ICHTHYOSIFORM DISORDERS

ICHTHYOSES & ERYTHROKERATODERMAS

- Disorders of cornification
- Abnormal differentiation and desquamation of the epidermis result in a defective epidermal barrier.
- Clinically all ‘Ichthyoses’ have generalized scaling of the skin
- Erythrokeratodermas: circumscribed areas of erythema and hyperkeratosis without obvious scaling.

ICHTHYOSES

- Disorder of keratinization that manifests as dry and scaly skin resembling ‘fish skin’
- Ichthys=fish

- Two broad types

1. Defects in keratin or intercellular substances(lipids, fillagrin) , limited to skin
2. Metabolic disease with involvement of other organs

TYPES OF ICHTHYOSES

Congenital ichthyoses

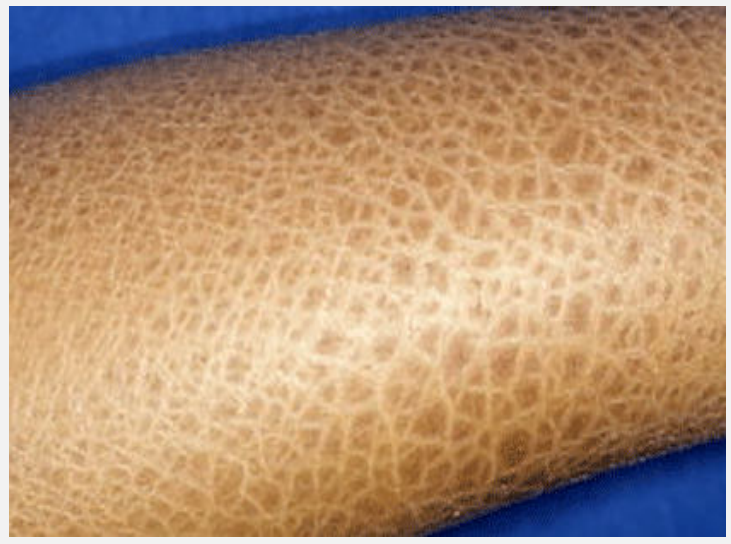
- Ichthyosis vulgaris
- X linked ichthyosis
- Lamellar ichthyosis
- Epidermolytic ichthyosis
- Non Bullous Ichthyosis erythroderma
- Epidermolytic ichthyosis (bullous CIE)
- Superficial epidermolytic ichthyosis (ichthyosis bullosa of Siemens)
- Ichthyosis hystrix Curth–Macklin

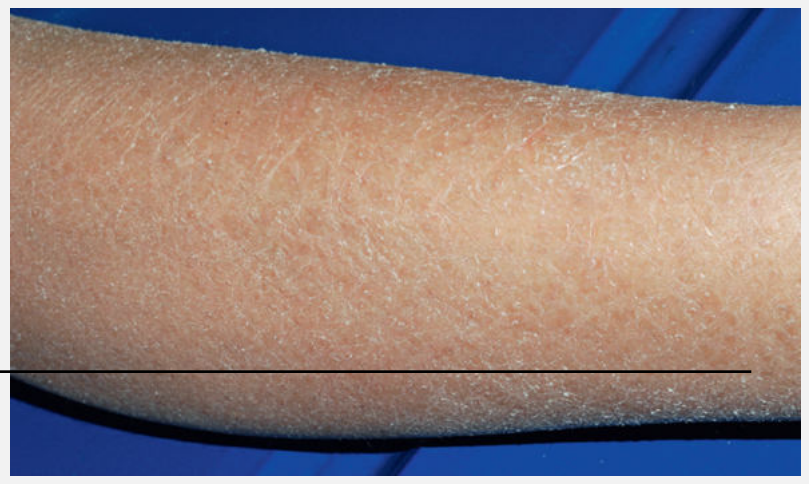
ICHTHYOSIS VULGARIS

- Autosomal dominant with high penetrance
- Molecular defect: Fillagrin deficiency in epidermis
- Resulting in retension hyperkeratosis
- Age of onset : 1-4 yrs
- Prevalence : 1 in 300
- Improvement in summer
- Lesions persists life long

