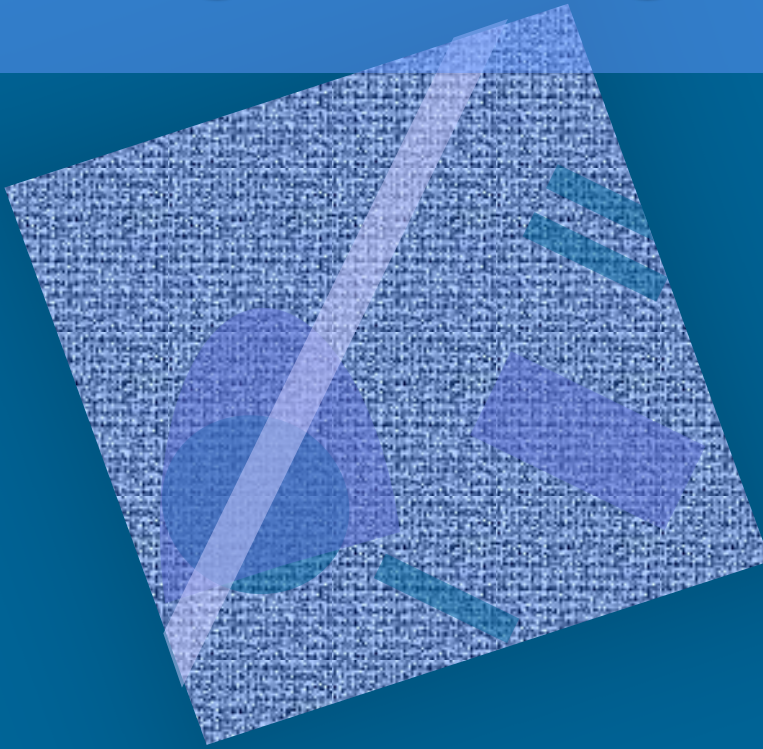


In the name of GOD



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 lentigines synd (Leopard)

- ✧ At birth- early death
- ✧ A.D
- ✧ Lentigines: Neck + Upper trunk
- ✧ ECG abnormality: Valvular + Conduction defect
- ✧ Ocular hypertelorism
- ✧ 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

Continue...

Clinical features

✴ Bulla :

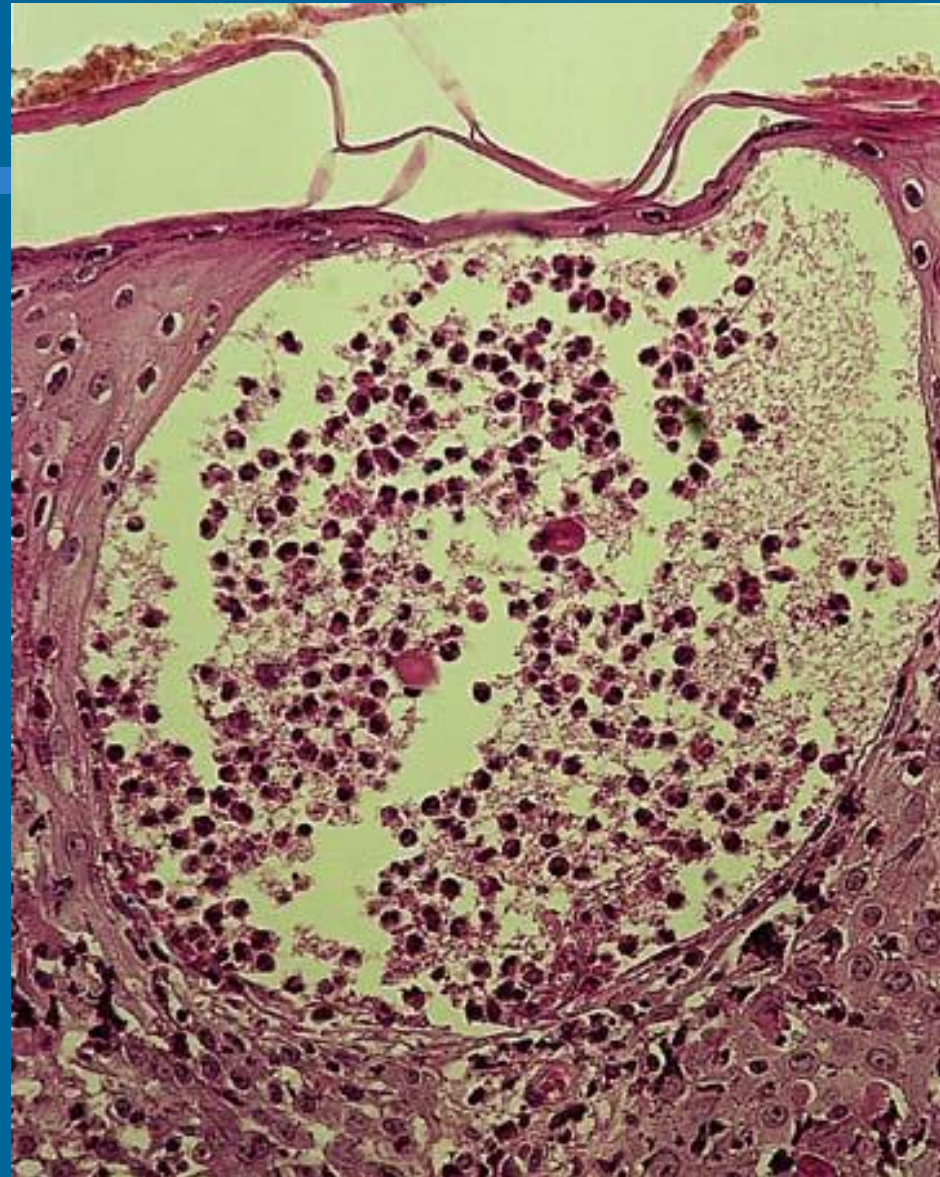
- ✴ clear tense, linear in limbs
- ✴ Days- month (until 6 months)

