Ichthyosis

Mohammad Al-Shloul

Mhmad Dayyat

Amro Osama

Definition

- Ichthyosis is a group of rare skin conditions that cause dry, scaly skin. It's usually something you're born with(congenital) , but it can happen later in life (Acquired) . It cannot usually be cured, but there are treatments to help the symptoms.

There is two types :

1) Congenital

2) Acquired

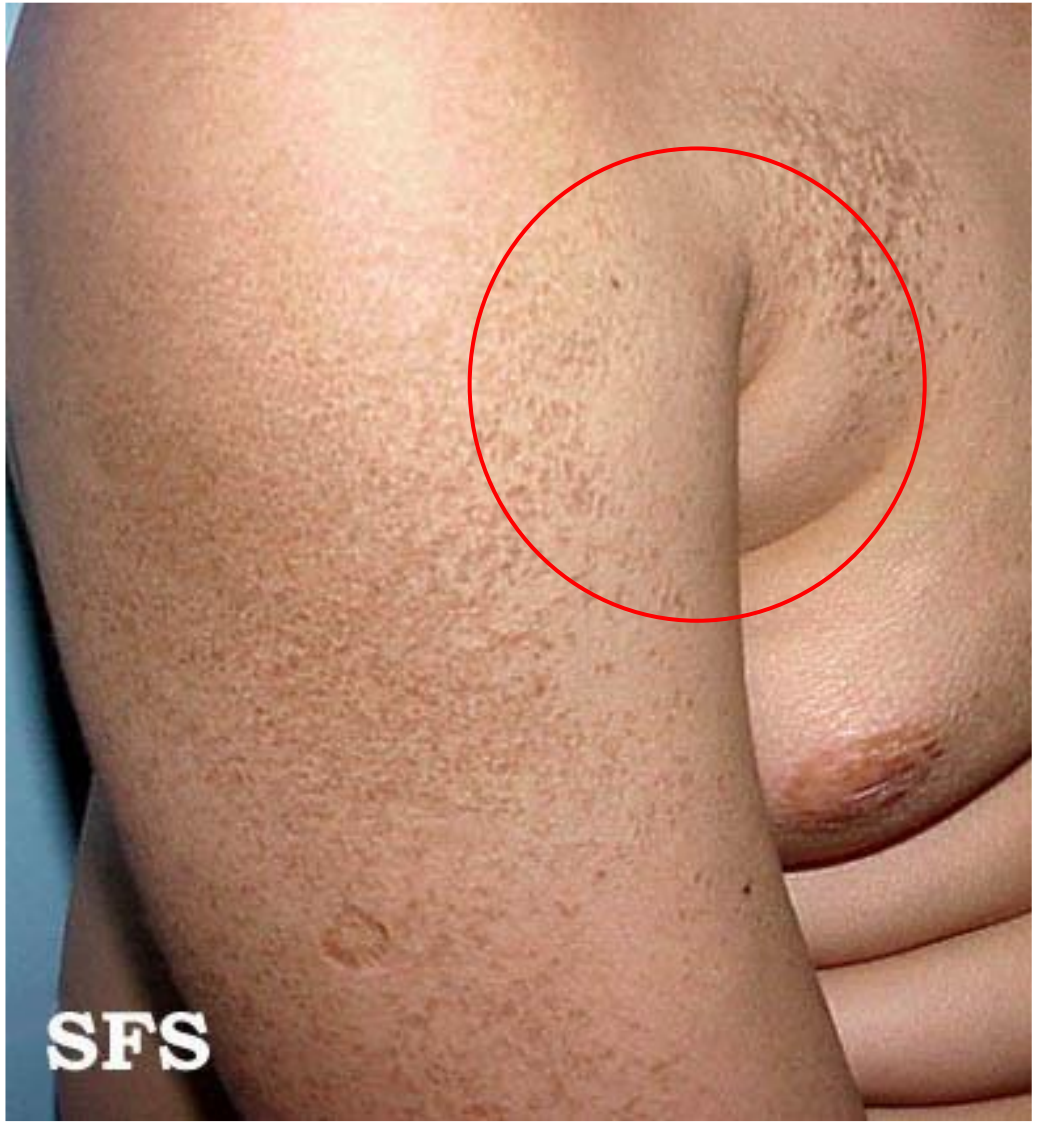
Congenital

- Ichthyosis vulgaris
- -Steroid sulfatase deficiency (x-linked recessive Ichthyosis)
- -Bullous–congenital ichthyosiform erythroderma (epidermolytichyperkeratosis)
- -Collodion baby
- -Lamellar ichthyosis
- -Congenital Ichthyosiform erythroderma
- -Netherton syndrome
- -Harlequine Ichthyosis

Ichthyosis vulgaris

- **Autosomal Dominant** Loss of function mutations in the filaggrin gene (FLG)
- -Not present at birth, appear later after few months
- -Dry skin , fine scales on extensor surfaces sparing the groin and flexural area
- -Increased skin marking of palms and soles , mild hyperkeratosis
- -Improves in summer , worsens with cold and dry weather
- -Associated with keratosis pilaris, Atopic dermatitis and asthma
- -Treatment emollients; keratolytic, ceramide containing lipid cream , urea
- -Improves with age

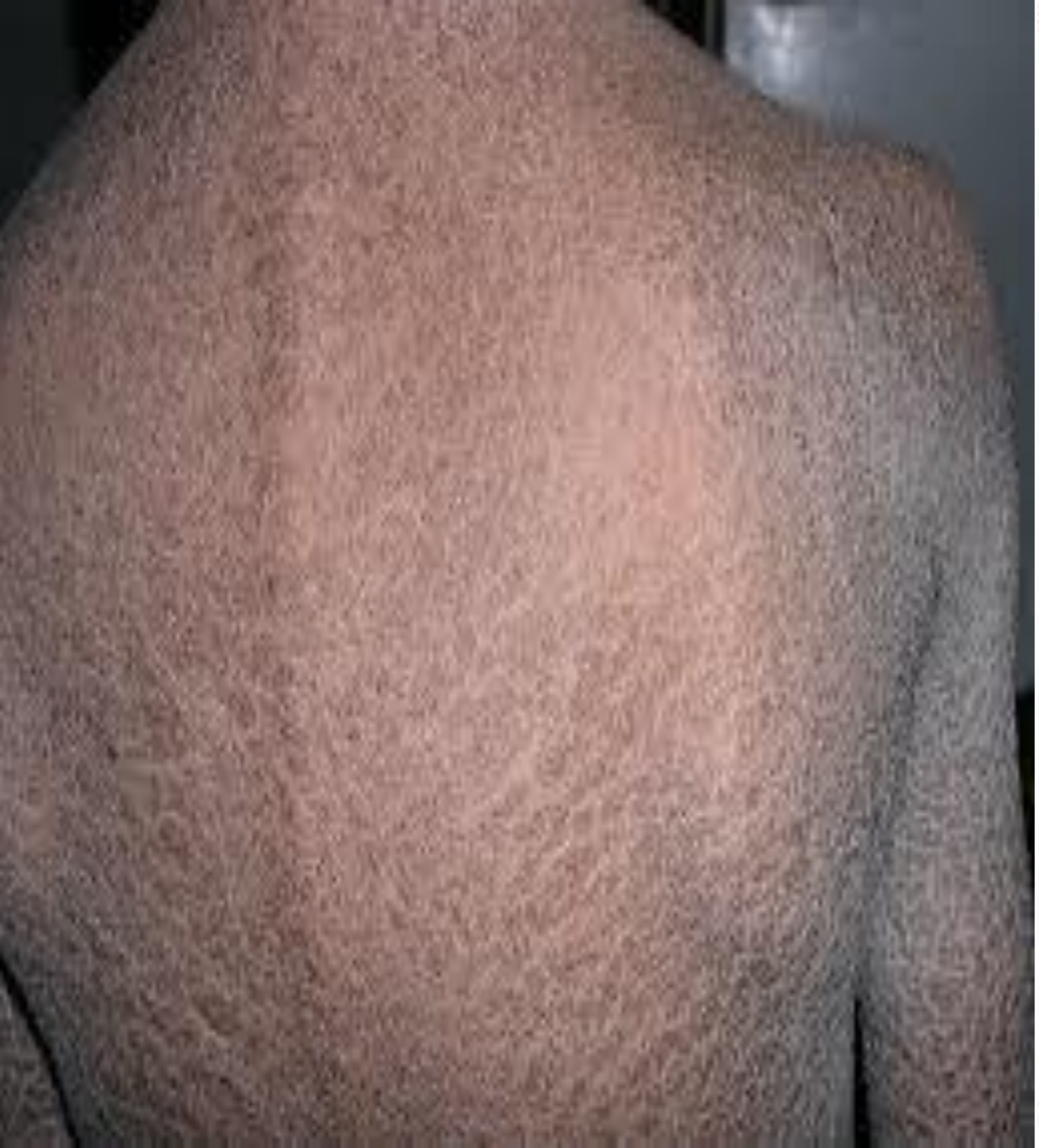
notice how it doesn't affect the flexures

