GENODERMATOSIS

FARAJ A. ALSARRAH

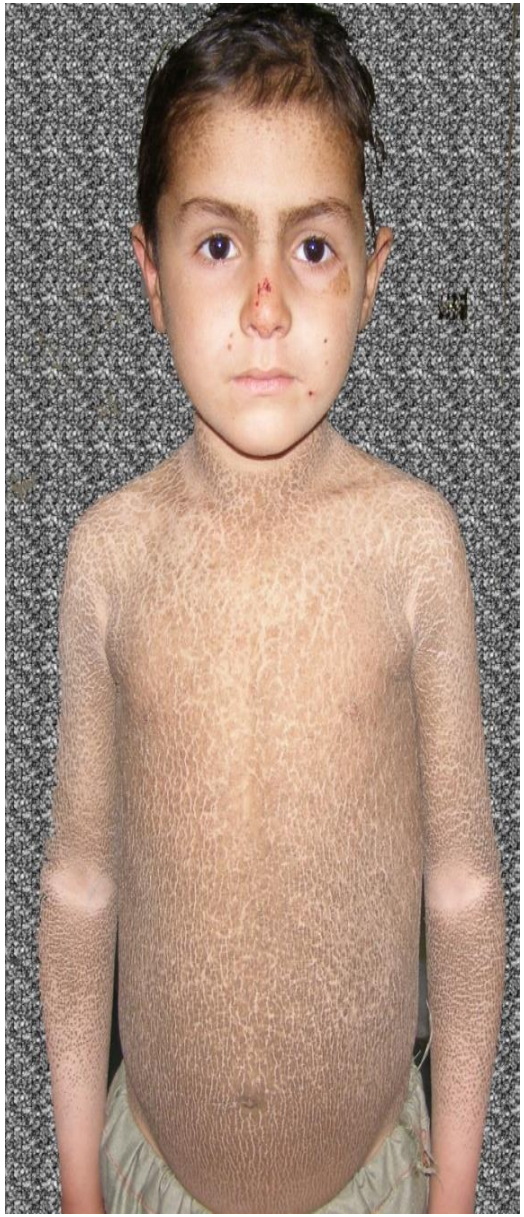
MBChB, MSc.

Department of Dermatology
Faculty of Medicine
Libyan international medical University

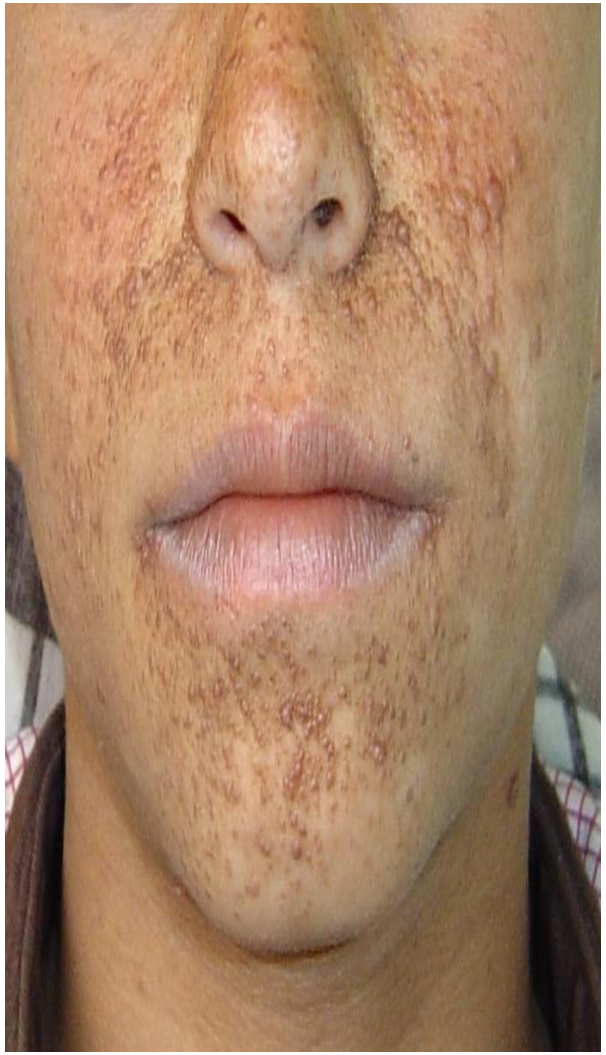
Genodermatoses

Are skin disorders caused by genetic chromosomal defects i.e. inherited, could be AD, AR or X-Linked.

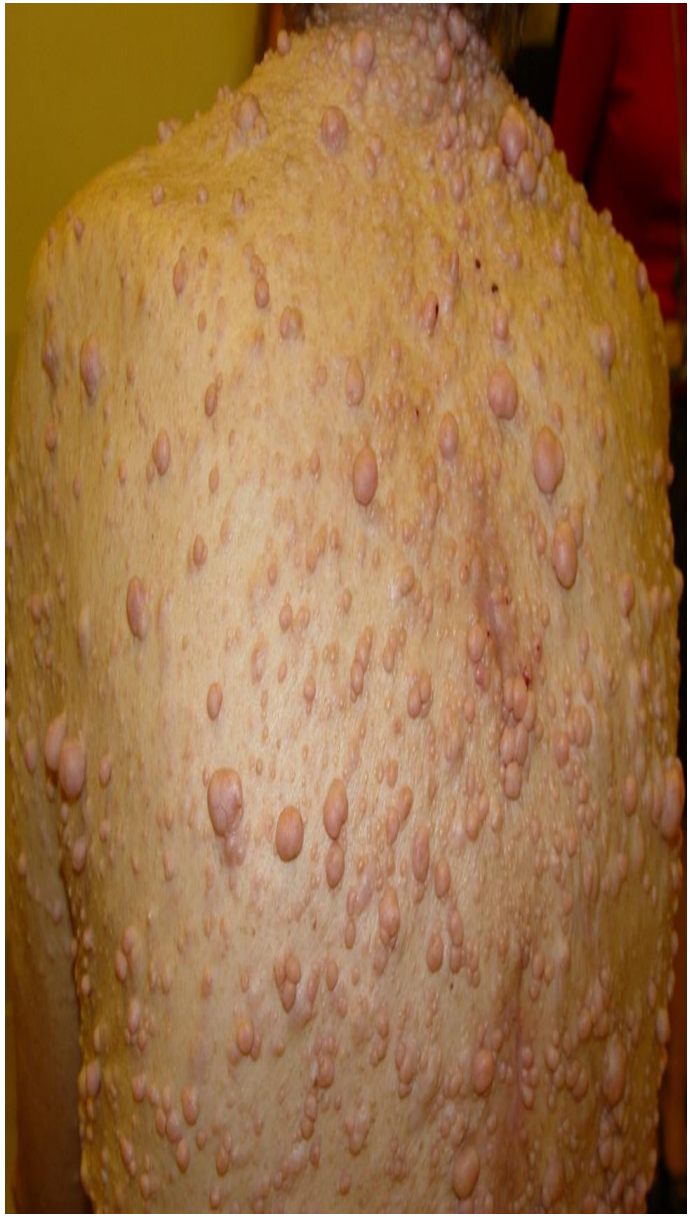
- **Autosomal dominant**: The affected individual has one copy of a mutant gene and one normal gene on a pair of autosomal chromosome.
- **Autosomal recessive:** the individual have two copies of the mutant gene.