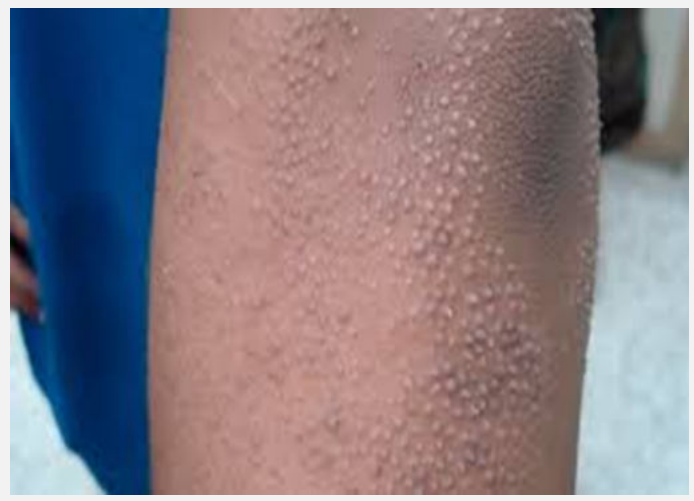
ICHTHYOSIS VULGARIS

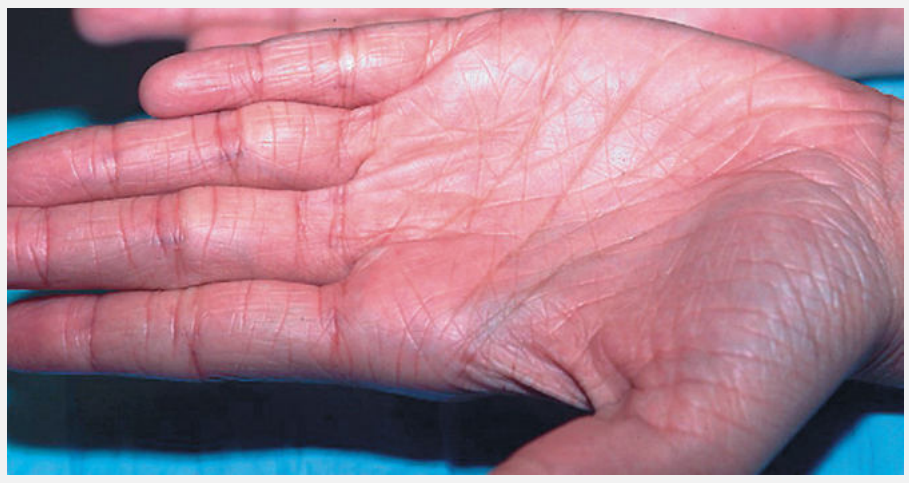
- Scaling : generalized
- Dirty brown polygonal scales: legs
- Fine, adherent scales on trunk, extremities
- Face, flexures spared
- Asymptomatic
- Asso. Features-
  - increased palmer lines, furrowed heels
  - Keratosis pilaris
  - Atopic dermatitis





- Treatment
  - Emolients , glycerine
  - 10 % Urea cremes
  - Kerolytic cremes with lactic and salicylic acid
  - Systemic acitretin rarely used