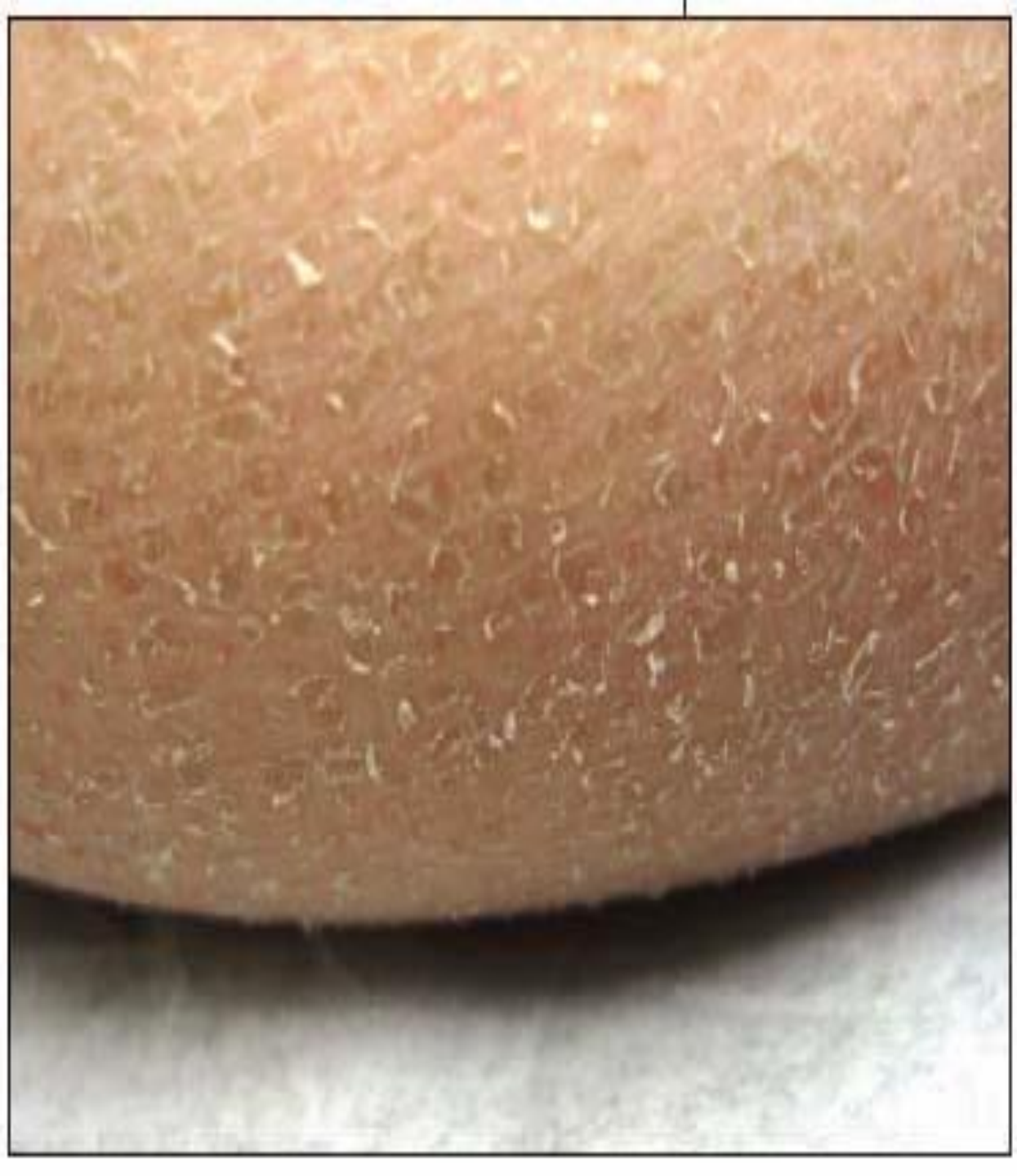


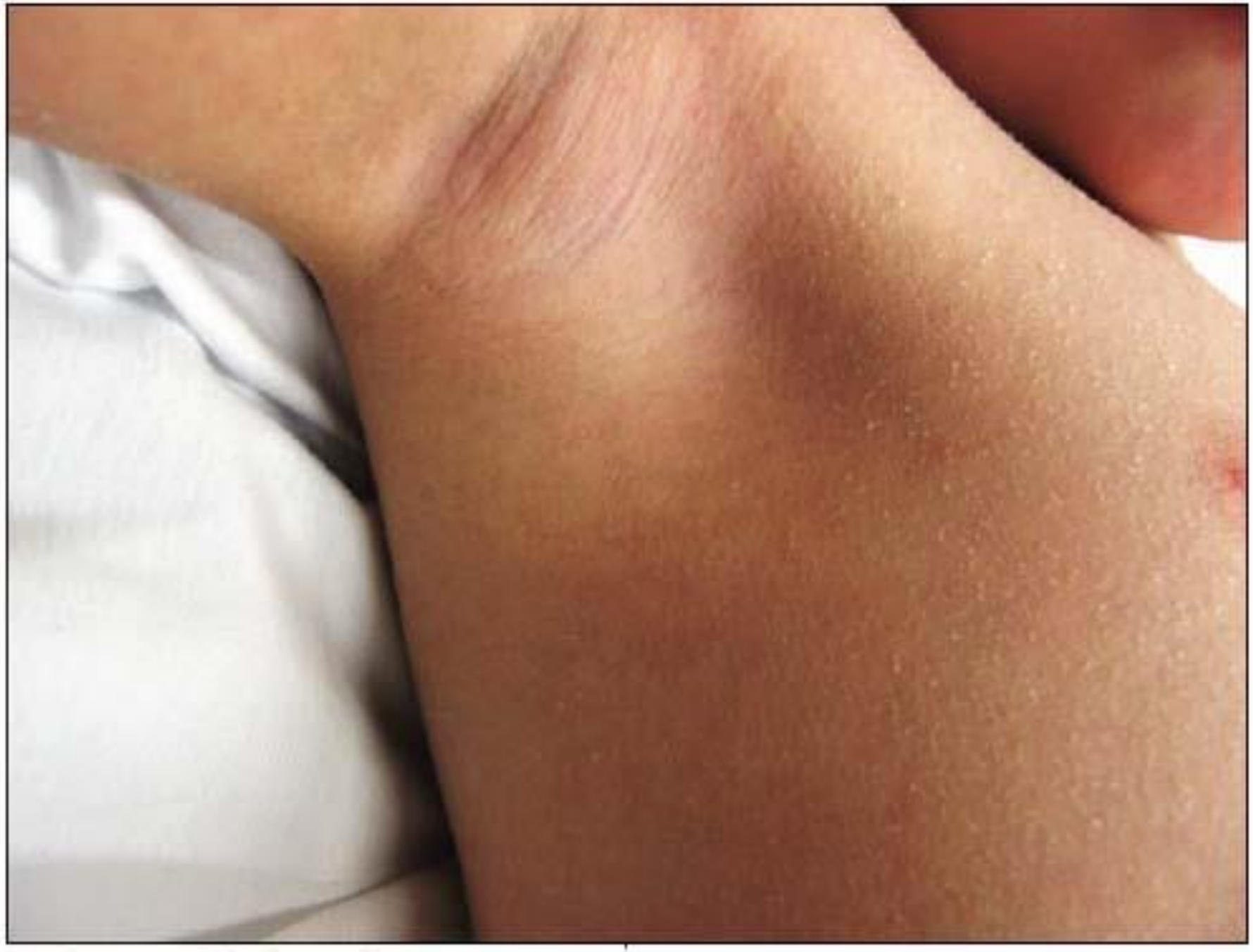
thickened skin of palms w/ exaggerated lines

Steroid sulfatase deficiency
(x-linked recessive ichthyosis)

- -X-linked recessive
- -Complete absence of steroid sulfatase is caused by complete deletion of the STS gene
- -90% are boys
- -Present within the first weeks after birth (mild erythroderma and peeling)
- -Typical large polygonal dark-brown scale with tight adherence to the skin develop later during infancy
- -Symmetrical involvement
- -Spare the palms , soles
- -May spare the flexures except the neck which is always involved (dirty neck)
- -Do not improve with age
- -Asymptomatic corneal opacities 10-50%
- -Cryptorchism