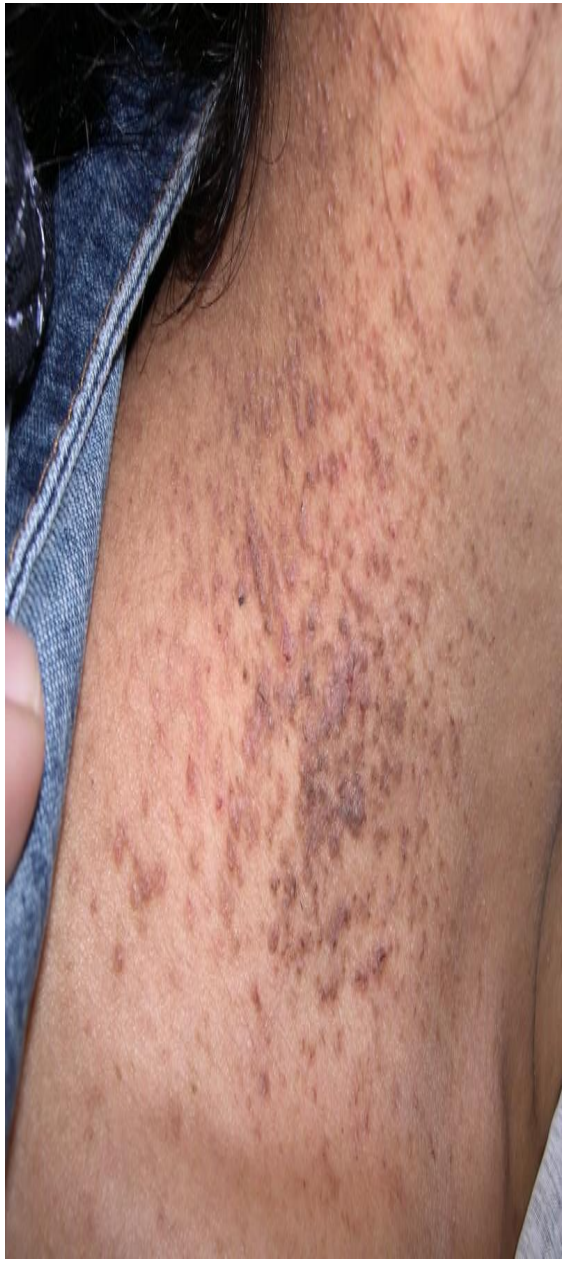
Ichthyoses



Tuberous Sclerosis

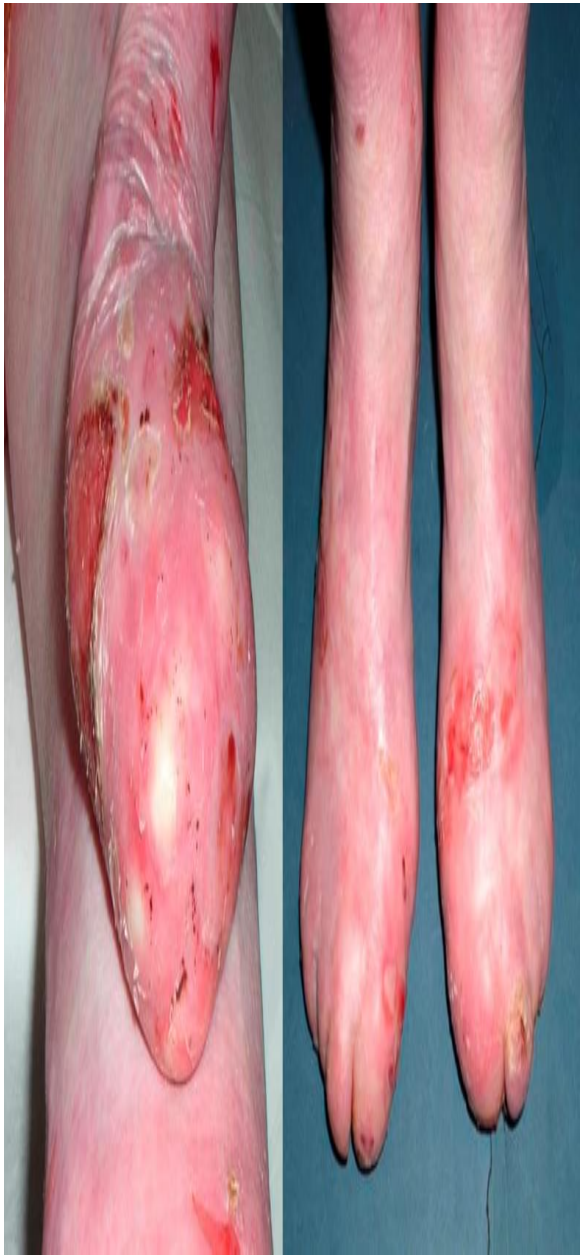


Neurofibromatosis

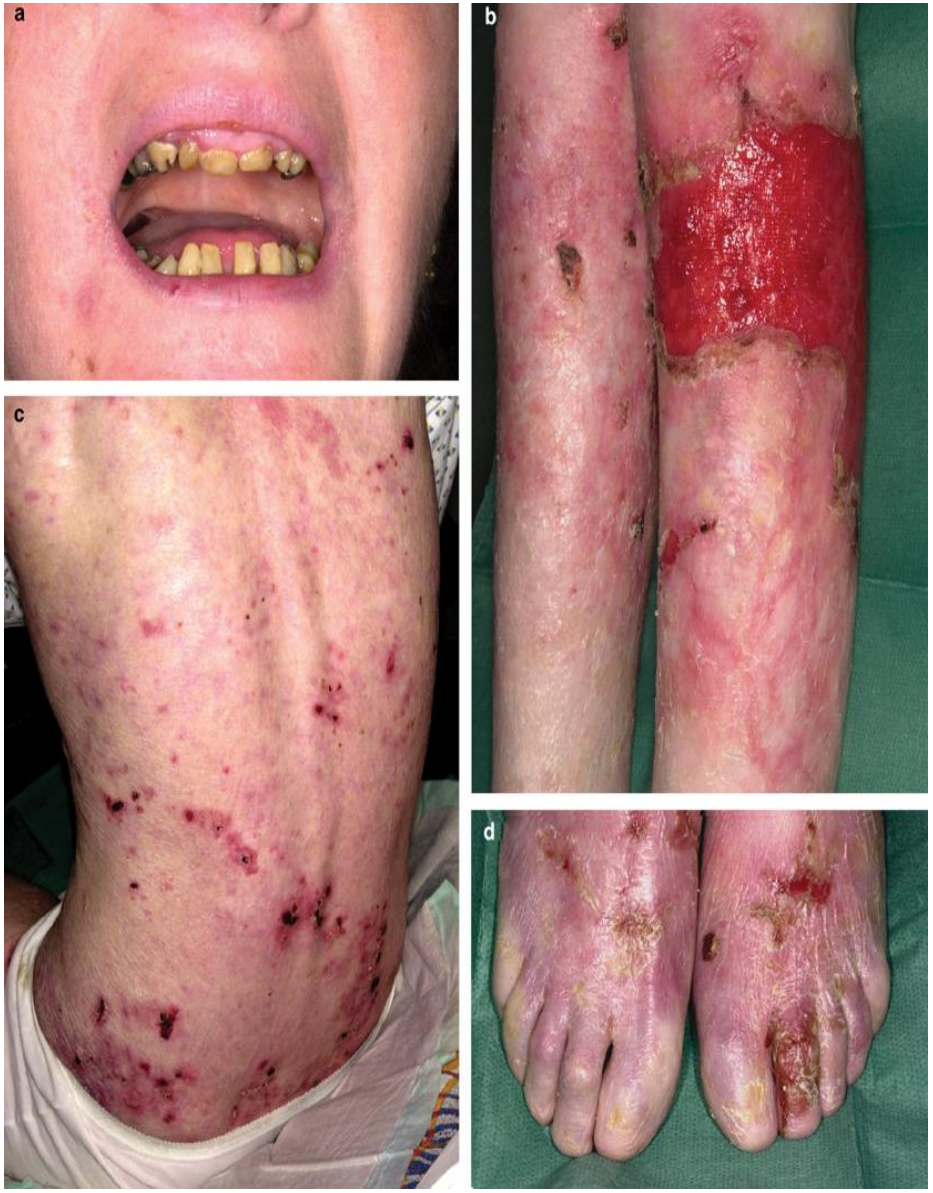


4 Darier Disease

Xeroderma Pigmentosum



Epidermolysis Bullosa



Epidermolysis Bullosa

Albinism

Genodermatoses

Are skin disorders caused by genetic chromosomal defects i.e. inherited.

1. **Ichthyoses.**
2. **Tuberous Sclerosis.**
3. **Neurofibromatosis.**
4. **Darier Disease.**
5. **Xeroderma Pigmentosa.**
6. **Epidermolysis Bullosa.**
7. **Albinism.**

Genodermatoses

The mode of inheritance could be autosomal or sex-linked depend on the locus of the defective gene.

Autosomal Dominant	X-linked	Autosomal Recessive
1.Ichthyoses vulgaris.	1.X-linked Ichthyoses.	1.Lameller Ichthyoses.
2.Tuberous Sclerosis.	2.Ocular Albinism.	2.Xeroderma Pigmentosa.
3.Neurofibromatosis.		3.Oculocutanous Albinism.
4.Darier disease.		4.EB Junctional.
5.EB Simplex.		5.EB Dystrophica.
6.EB dystrophica.		

Ichthyosis.

Group of genetically inherited conditions characterized by accumulation of visible scales on the skin surface.

Classified into 3 major forms:

1. **Ichthyosis vulgaris.AD**
2. **X-Linked Ichthyosis.**
3. **Lamellar Ichthyosis.AR**

Ichthyosis Vulgaris

AD

Childhood

Fine scale

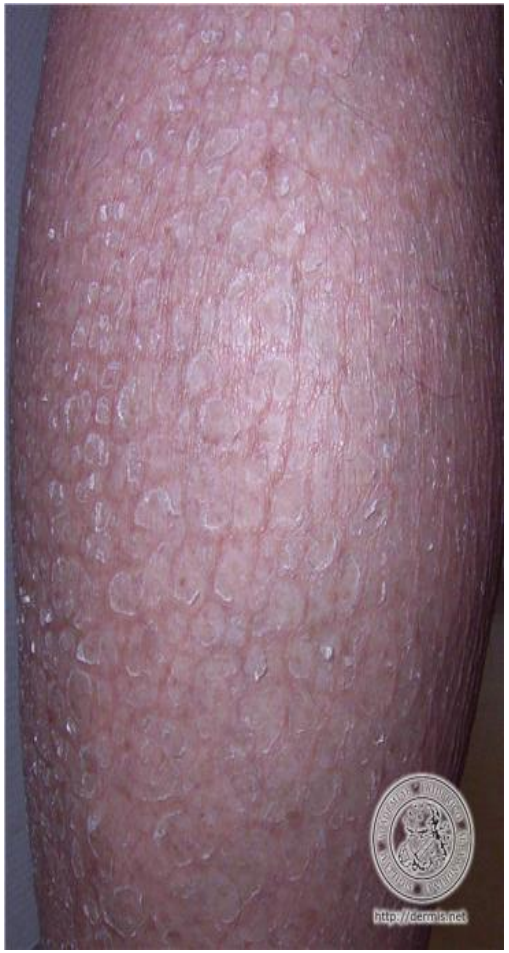
Extensor, palm,sole.

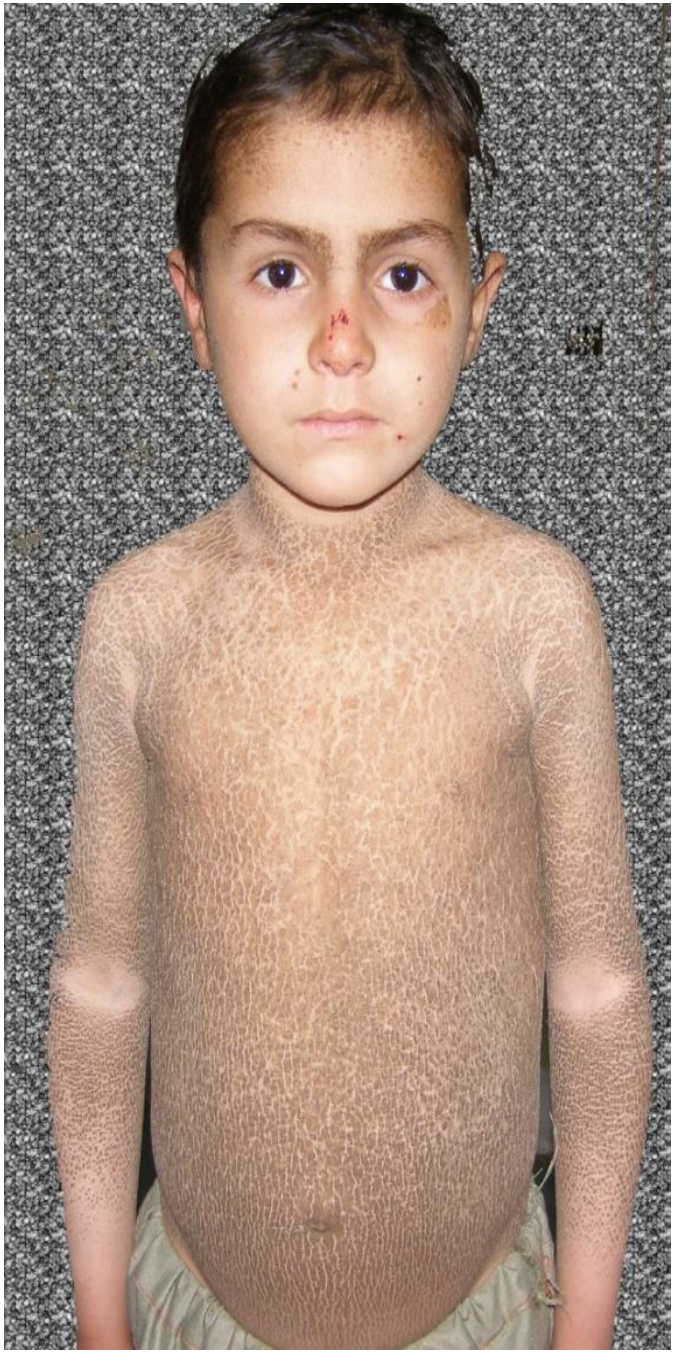
Ass; Atopy,

keratosis Pilaris,

palmoplanter hyperkeratosis.

Improve with age.





X-linked Ichthyosis

Infancy

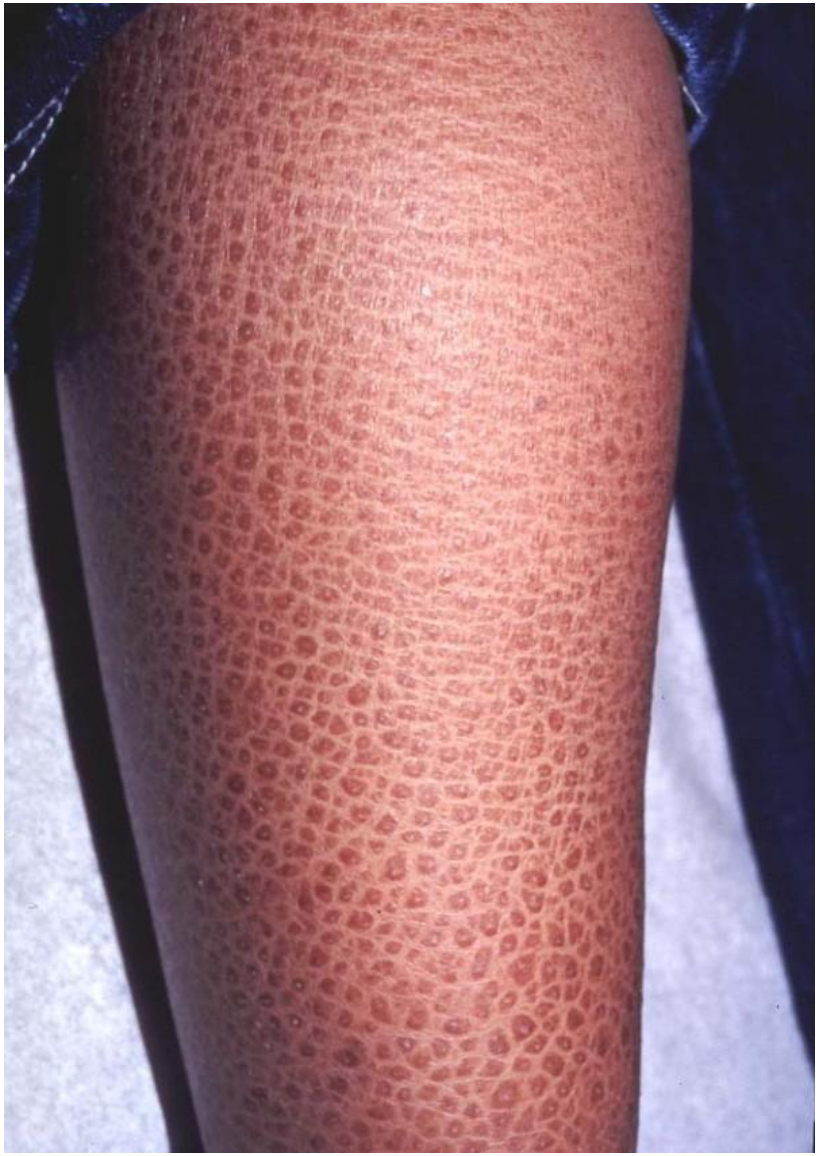
Coarse brown scales

spare palm, soles

Ass; corneal opacity, cryptorchidism.