✴ Linear warty:

- ✴ Back of hands and feet(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...



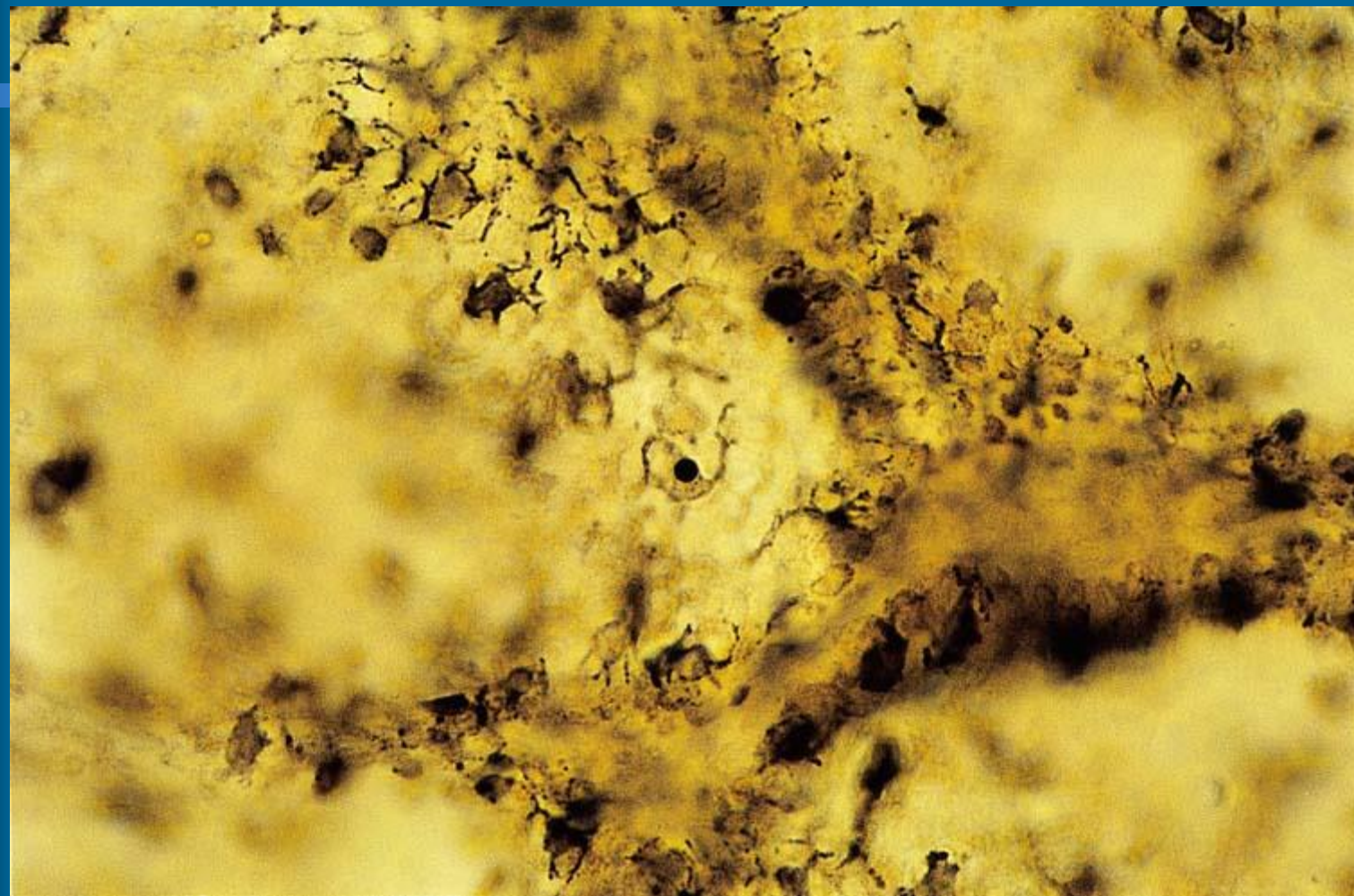
continue ...