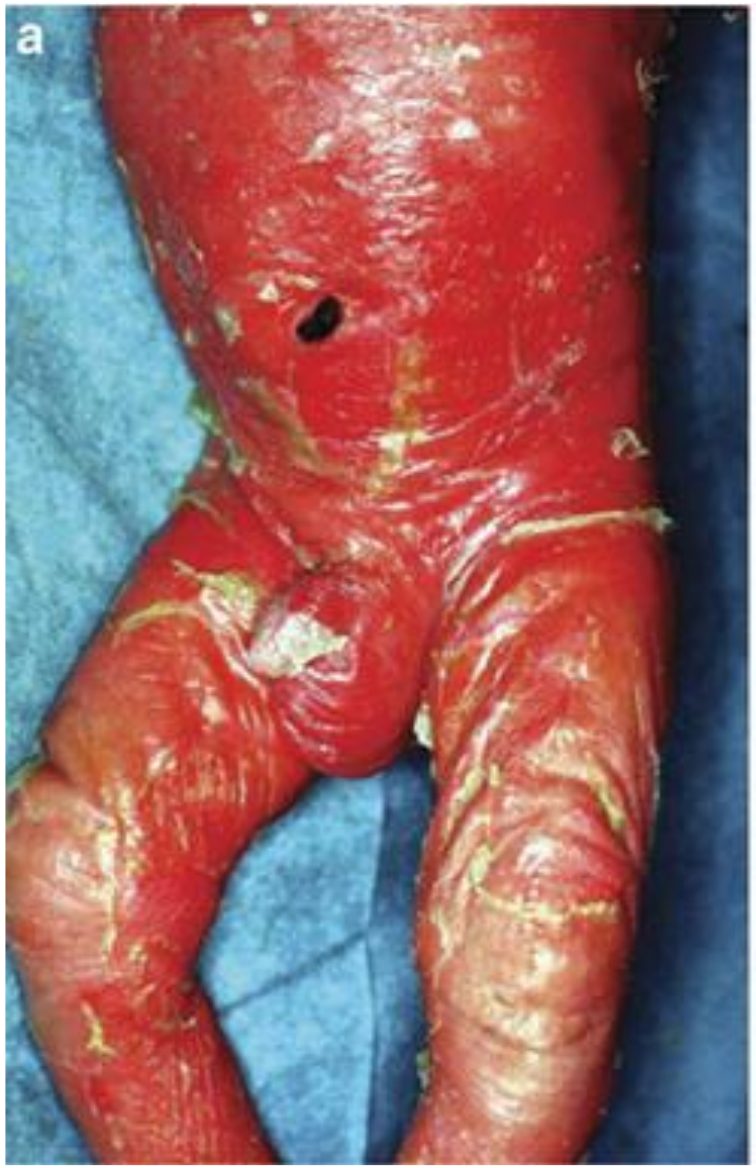
Difference between I.V. and X-linked Ichthyosis (clinically)

Traits	Ichthyosis vulgaris	X-linked Ichthyosis
Severity	Mild	Moderate
Affected sex	Both sexes	Only male
Onset	in between 3-12 months of age	before 3 months of age
Scales	Finer	Coarser and darker
Affected area		Abdomen is more affected than back
Spared area	Flexures and face	Palms and soles
Association with KP & AD	Present	Absent
Eye involvement & hypogonadism	Absent	Present

Bullous congenital ichthyosiform erythroderma Epidermolytic hyperkeratosis

- -Autosomal Dominant , 50% are sporadic
- - Caused by heterogeneous mutation in the gene encoding keratin 1(KRT1) and keratin 10(KRT10)
- -Present at birth with erythroderma and erosions
- -over time erythroderma decrease and hyperkeratosis prevails
- -Different clinical forms of presentation in different families
- -Chronic , disfiguring with great impact on social life
- -Treatment : according to the age , emollients, keratolytic, Retinoids

- Note : impaired skin barrier function -> complications like dehydration then it may cause scales that will be infected by bacteria, and it will cause (sepsis)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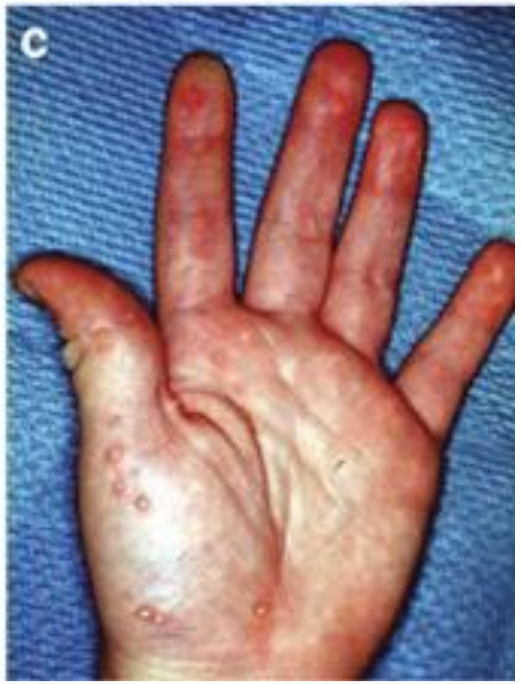
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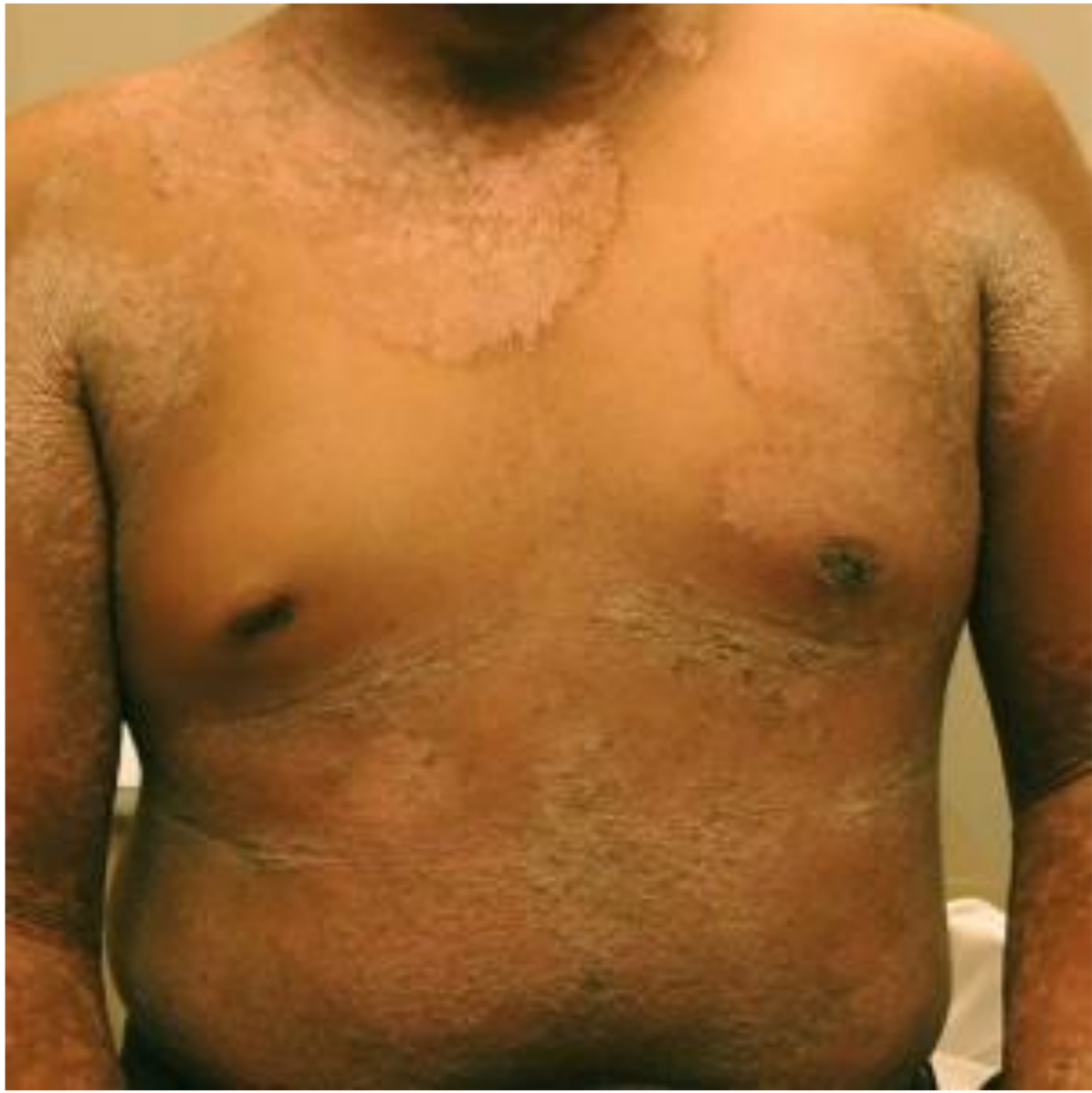