Risk for testicular malignancy & ALL.

Worsen with age.



Lamellar Ichthyosis

AR, onset At birth
(Collodion baby).

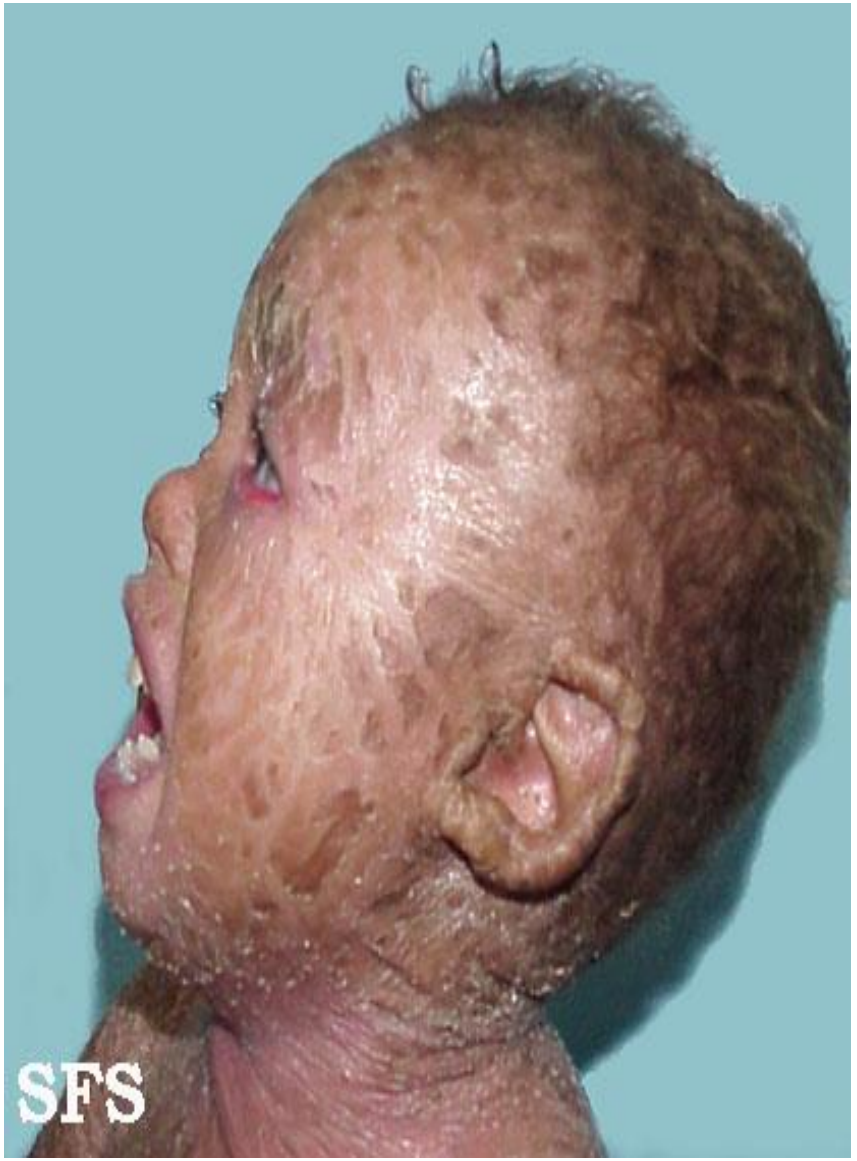
Scales are Dark plate
like, large rectangular.

prominent in LL.

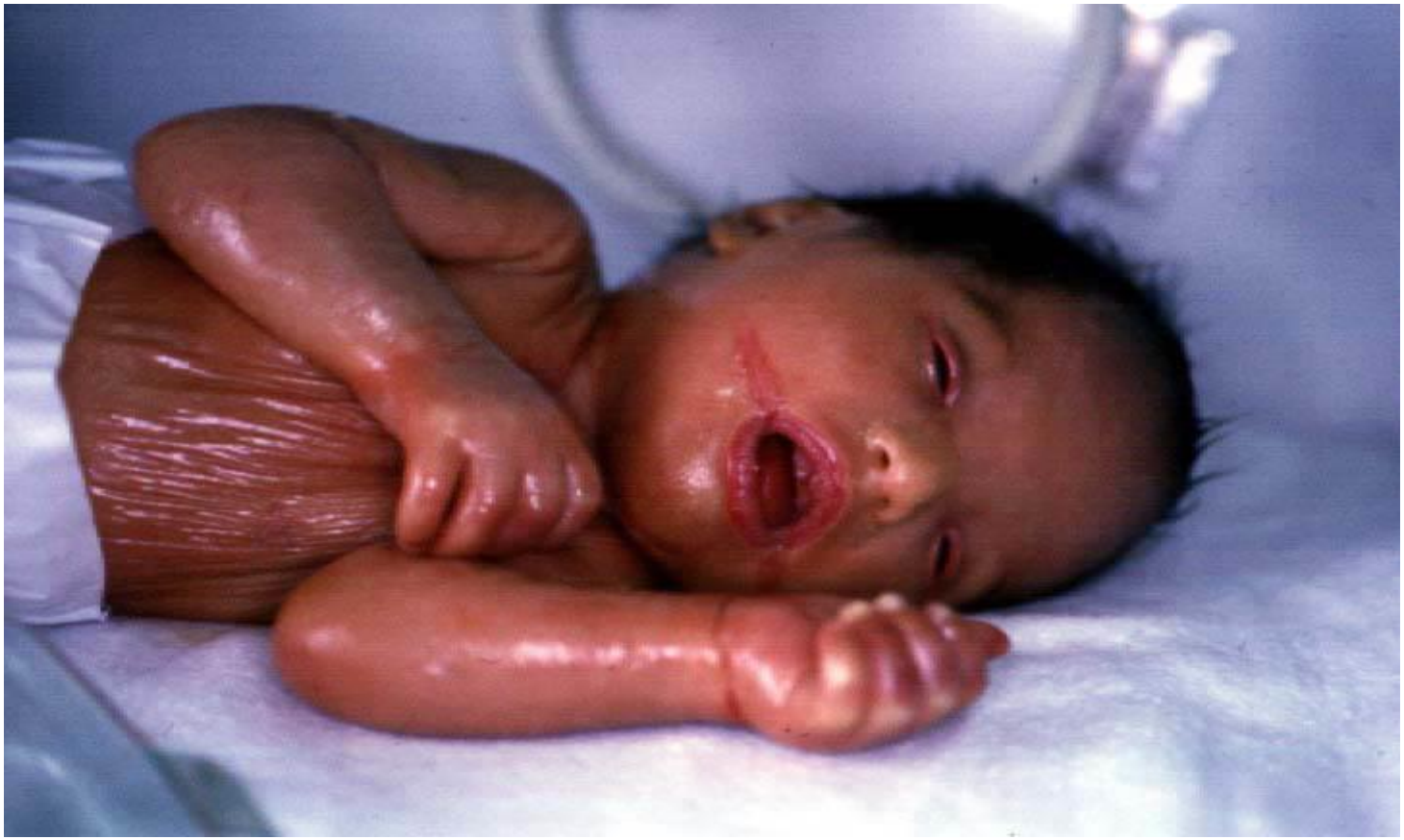
Ass; ectropion,

nail dystrophy &
Scaring alopecia.

persist



Collodion baby



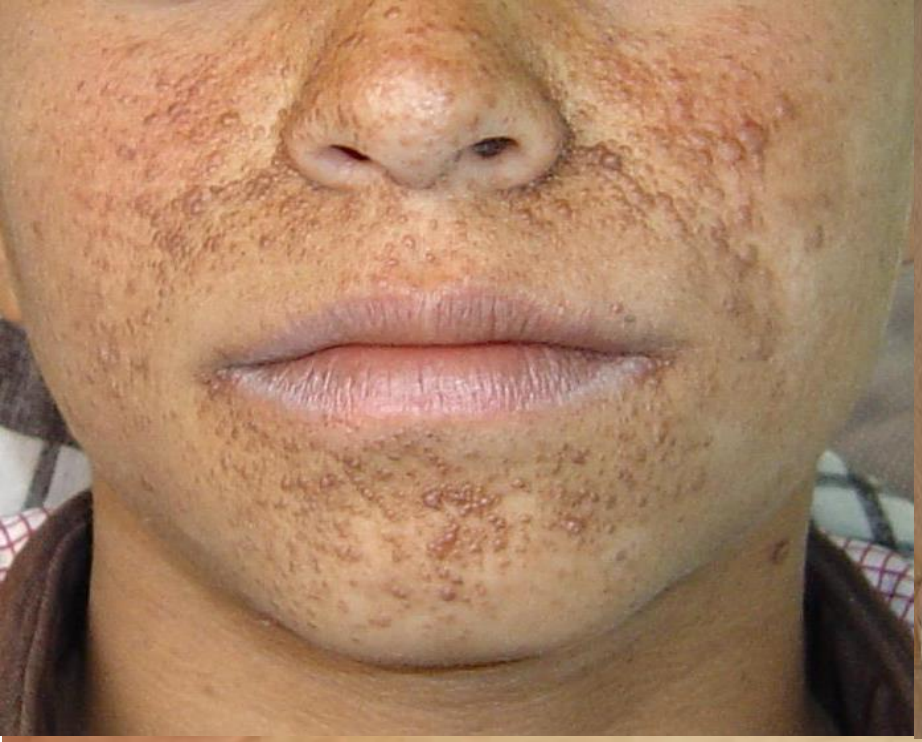
Lamellar icthyosis appear at birth & present throughout life, The newborn encased in a collodion membrane sheds ein 2 wks.