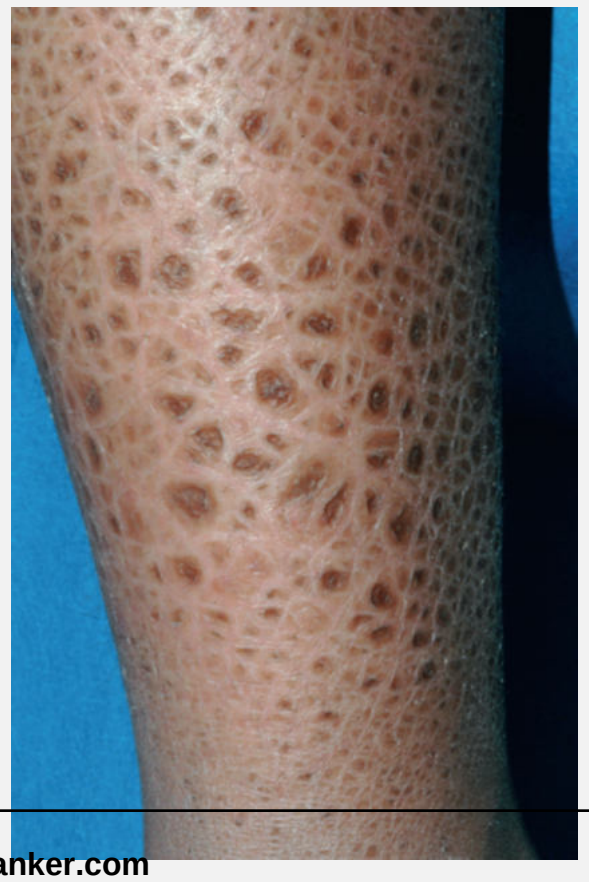




X LINKED RECESSIVE ICHTHYOSIS

- X linked recessive
- Males (females carrier)
- Defect : Steroid sulfatase deficiency
- Age in onset: infancy
- Fine to large, dark, adherent scales
- Extremities, trunk, neck and lateral face
- Preauricular area involvement: pathognomic
- Involves flexures
- Neck:almost invariably involved, “dirty neck”.
- Palms, soles spared

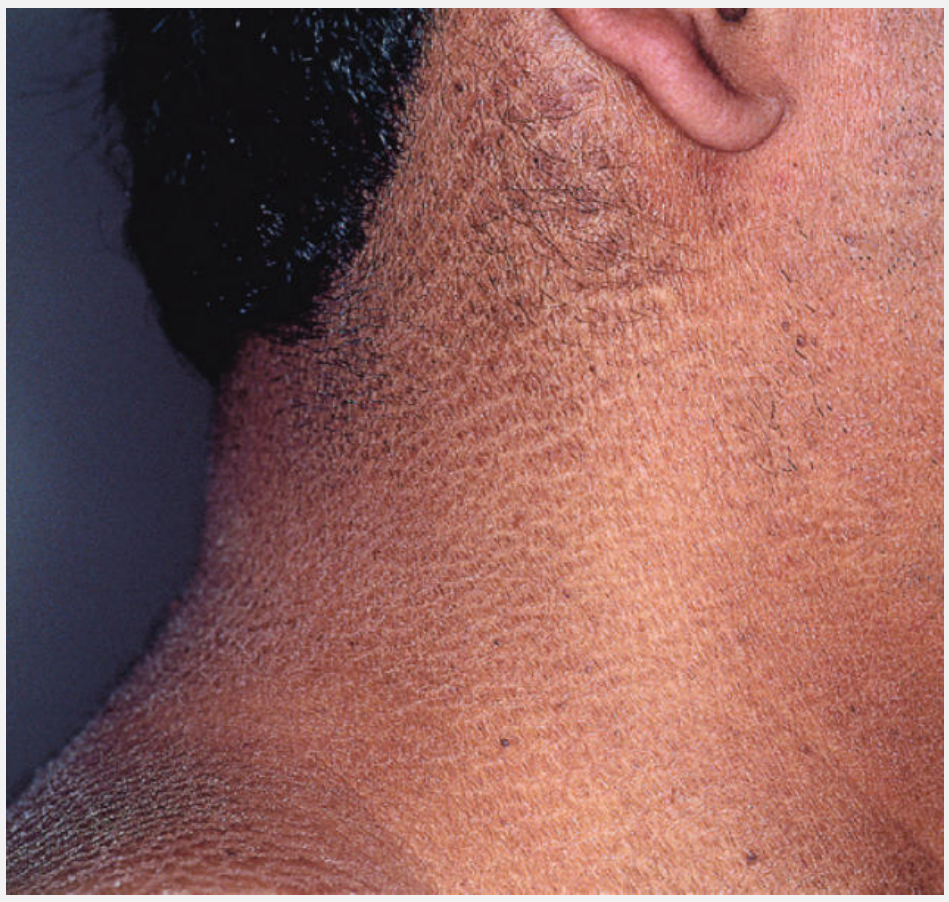
Prominent dark-brown scales - lower legs.
Smaller dark-brown scales -trunk, with sparing of skin folds



X LINKED RECESSIVE ICHTHYOSIS

- Asso. Features
- Cryptorchidism
- Corneal opacity
- Failure of spontaneous labour in carrier females
- Topical
  - Humectants- Propylene glycol
  - Keratolytics – salicylic acid
  - Retinoids
- Systemic Retinoids – rarely necessary

Dark scales on the neck, "dirty neck".