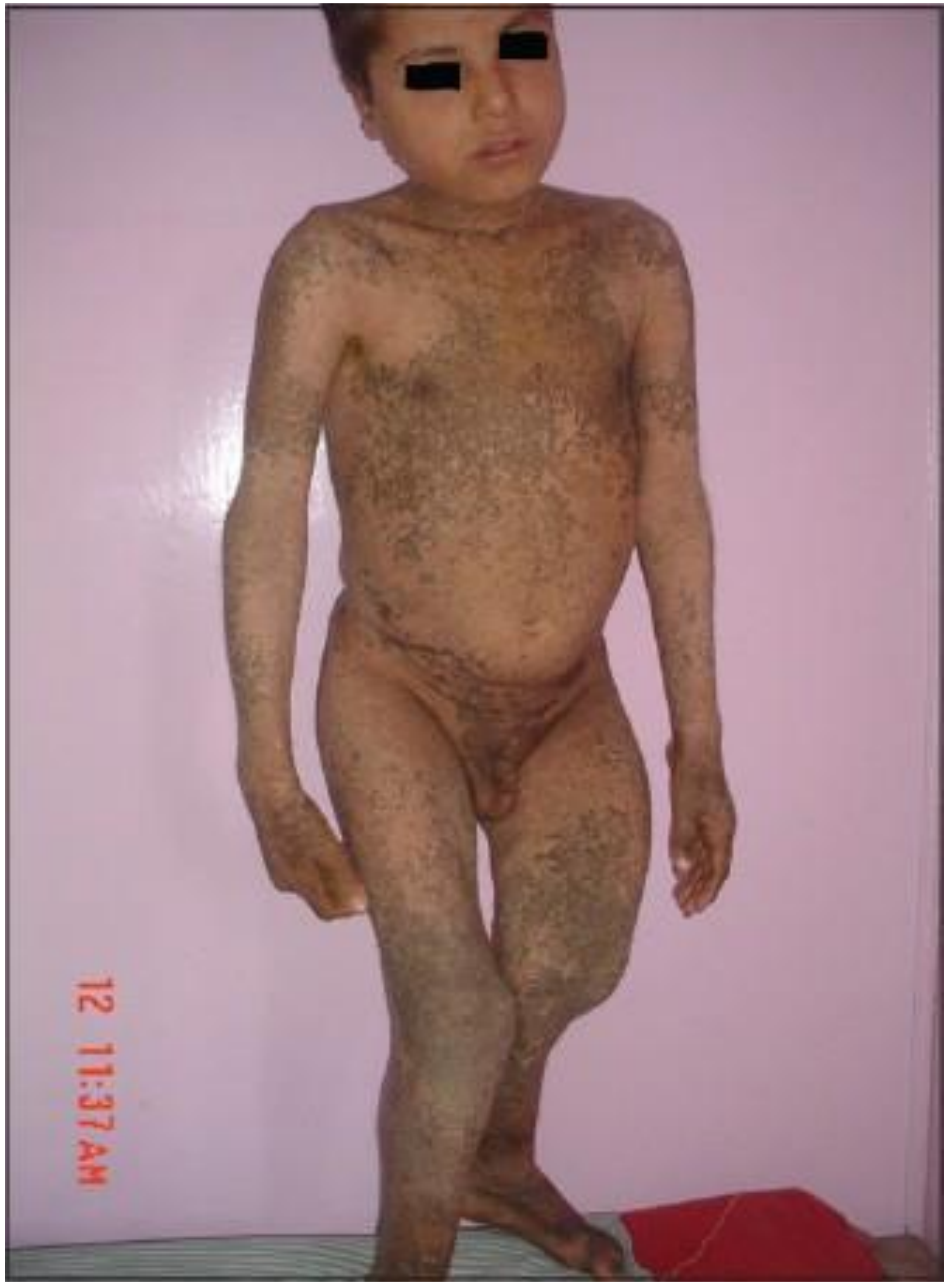
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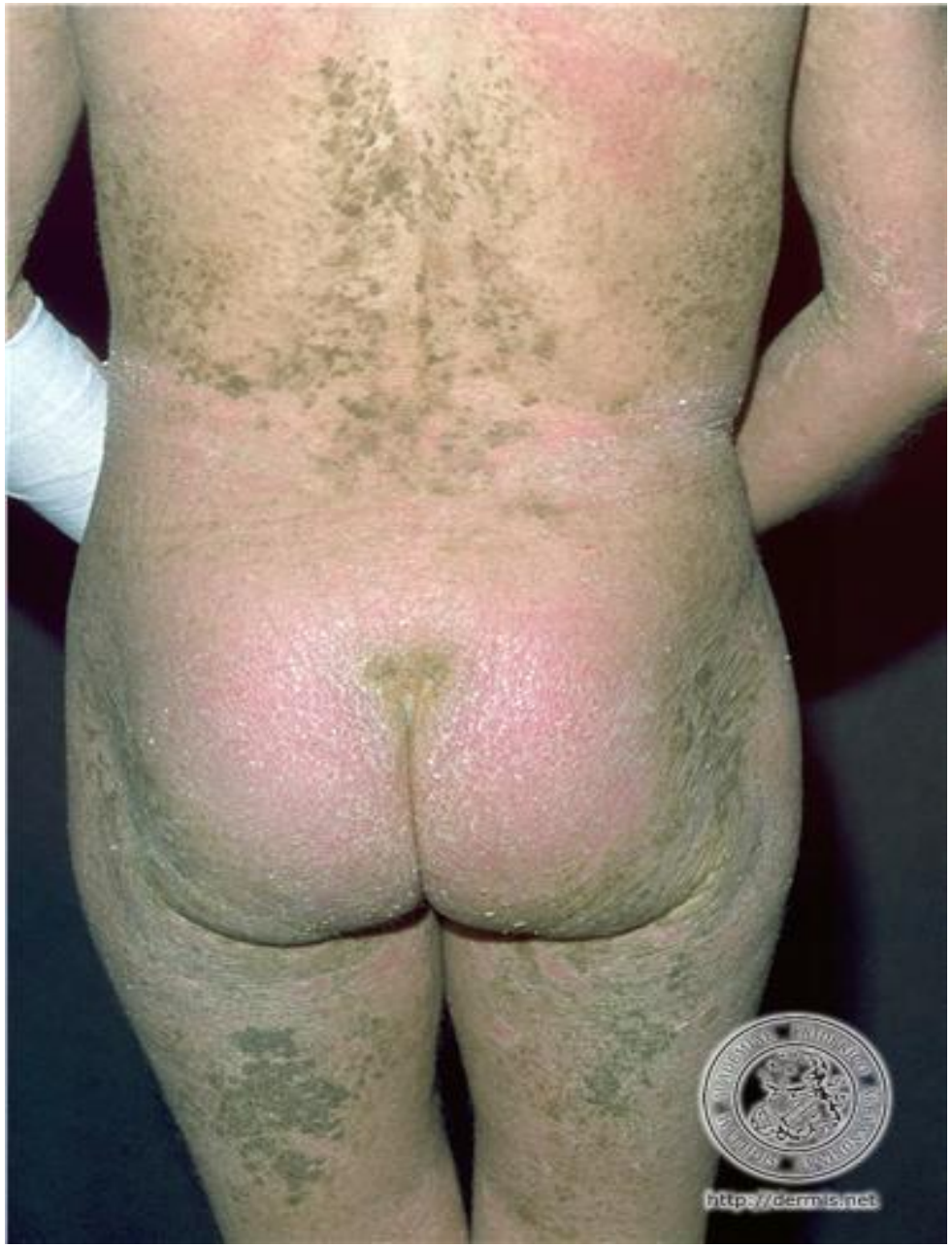
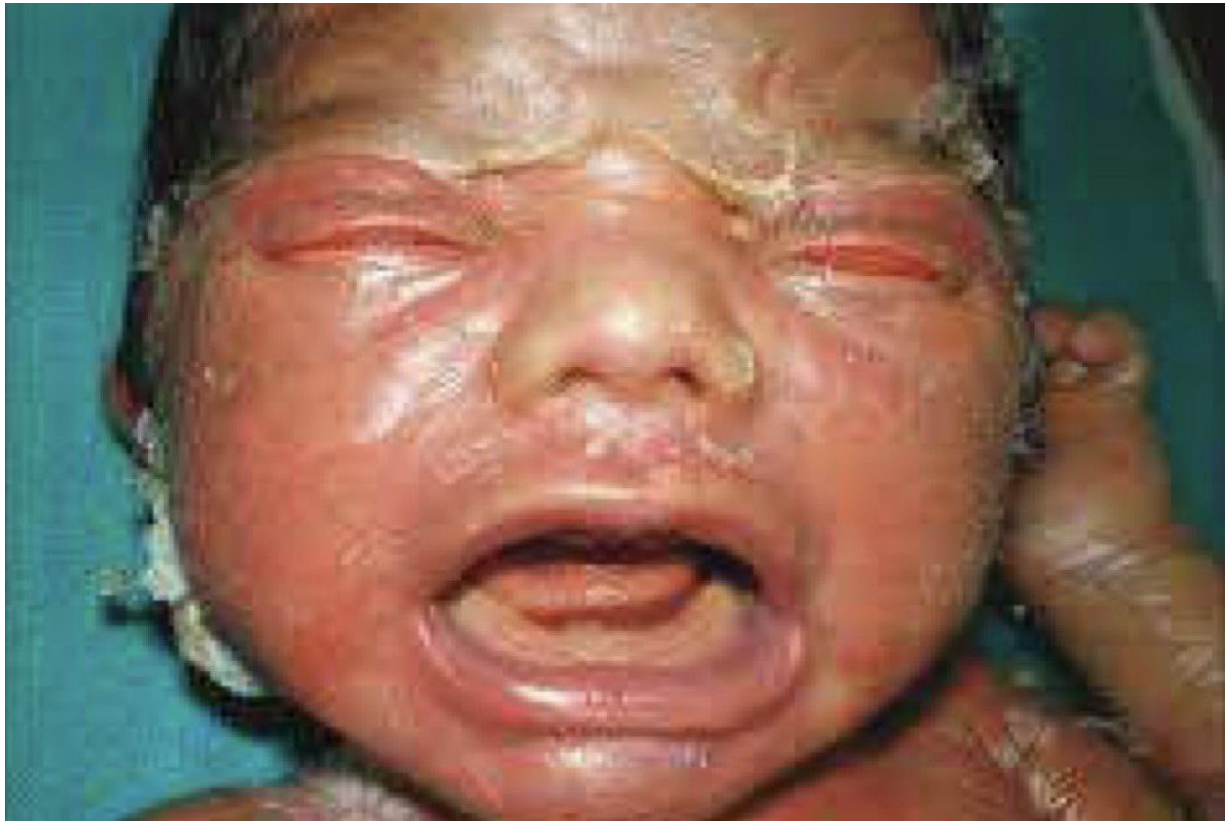
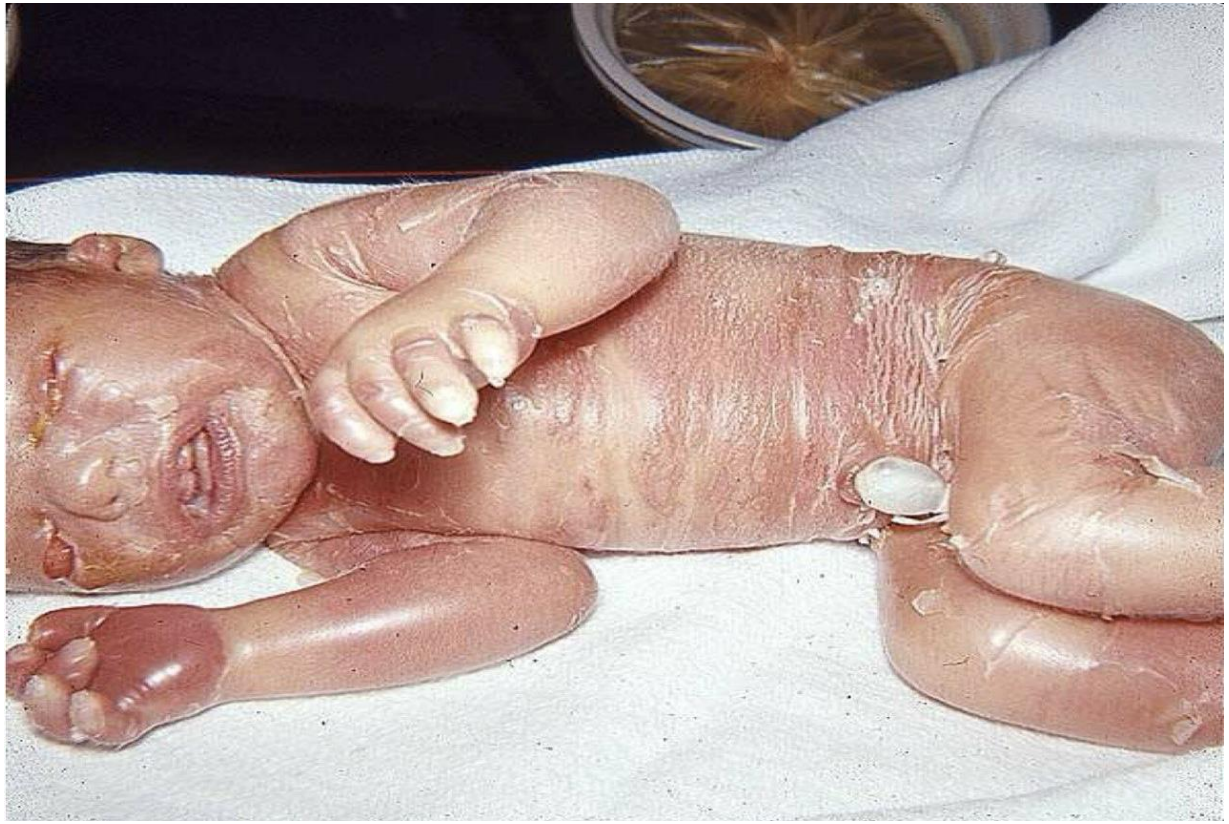