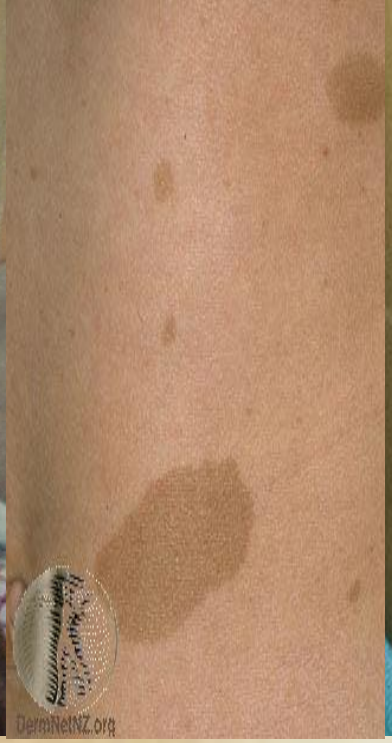
Ichthyosis types/ features	Ichthyosis Vulgaris	X-linked Vulgaris	Lamellar Ichthyosis
Inheritance	AD	X-linked	AR (Collodion baby)
Onset	Child	Infancy	birth
Scales	○ Fine scale ○ Extensor, palm & sole.	❑ Coarse brown scales. ❑ spare palm, soles	❖ Scales are Dark plate like, large rectangular. ❖ prominent in LL.
Association	Atopy, keratosis Pilaris, palmoplanter hyperkeratosis.	corneal opacity, cryptorchidism. Risk for testicular malignancy & ALL.	ectropion, nail dystrophy & Scaring alopecia.
prognosis	**Improve with age.**	**Worsen with age.**	**persist**

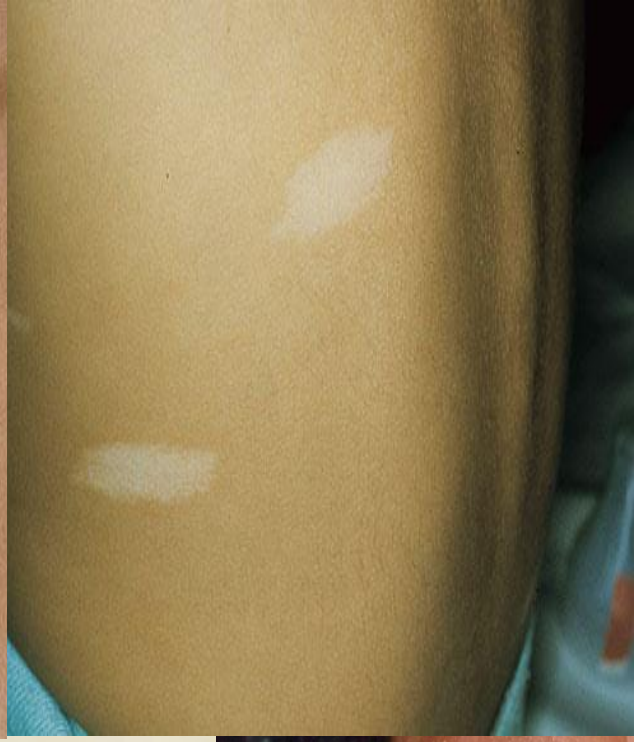
Tuberous Sclerosis (Epiloia)

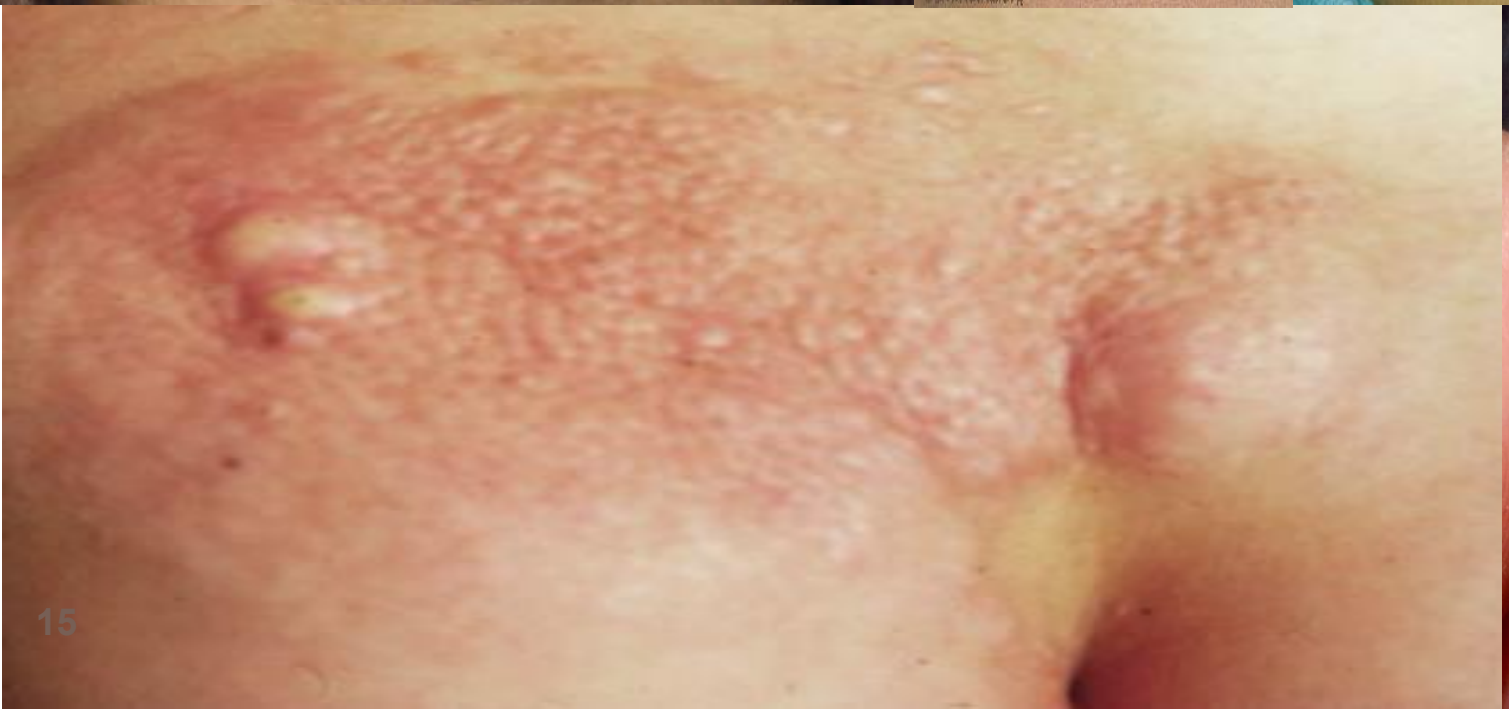
- AD, hereditary disorder of the skin & **internal organs**.
- CB triad; **E**pilepsy, **M**ental Retardation & **C**ut lesions.
- **The characteristic skin lesions are;**

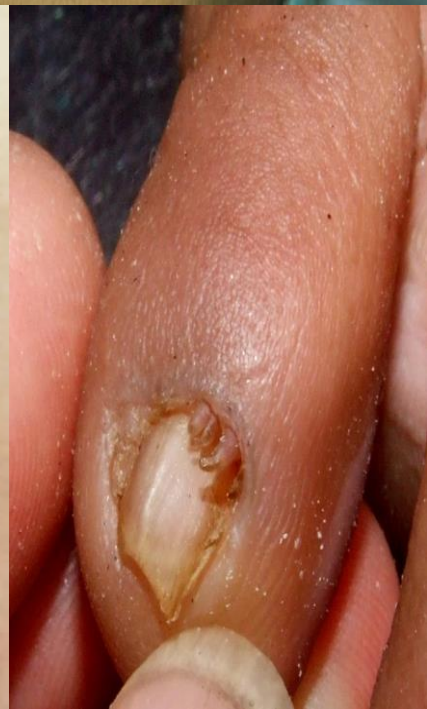
1. **A**denoma Sebaceum; symmetrically papular lesions on Naso-labial fold, cheeks & chin, (Angio-fibroma on histopathology).
2. **P**eri-ungual Fibromas ; Angio-fibroma in nails.
3. **A**sh-leaf Hypopigmented Macules; Earliest sign, At birth on trunk.
4. **C**afé-au-lait macules; appear after few months of life, need <6 in number for the Dx.
5. **S**hagreen patch; plaques in lumbosacral region(orange peel appearance).




DermNetNZ.org





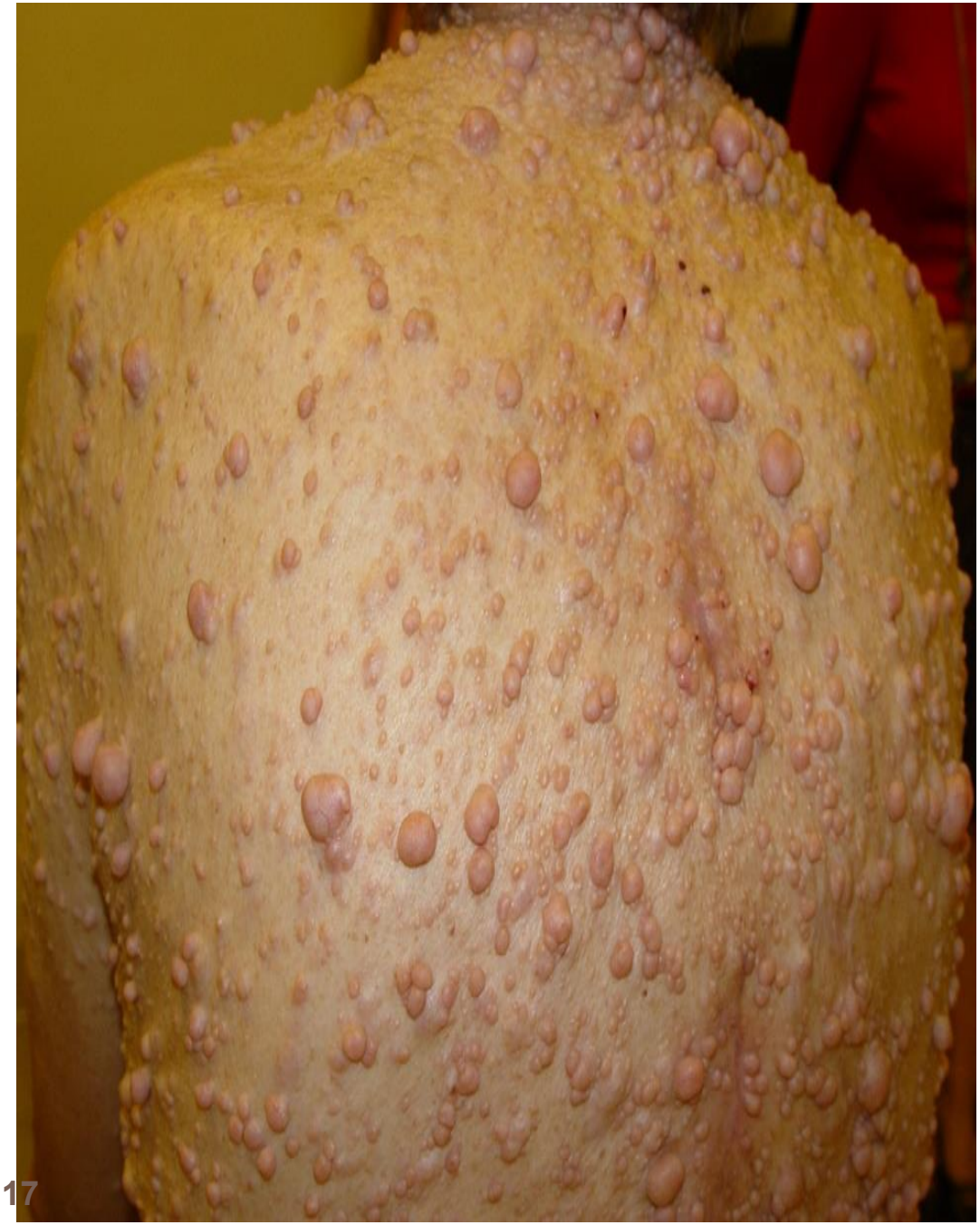


Neurofibromatosis
(Von Reckling hausen disease)

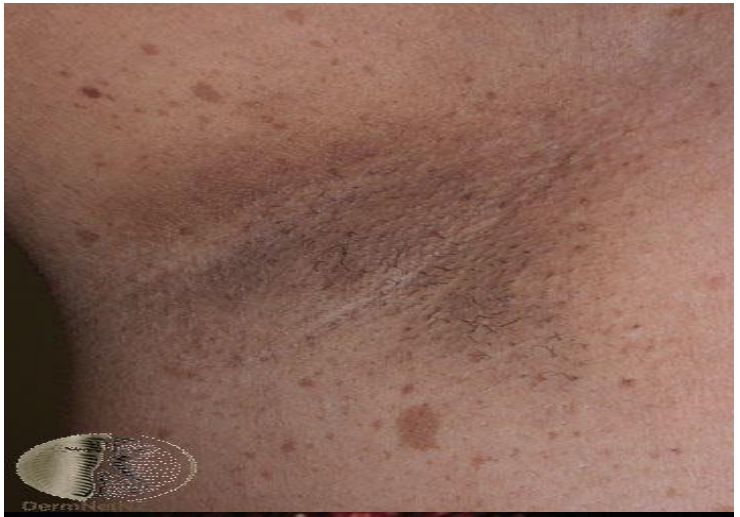
AD, CB; **C**utaneous changes, **L**isch nodules, **b**one abnormality & **V**arious **I**nternal **Tumors**.