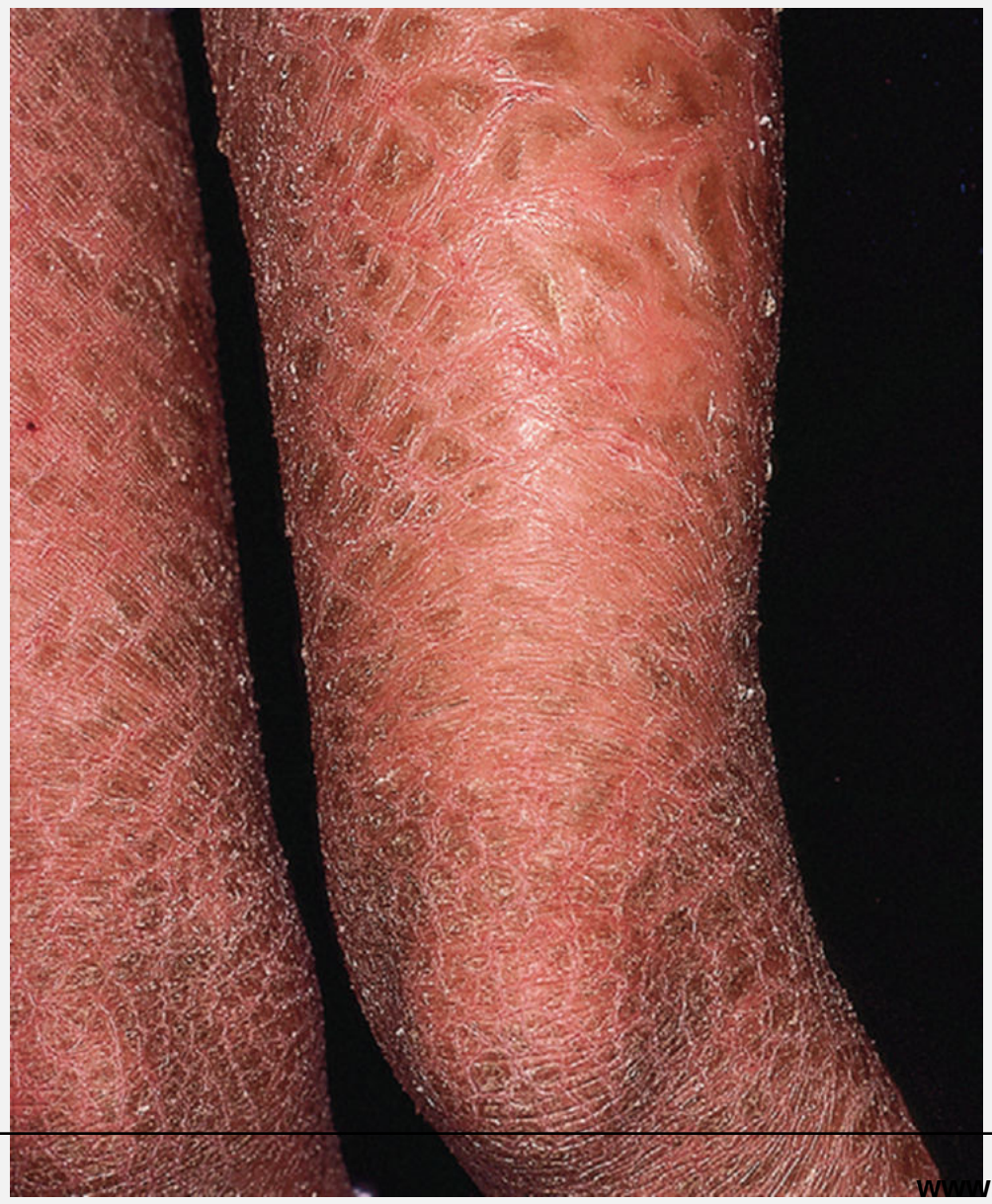


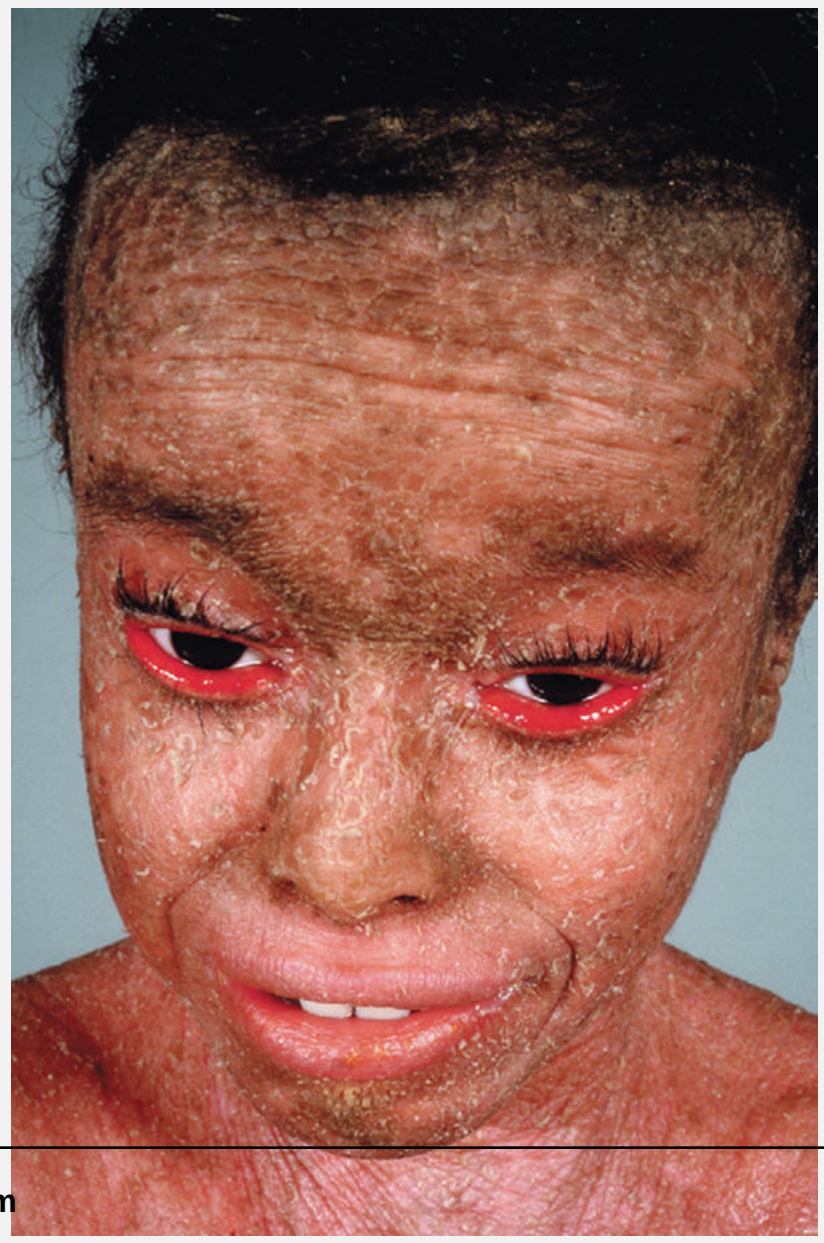
LAMELLAR ICHTHYOSIS

- Autosomal recessive
- Transglutaminase-1 deficiency
- This enzyme is expressed in upper layers of the epidermis,
- facilitates formation of the cornified cell envelope by cross-linking numerous structural proteins to the lipid envelope.
- Age of onset: Birth as colloidion baby
- Persists throughout life