http://dermis.net

Collodion baby

- **Inheritance:** commonly autosomal dominant
- **Defect:** premature birth
- **Age on presentation:** at birth covered with a taut shiny and transparent membrane that resemble a plastic wrap
- **Clinical presentation:**
  - ectropion, eclabium, hypoplasia of nasal and auricular cartilages
  - After birth the membrane dries, cracks and breaks up, fissure develop
  - Dehydration, hypoxia, malnutrition and pulmonary infection may result (need special care)
  - Within 2 weeks, the membrane peels off → congenital ichthyosiform erythroderma or lamellar ichthyosis
  - In some cause normal skin appear or mild exfoliation
- **Treatment:** topical antibiotics and emollients

Collodion baby



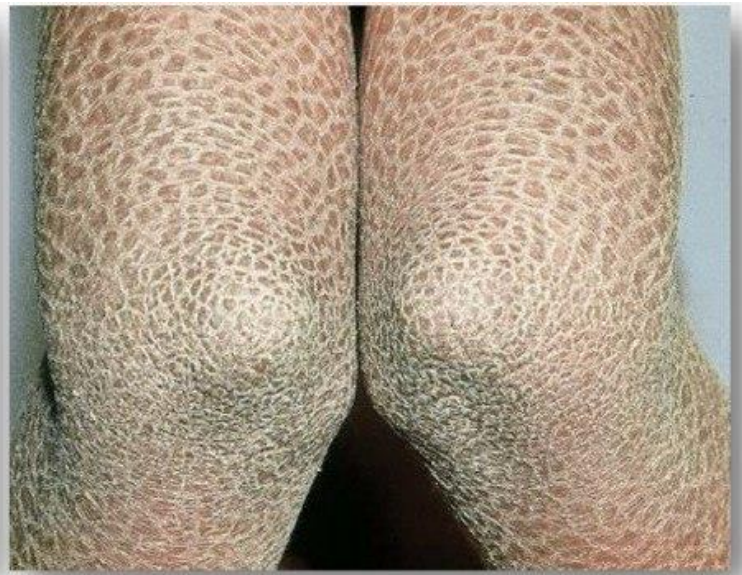


Lamellar ichthyosis (non-bullous ichthyosiform erythroderma)

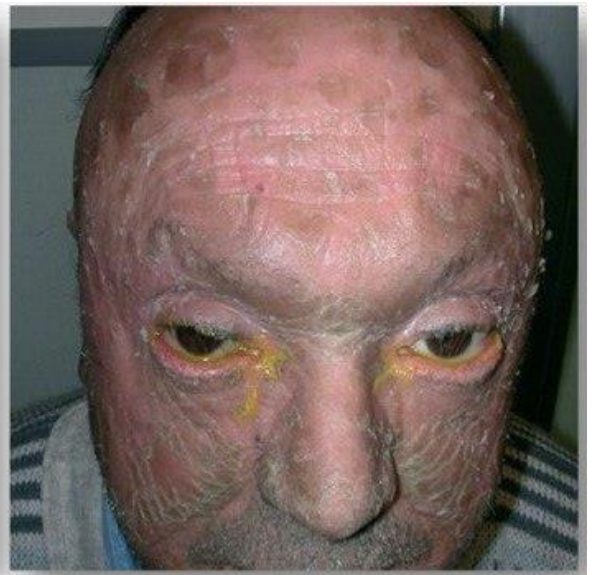
- **Inheritance:** Autosomal recessive
- **Defect:** transglutaminase deficiency due to mutation of TGMI gene
- **Age on presentation:** appears at birth as collodion baby
- **Clinical presentation:**
  - severe disorder
  - large, dark-brown plate-like scale
  - ectropion, eclabium, hypoplasia of nasal and auricular cartilages
  - scarring alopecia because of taut skin
  - mild to sever palmoplantar keratoderma
- **Treatment:** acitretin from early childhood

Lamellar ichthyosis

Very large dark scales, Ectropion, eclabium, deformed nose and ears and hair loss