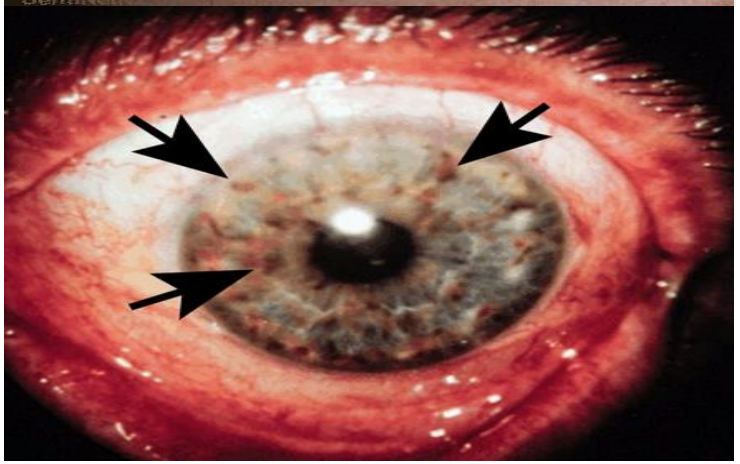
The Cutaneous manifestations are:

1. **C**afé-au-lait macules; for Dx of NF **Six** or more > 0.5 cm in children &1.5cm in adults is required.
2. **A**xillary freckles (Crowe`s sign).
3. **C**utaneous neurofibromas: on the trunk by pressure invaginate give button-hole sign. for Dx Two or more.
4. **P**igmented iris hamartomas (Lisch nodules), often identified only through slit-lamp examination by an ophthalmologist.