- ✱ Bone lesion side → pigmentation ↑
- ✱ $\text{Ca} + \text{P} \rightarrow \text{Normal}$
- ✱ Precocious puberty in girls (under 10 years)
- ✱ Accelerated growth in childhood

Nourofibromatosis

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

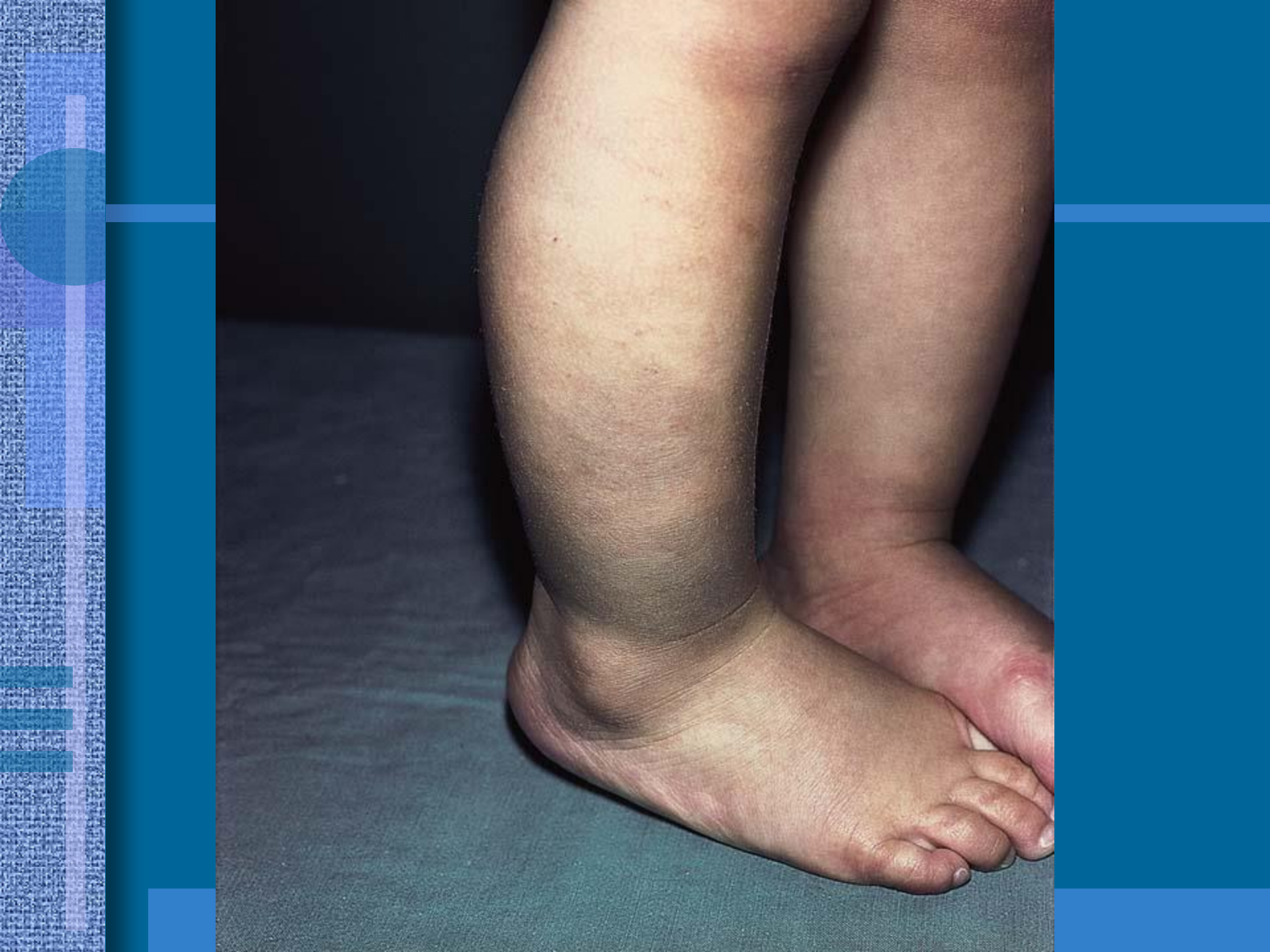




Mongolian spot

- ✧ 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 $1/20,000$ Birth

Clinical features

✧ Marked dilution of pigmentation of :

✧ Skin

✧ Hair

✧ Eyes

✧ Tyrosinase negative:

✧ skin → Pink

✧ hair → white

✧ eyes → red reflex

Continue...

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