Very large disfiguring scales

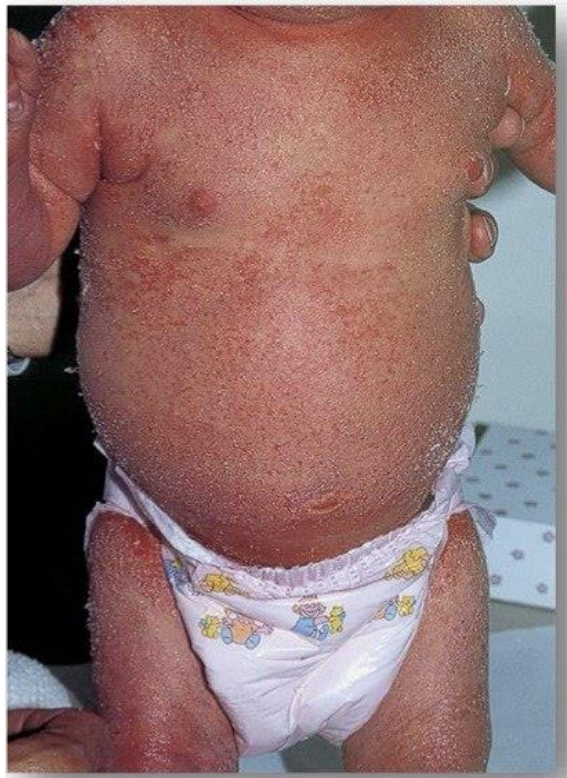


Ectropion, with exposure keratitis as a complication

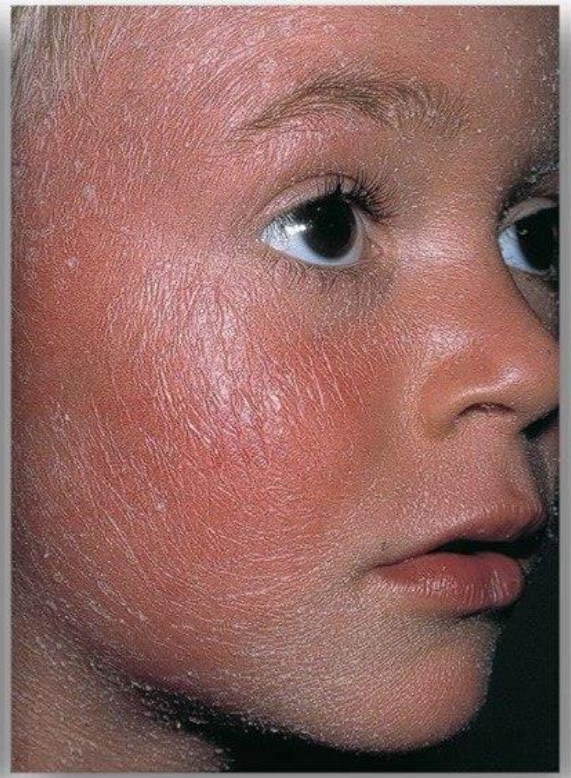
Congenital icthyosiform erythroderma

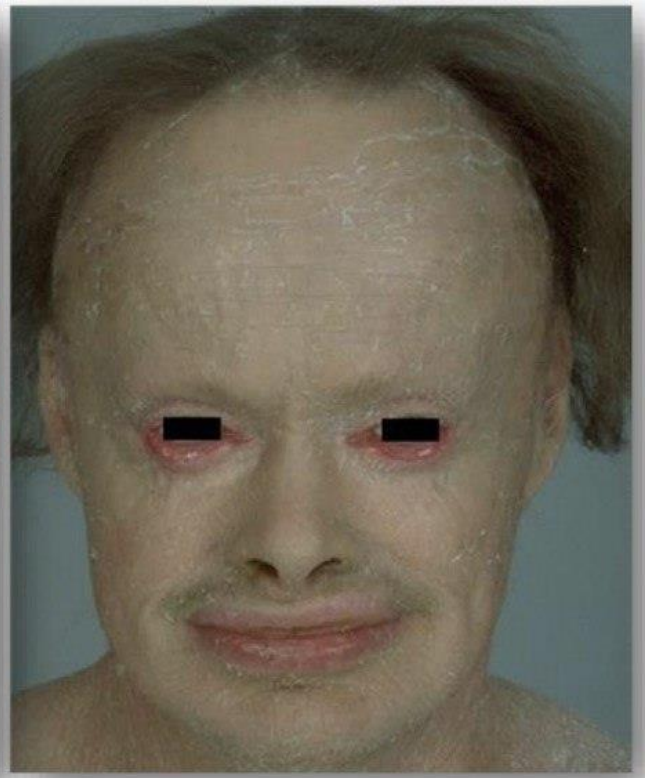
- **Inheritance:** Autosomal recessive
- **Age on presentation:** appears at birth as collodion baby
- **Clinical presentation:**
  - Generalized erythroderma with persistent scalling throughout life
  - Milder presentation than lamellar ichthyosis
  - Ectropion
  - Scarring alopecia
  - Sever palmoplantar keratoderma
  - Bright erythroderma, generalized, white powdery scales
- **Treatment:** acitretin as lamellar ichthyosis

Congenital icthyosiform erythroderma



Erythroderma with mild scaling.



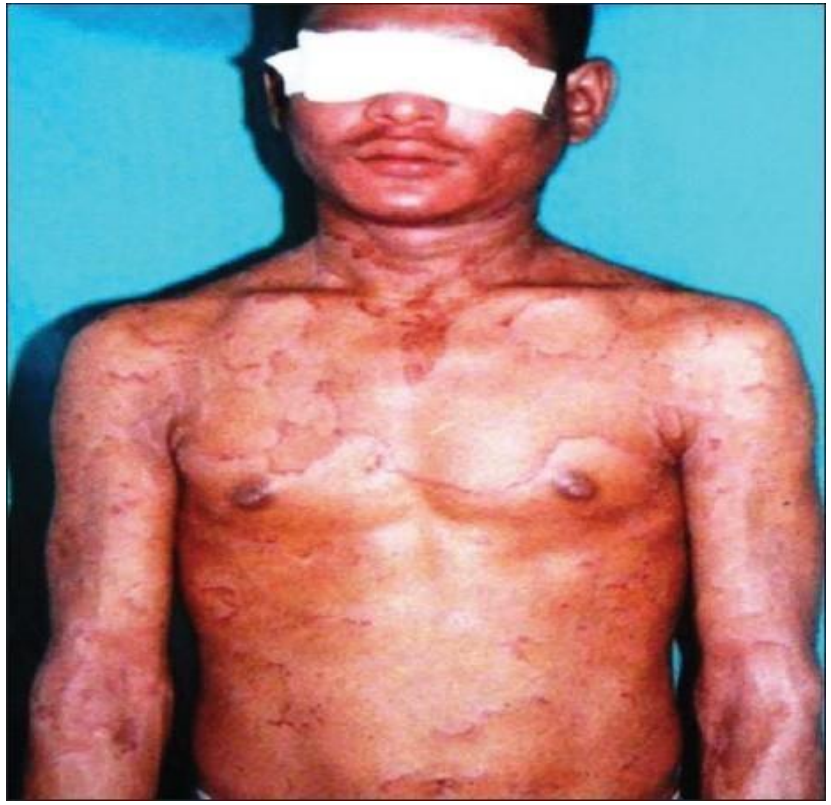


Scaling, scarring, Ectropion, Eclabium, deformity of the ears

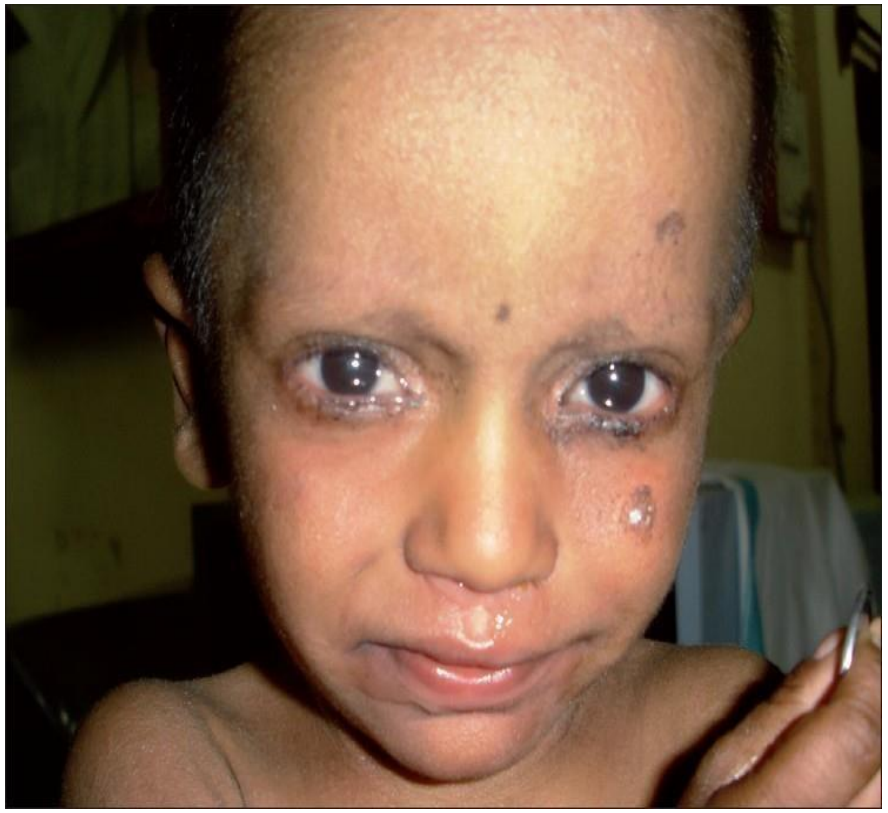
Netherton syndrome

- **Inheritance:** Autosomal recessive
- **Triad of** congenital ichthyosis, Trichorrhexis invaginata and atopy
- **Age on presentation:** Present at or soon after birth with erythroderma and scaling, no collodion baby
- **Clinical presentation:**
  - Gradually evolves into circinate scaling and erythematous plaques (Ichthyosis linearis circumflexa) over trunk and extremities and change over time.
  - Eczematous pruritic plaques due to atopy.
  - Scalp involvement.
  - Hair shaft abnormality since infancy (bamboo hair) improves with age
- **Associations**:
  - Elevated serum IgE due to atopy.
  - Increased susceptibility to infection.
  - Mental retardation can occur
- **Treatment:** If symptomatic: emollients, Retinoids and phototherapy

Netherton syndrome



Ichthyosis linearis circumflexa.