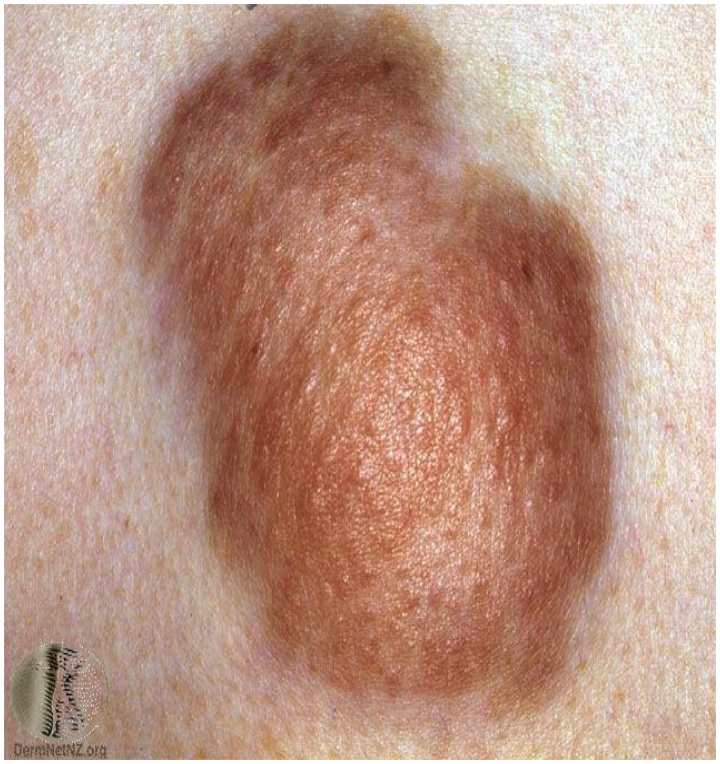


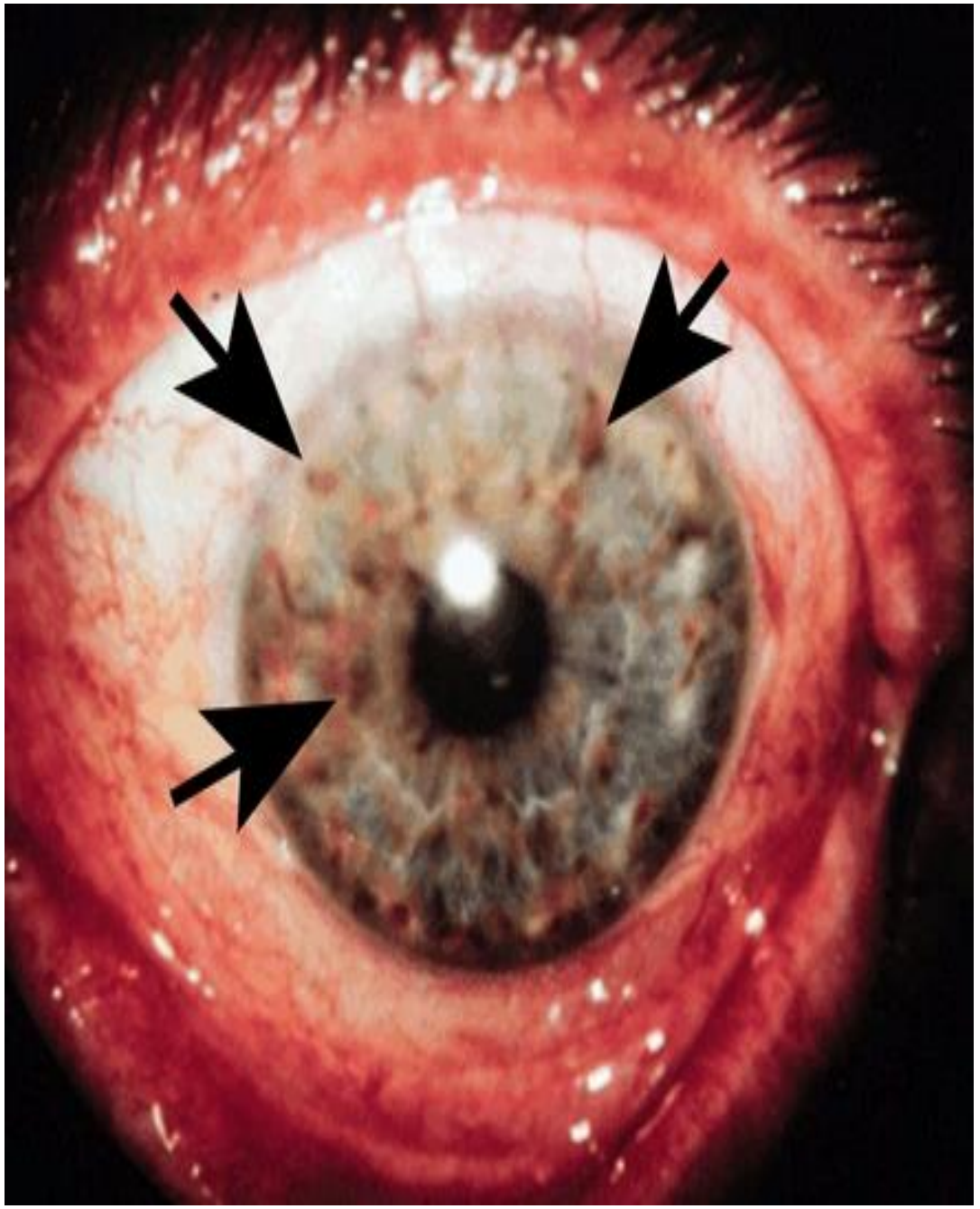

DermNetNZ.org


DermNetNZ.org



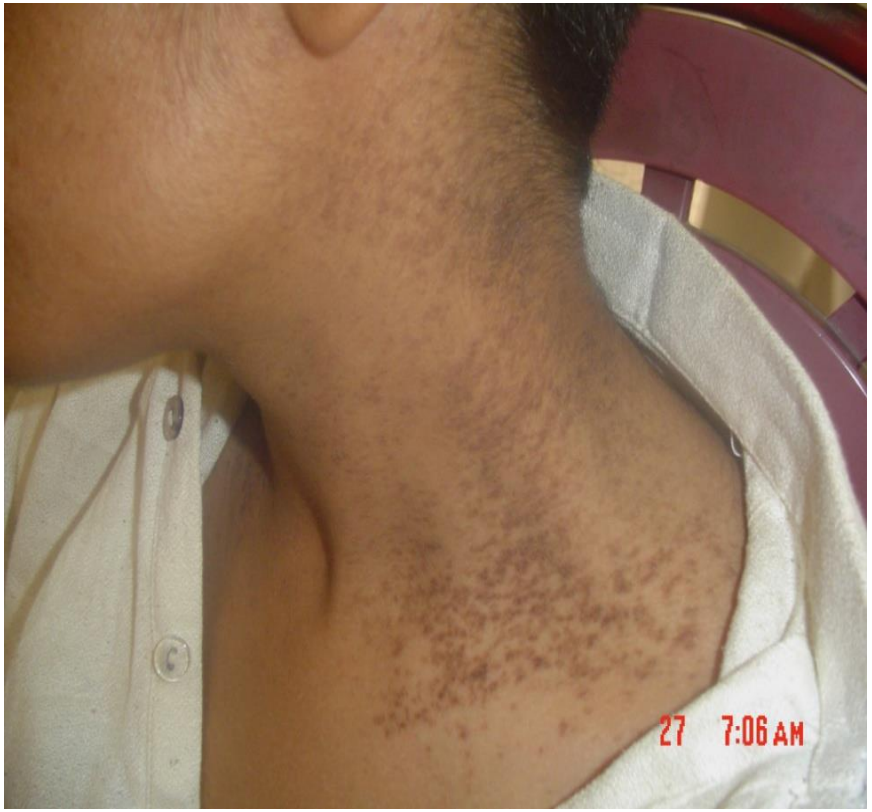
Neurofibromatosis

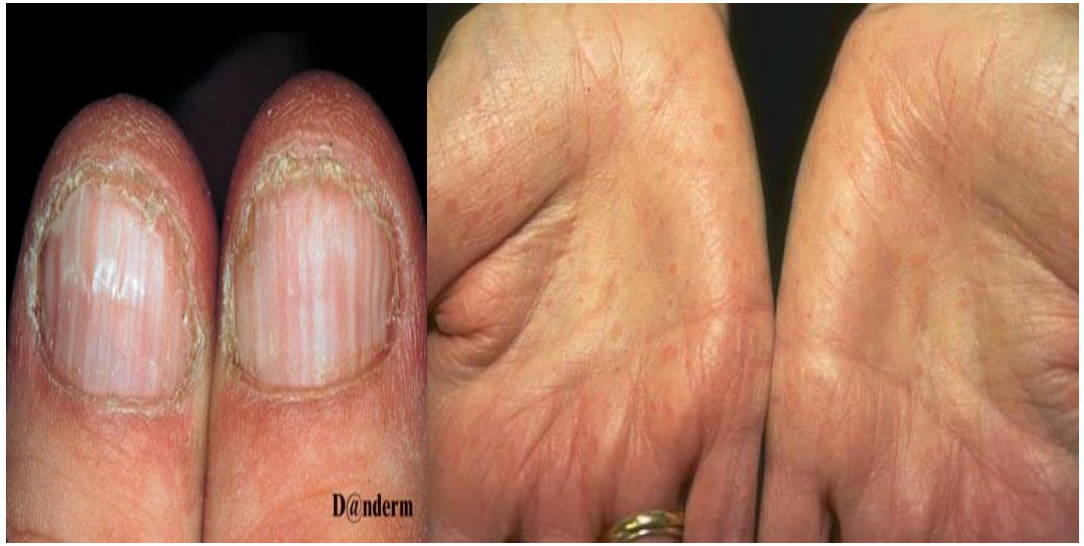


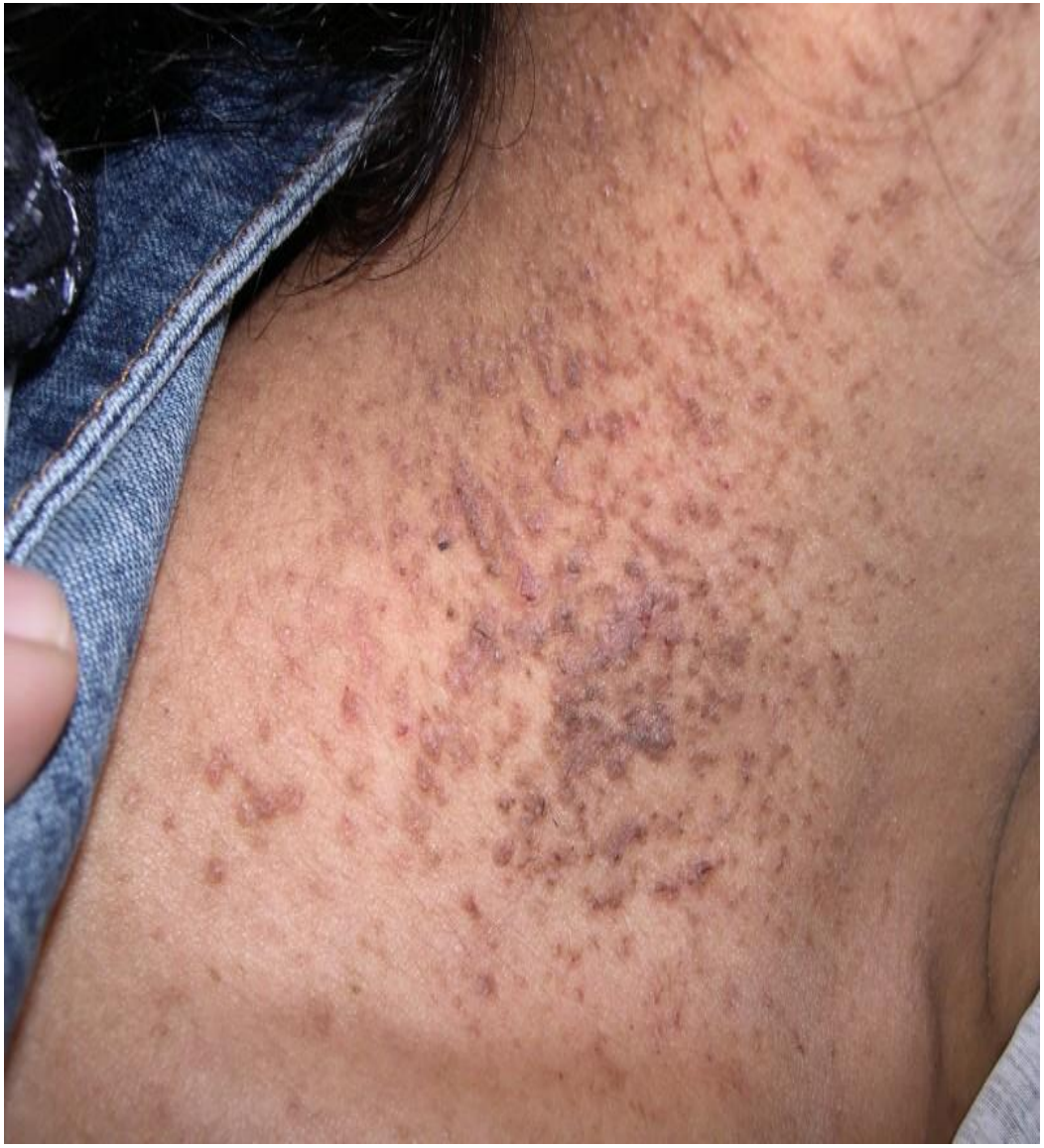


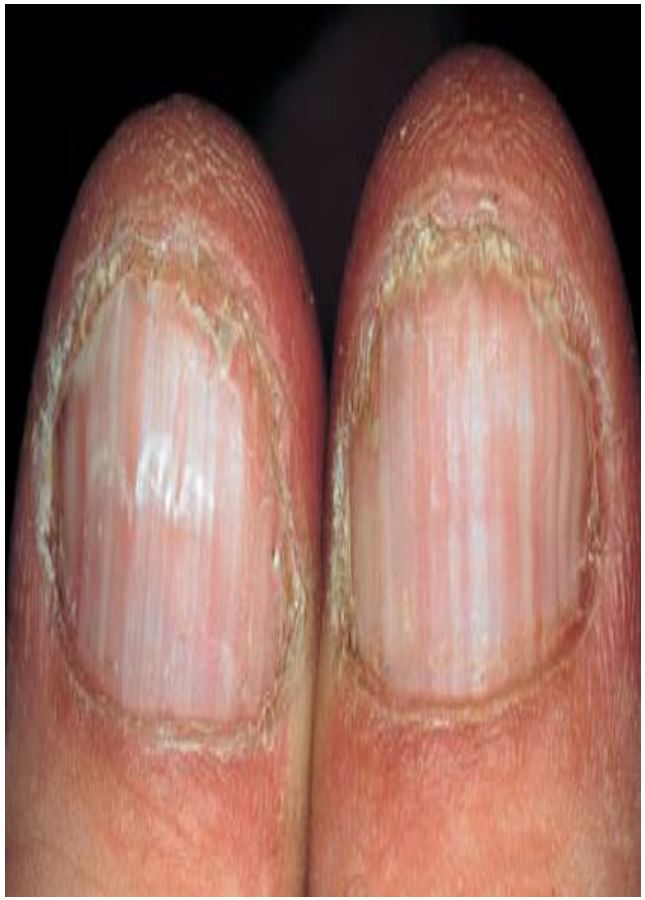
Dariers Disease

- AD, Chronic disorder of keratinization.
- Dirty skin color with yellowish brown papules in seb areas.
- Scales, crust.
- Pitting palms & soles.
- Nail: distal notch.
- Whitish papules in oral mucosa.
- Onset during puberty.
- Retinoids used as Rx.



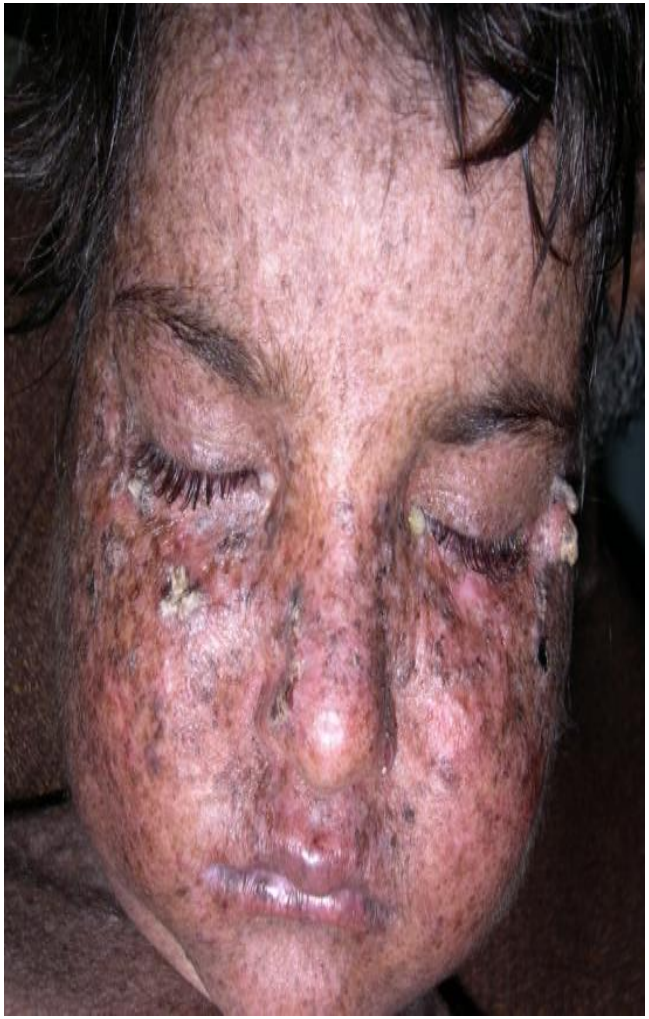






Xeroderma Pigmentosum(xp)

Rare AR disorder characterized by sever photosensitivity, photophobia and early development of cutaneous malignancy due to abnormality of DNA repair system.



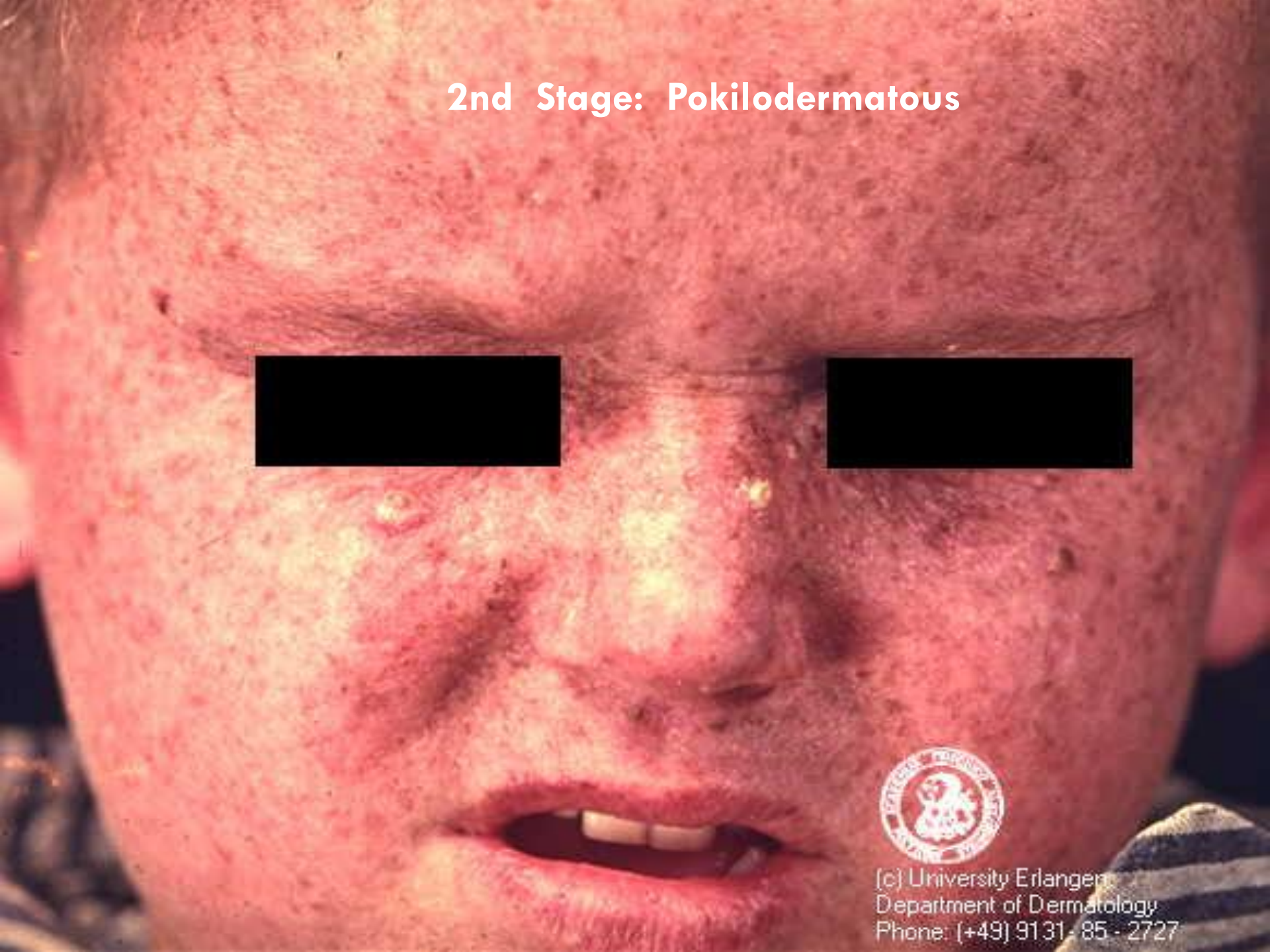
Clinical Picture :

1st Stage: Sever Photosensitivity & Photophobia.

