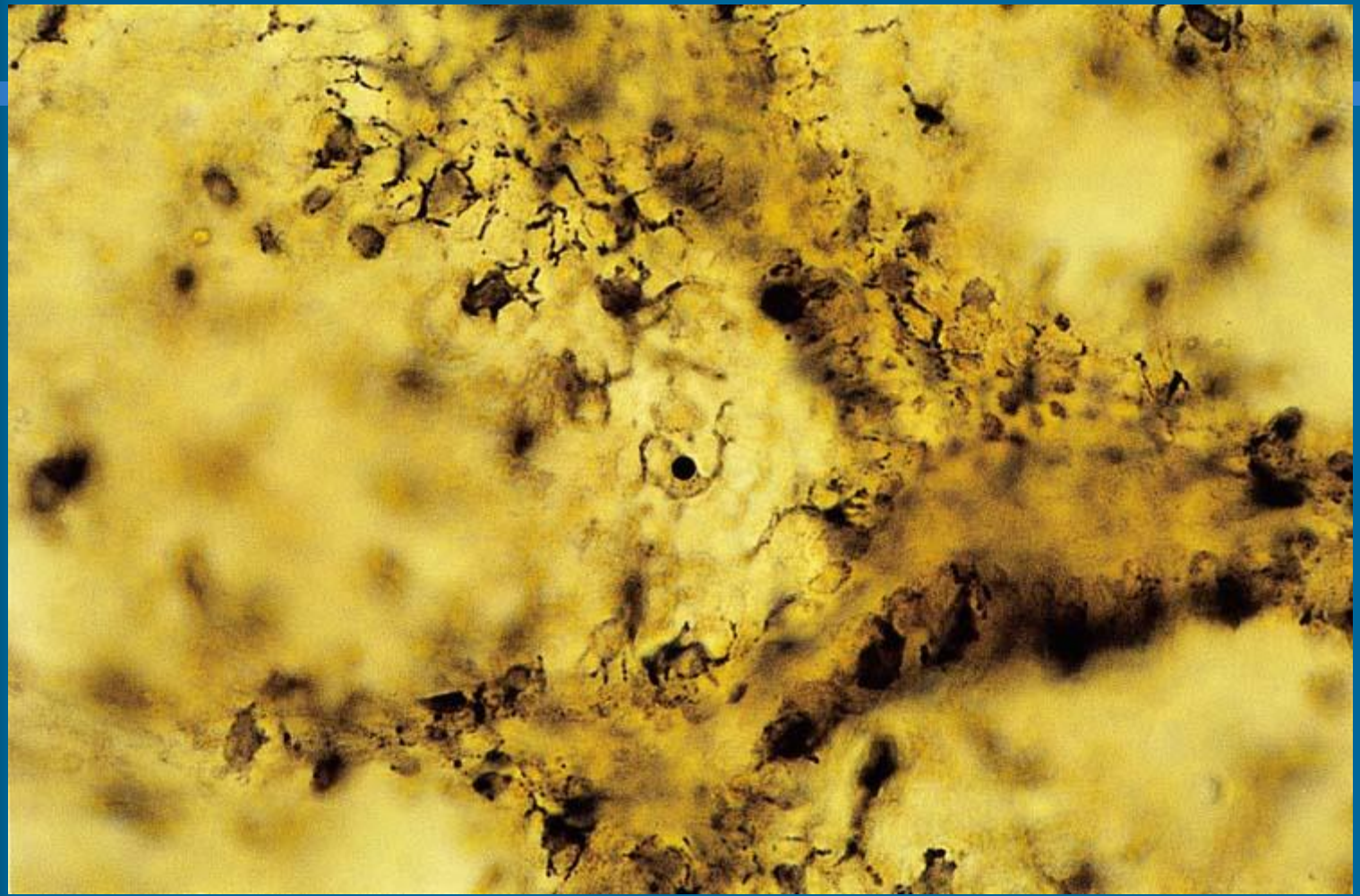
continue ...

- \* Bone lesion side → pigmentation ↑
- \* Ca+ P → Normal
- \* Precocious puberty in girls (under 10 years)
- \* Accelerated growth in childhood

# Nourofibromatosis

- \* Cafe- au- lait (90%)
- \* 10% normal variation
- \* Six or more → dis.





# Mongolian spot

- ✦ 90% mongoloid baby
- ✦ Lumbosacral ( leg occasionally)
- ✦ Poorly circumscribed blue- black
- ✦ Mistake for bruise
- ✦ Fade in early childhood (usually)
- ✦ Extrasacral → prolong





# Nevuse of Ota

- \* Hyper pigmentation affect one side of face  
( area supplied by ophthalmic and maxillary branch of trigeminal nerve)
- \* At birth (usually)
- \* Blue- brown color
- \* Sclera ⊕
- \* Deafness ( ipsilateral)

## Treatment:

\* Laser

\* Cryo







# Nevuse of ITO

- ✦ Increased pigmentation in area supply by post. Supra clavicular + lateral brachial
- ✦ Common in Japanese



# Hypomelanosis

# Piebaldism

- ✦ Or partial Albinism
- ✦ Absence of melanocytes and melanosomes in area
- ✦ Patch of depigment → at birth
- ✦ Unchange throughout life
- ✦ Most common: Frontal median
- ✦ White fore lock (100%) (rarely only lesion)
- ✦ Others site: upper chest+ Abdomen+ limbs
- ✦ Hand + feet+ back → Normal



# Treatment

- ✦ Photoprotection

- ✦ Skin grafts

- ✦ Psoralen therapy



# Incontinentia pigmenti achromians of ITO

- ✦ Whorled depigmentation
- ✦ Unilateral or bilateral
- ✦ Like incontinentia pigmenti (without bulla-warty)
- ✦ Birth
- ✦ Bone + eyes+ C.N.C dis.



# Albinism

- ✧ Partial or complete failure of melanin production in skin and eyes
- ✧ melanocyte → Normal (Number)
- ✧ Albinism:
  - ✧ Tyrosinase
  - ✧ Negative → most of melanosom in stage 1,2
  - ✧ Positive → most in stage 3
- ✧ Albinoidism: Involve skin+ hair (not eyes)
- ✧ Incidence: in UK  $\frac{1}{20,000}$  Birth

# Clinical features

✦ Marked dilution of pigmentation of :

✦ Skin

✦ Hair

✦ Eyes

✦ Tyrosinase negative:

✦ skin → Pink

✦ hair → white

✦ eyes → red reflex

continue...

- ✦ Tyrosinase positive: some pigment seen in iris+ skin + hair ( yellow color)
- ✦ Dark brown freckle
- ✦ Photophobia (in both type)
- ✦ Nystagmus in 100%





continue...

✦ ↑ actinic keratosis + S.C.C+ melanoma

✦ Treatment :

✦ No treatment

✦ Photo protection

✦ Regular examination for pre malignant lesion





The END