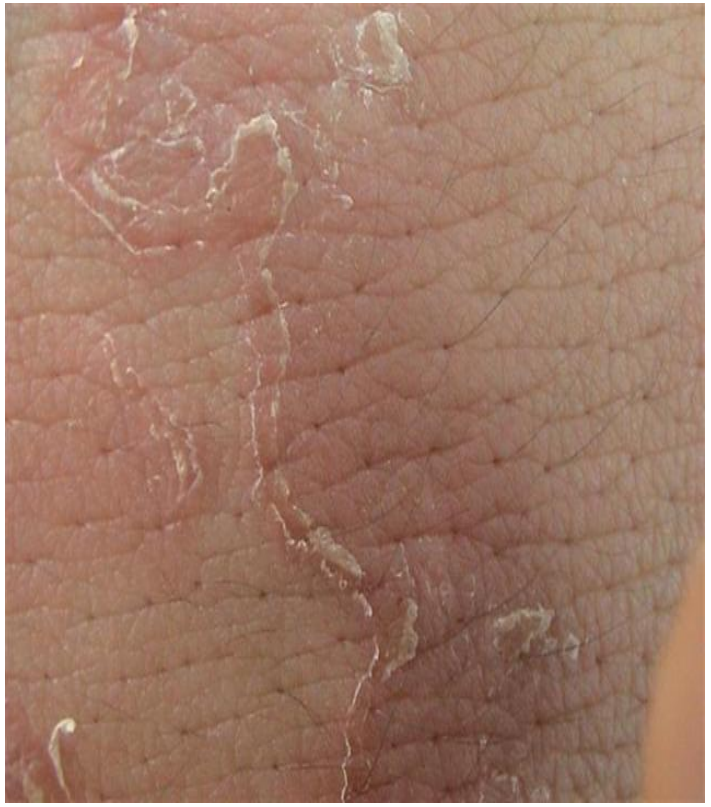
Scalp alopecia

Hair defects with eczematous features on the face

Netherton syndrome

Bamboo hair which is a feature of Trichorrhexis invaginata.

Trichorrhexis invaginata under light microscopy.

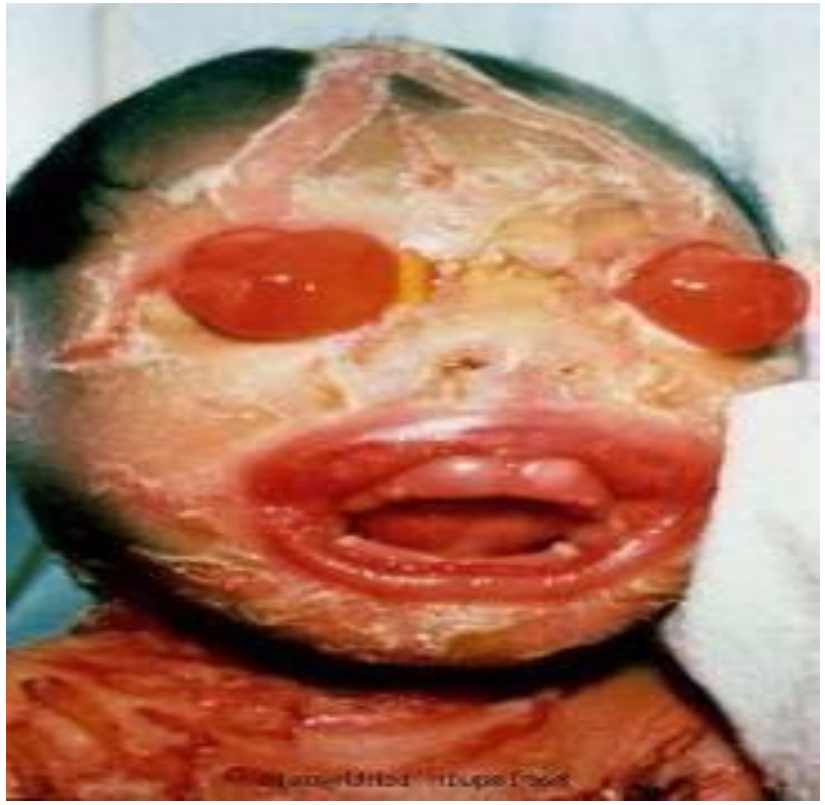


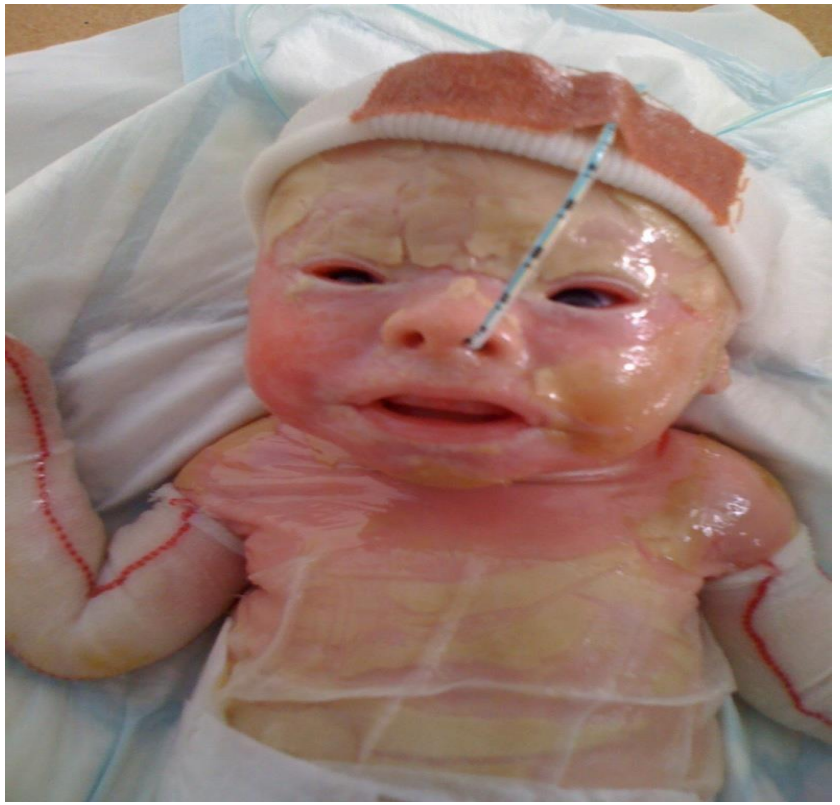
Double edge scaling which is a feature of Ichthyosis linearis circumflexa.

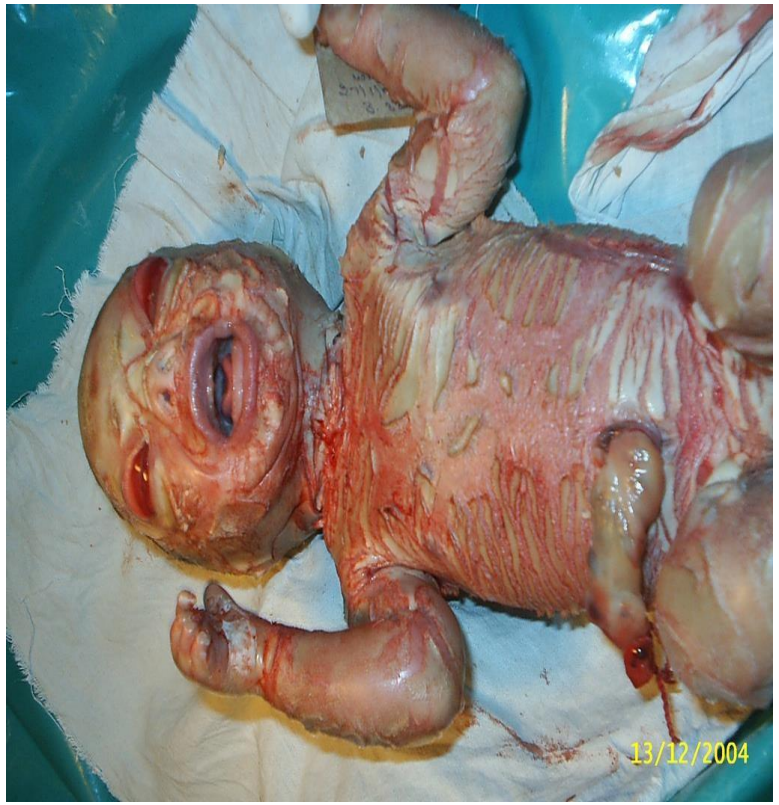
Harlequin Ichthyosis

- Most extreme and distinct form of congenital ichthyosis.
- **Age on presentation:** Premature baby, die within few days or weeks After birth
- **Clinical presentation:**
  - Encased in a hard, armor-like thick stratum corneum that severely immobilizes the baby.
  - After birth this taut cast cracks and form large, yellow adherent plates with deep fissures resembles a harlequin`s costume.
  - Ectropion – eversion of eyelids.
  - Eclabium – eversion of lips.
  - Microcephaly.
  - Edematous hand and feet, digits are well developed.
  - Eyelashes and eyebrows are missing.
- **Treatment:**
- Need special care for water and electrolyte balance and prevention of sepsis
- Retinoids