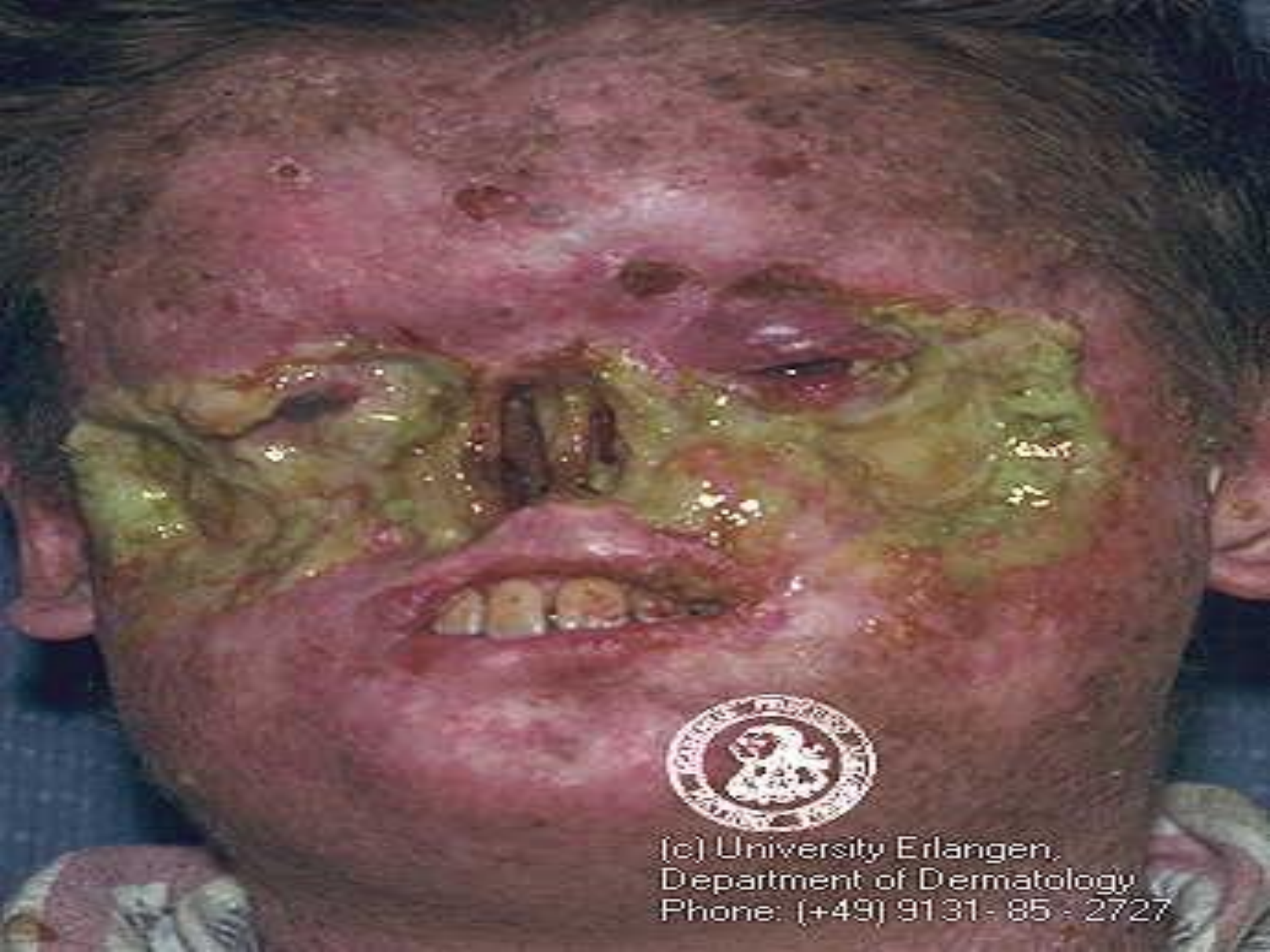
2nd Stage: Pokilodermatosis; freckling, mottled pigmentation, telangiectasia & skin atrophy.

3rd. Stage: Cut. Malignancy.

1st Stage: Photosensitivity


2nd Stage: Pokilodermatous
(c) University Erlangen
Department of Dermatology
Phone: (+49) 9131 - 85 - 2727

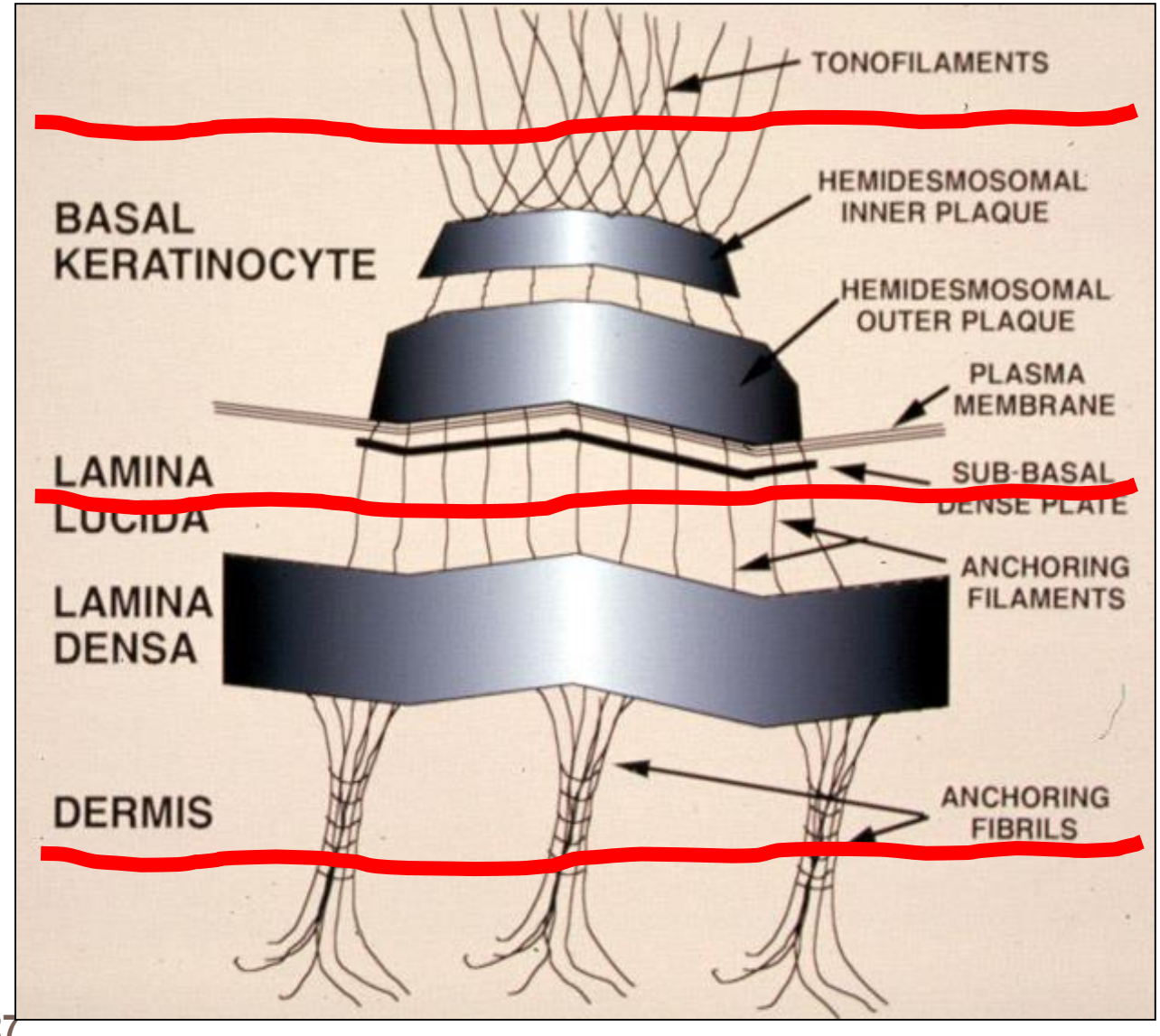

(c) University Erlangen,
Department of Dermatology
Phone: (+49) 9131 - 85 - 2727
3rd. Stage: Cut. Malignancy.



Epidermolysis Bullosa (EB)
(mechano-bullous diseases)

- Its a Group of inherited bullous disorders characterized by blistering of the skin & MM in response to minor or significant trauma.
- All share in; Genetic, Trauma & Blister formation.
- There are >20 types but according to ultra-Structural site of the blister they are classified into **3** major forms;
  1. EB **S**implex AD
  2. EB **J**unctional AR
  3. EB **D**ystrophic AR & AD

Epidermolysis Bullosa (EB)



EB SIMPLEX

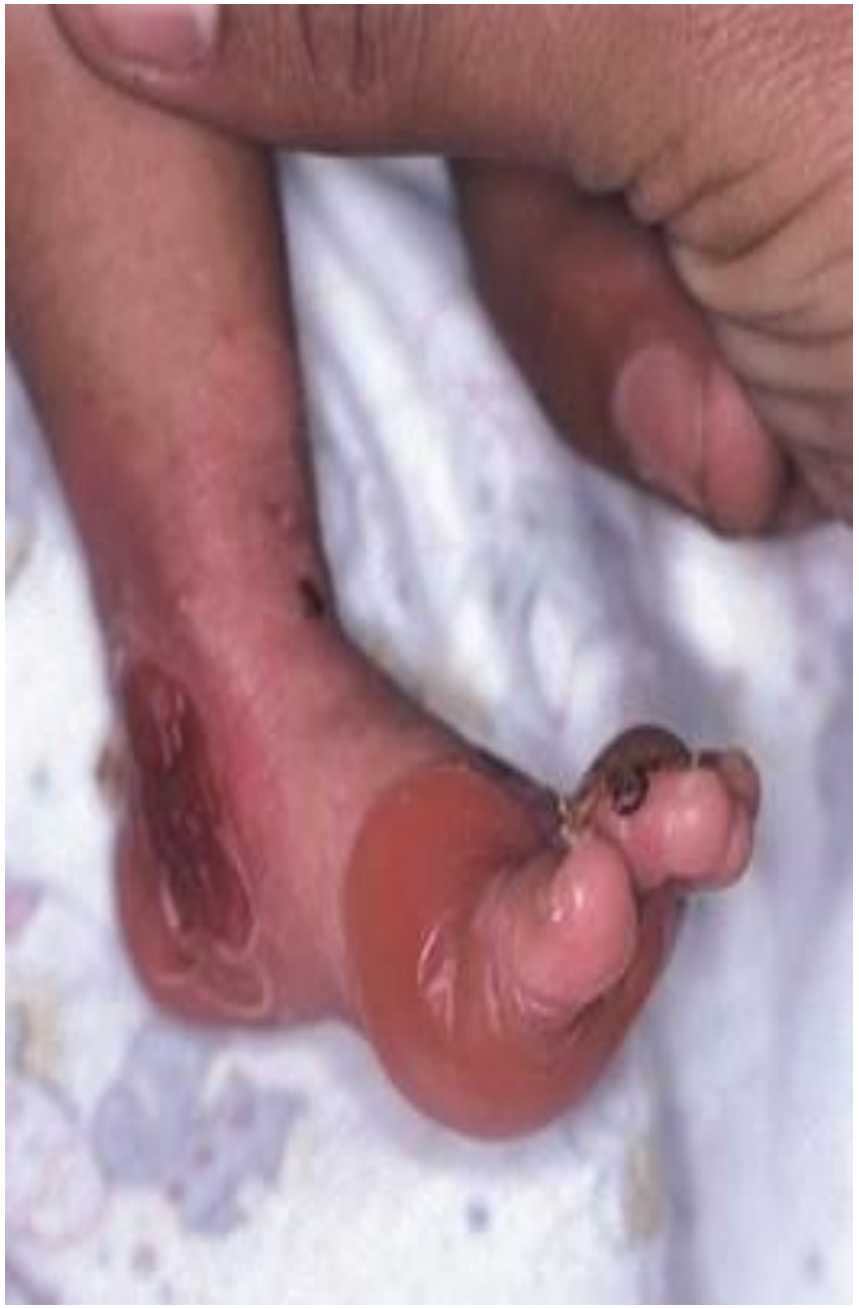
JUNCTIONAL EB

DYSTROPHIC EB

27

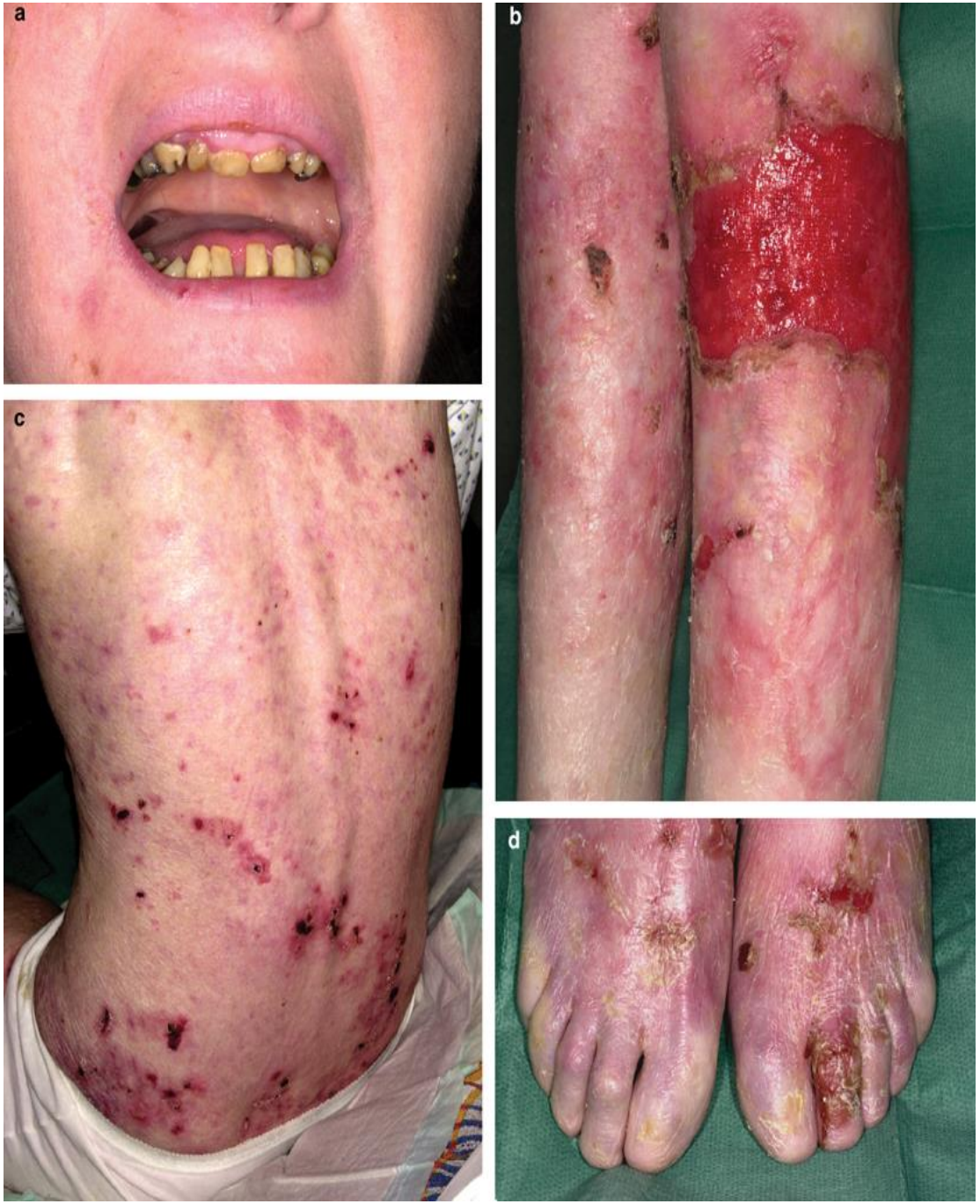
Epidermolysis Bullosa Simplex;

- AD.
- infancy & childhood.
- Site of blister HP; Intra-epidermal.
- good prognosis.



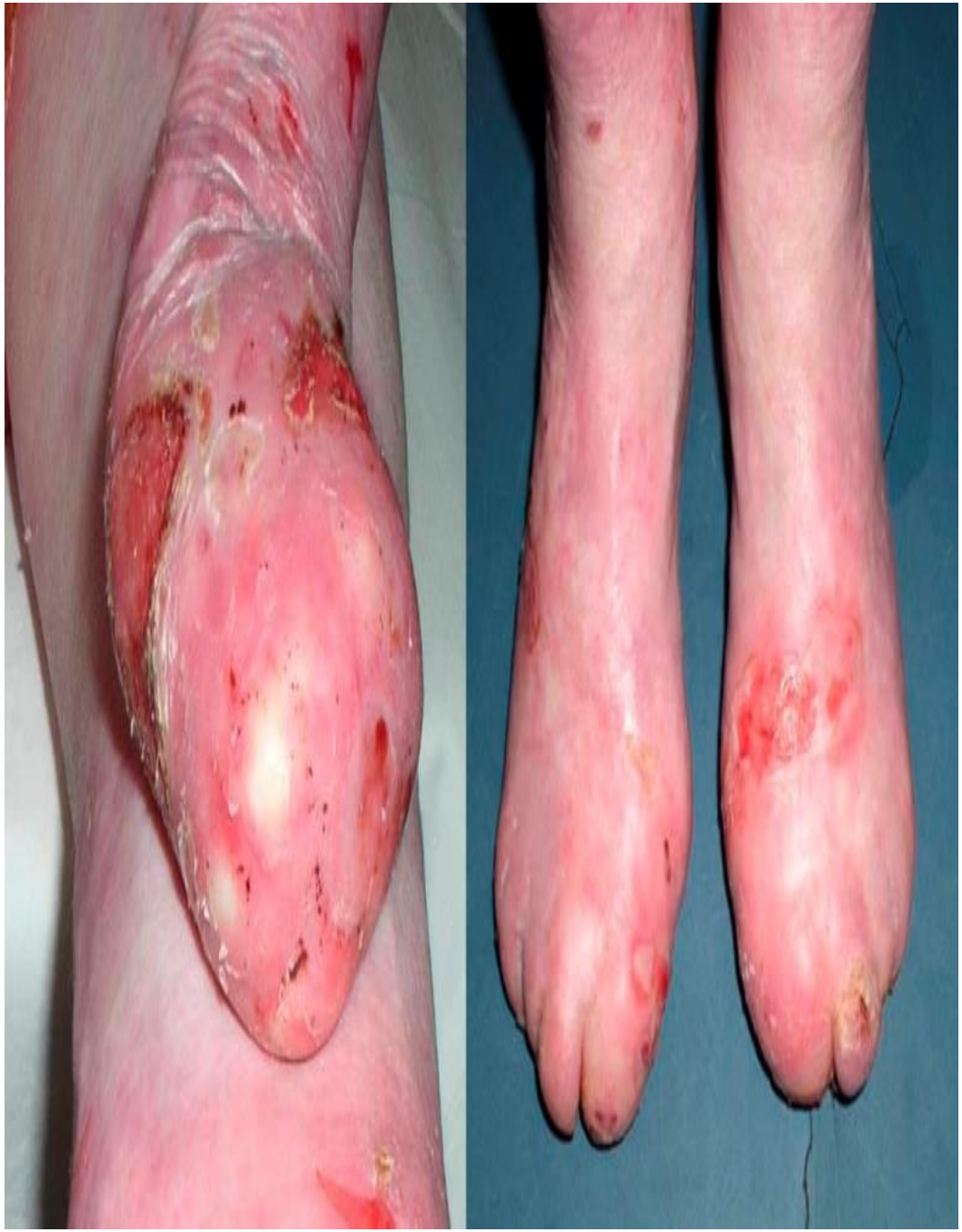
Junctional EB;

- AR.
- at birth.
- Site of blister HP; Basement membrane zone
- blister generalized.
- Poor prognosis.



Dystrophic EB;

- AD & AR.
- at birth.
- Site of blister HP; Anchoring fibers.
- Scaring & loss of function.
- Poor prognosis.

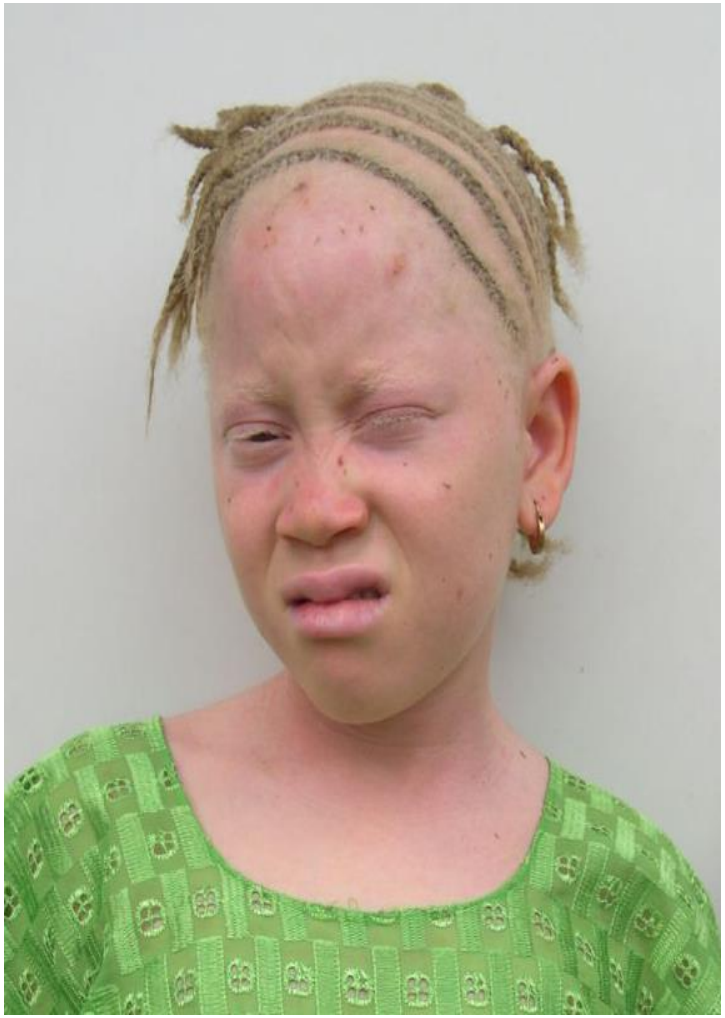


EB types/ features	EB simplex	EB junctional	EB Dystrophic
Inheritance	AD	AR	AD/AR
Onset	Infancy/child	birth	birth
Blister extend	Localized to friction	generalized	generalized
MM	Normal	involved	Involved
Teeth	N	Defected	N/defect
Hair & nails	N	Affected	Affected
Scarring	Absent	Absent ± present	Present
Site of blister HP	Intra-epidermal	Basement membrane zone	Anchoring fibers.
prognosis	good	poor	Poor

Albinism

Group of genetic disorders (AR) of melanin pigment synthesis within melanocytes of the skin, hair and eyes, in which there is complete or partial absence of pigment (defect in enzymes).

- Associated with photophobia, nystagmus and astigmatism.
- Lack of skin pigmentation makes it more susceptible to sunburn and skin cancers.



MCQ

Q; Regarding genodermatosis disorders?

a. Xeroderma Pigmentosum is AD disorder characterized by sever photosensitivity, photophobia and early cutaneous malignancy.

b. Ichthyosis Vulgaris is AD affecting the child & worse with age.

c. Ash-leaf macules & Adenoma Sebaceum are a features of Epiloia.

d. Crowe`s sign & button-hole sign are not seen in Neurofibromatosis.


Thank you!