Harlequin Ichthyosis







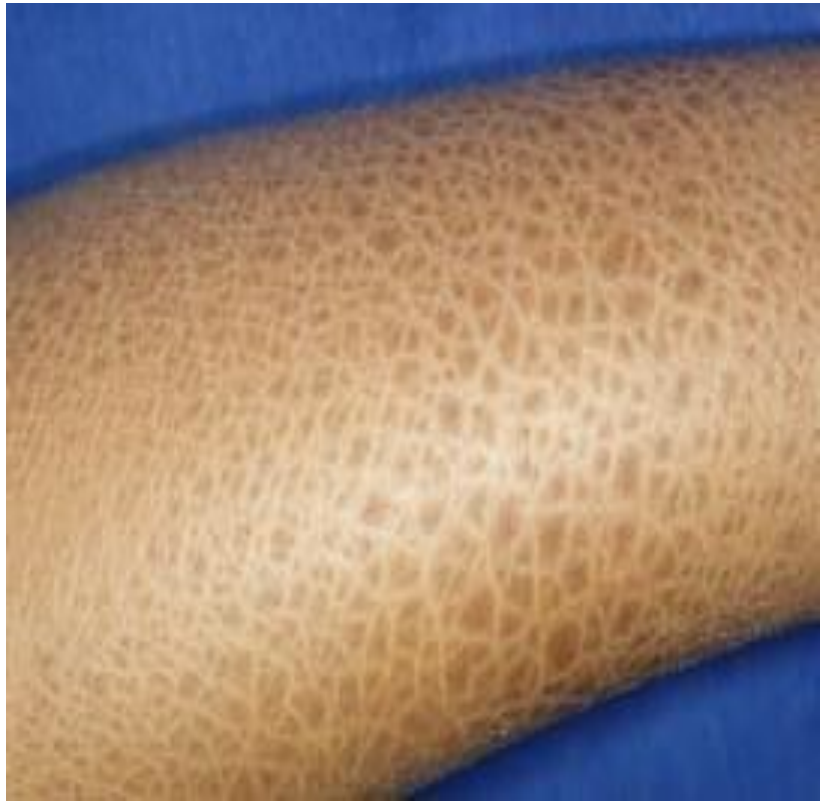
Armor plate like lesions, Ectropion and eclabium

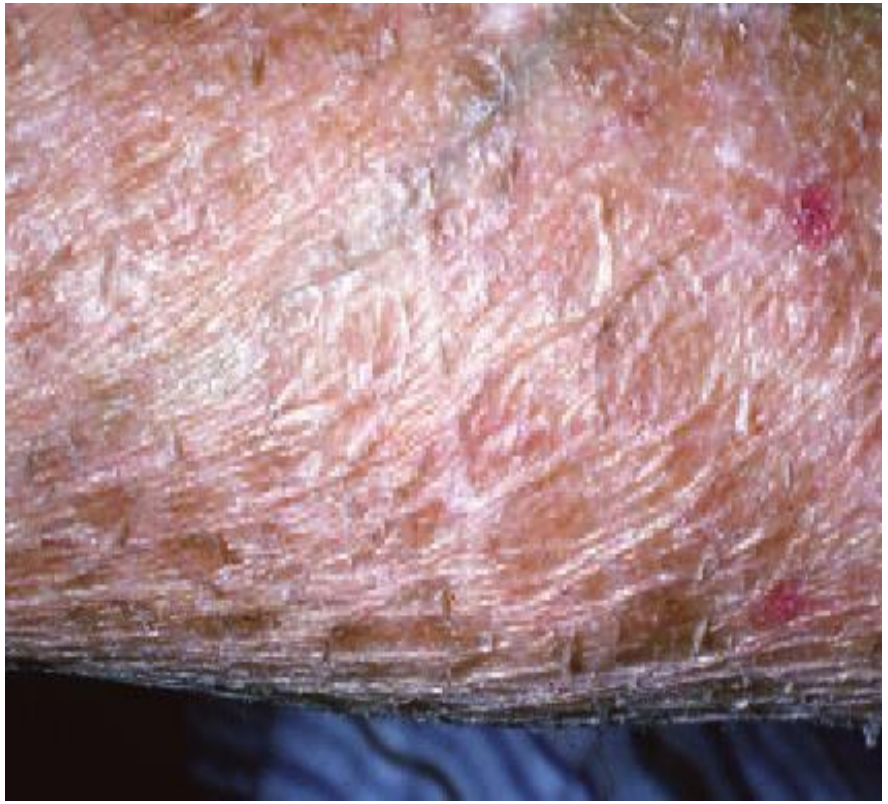
Acquired Ichthyosis

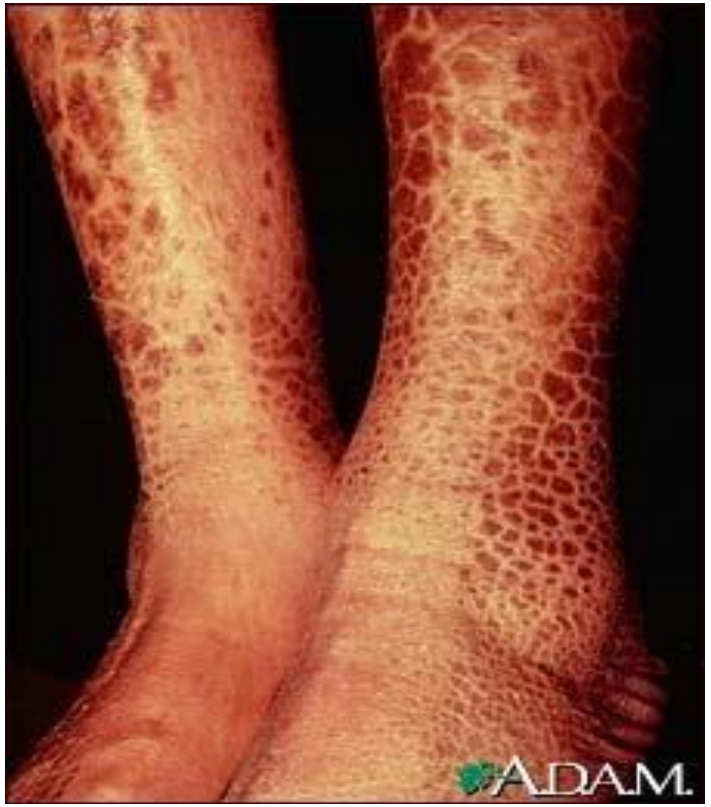
- Any patient presented with acquired Ichthyosis must be investigated to rule out these causes (Mention the causes of acquired Ichthyosis):

1. Sarcoidosis.
2. Polycythemia rubra Vera.
3. Cutaneous T cell lymphoma.
4. Leprosy.
5. TB.
6. Hodgkin`s lymphoma.
7. Lupus erythematosus.
8. Dermatomyositis.
9. Carcinomas.
10. Thyroid disease.

Acquired Ichthyosis







Dry, cracked, scaly skin

THANK YOU

يا ربّ برحمتك فرّج عن إخواننا
برحمتك أنجِّ المستضعفين من المؤمنين
أفرغ عليهم صبرًا ويقينًا وأنسًا ورحمة وحنانًا من لدنك
اجبرهم واربط على قلوبهم واستر عوراتهم وآمن روعاتهم
وعوّضهم
اجعلهم في ولايتك ومعيّتك وعنايتك وأطعمهم من جوع وآمنهم
من خوف
واشفِ جرحاهم وداوي مرضاهم وتقبّل وارحم شهداءهم ..
اللّهمّ كن مع إخواننا المجاهدين اغفر لهم ذنوبهم وإسرافهم في
أمرهم وثبت أقدامهم وانصرهم على القوم الكافرين
خُذ بأيديهم وسدد رميهم ورأيهم وبارك في قوتهم وقوّتهم وأنزل
عليهم مدداً من عندك
ثبّتهم وأخلص نيّاتهم واجعلها في سبيلك وإعلاء كلمتك، ونصرةً
للمستضعفين

يا منتقم انتقم من كل مُجرمٍ ظالم نكّل بعبادك ..
يا ربّ اشفِ صدورنا بانتقامك يا عظيم