LAMELLAR ICHTHYOSIS

- Over the first weeks , the collodion membrane is replaced by generalized scales.
- Scales are large, brown, plate-like:mosaic or bark-like pattern
- scales are centrally attached with raised borders, often leading to superficial fissures.
- minimal to no associated erythroderma
- Asso.
  - Heat intolerance : Intraepidermal constriction of sweat ducts
  - Scarring alopecia,
  - Eclabium
  - Severe ectropion - madarosis, conjunctivitis, incomplete lid closure with ensuing keratitis.
  - Palmoplanter keratoderma



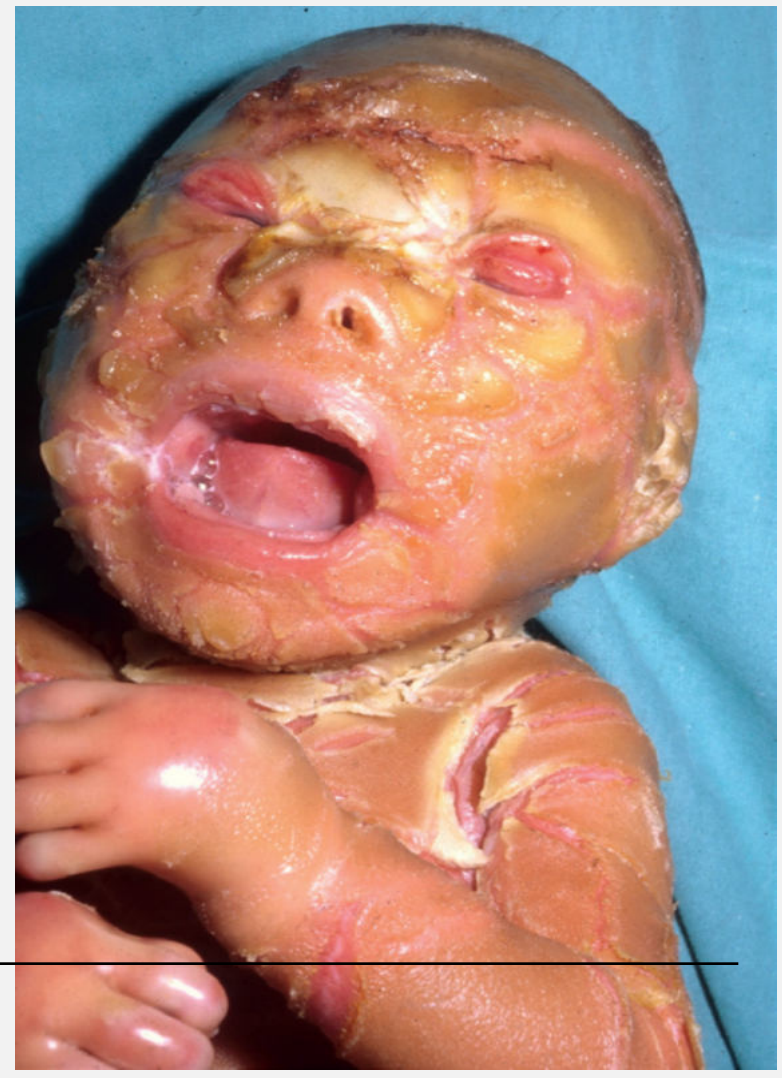


LAMELLAR ICHTHYOSIS

- Neonatal care as in collodion babies.
- Oral retinoids: Acitretin from early childhood.
- Topical
  - Vitamin D3 derivatives,
  - Tazarotene
  - Lactic acid and
  - Propylene glycol

HARLEQUIN ICHTHYOSIS

- Autosomal recessive
- Rare
- Age of onset: Birth
- Very thick, yellow–brown plates of scale that tightly encase the neonate: ARMOR like
- severe immobilization and restricts ventilation
- Extreme ectropion,
- Eclabium
- Ear deformities
- Microcephaly



HARLEQUIN ICHTHYOSIS

- Shortly after birth, the hyperkeratotic cast cracks
- Large, yellow, adherent plates
- Broad, deep, intensely red fissures
- Increased transcutaneous loss of water, electrolyte imbalances temperature instability.
- Skin infections or sepsis
- Hands and feet are edematous , covered by a mitten-like casing
- Autoamputation

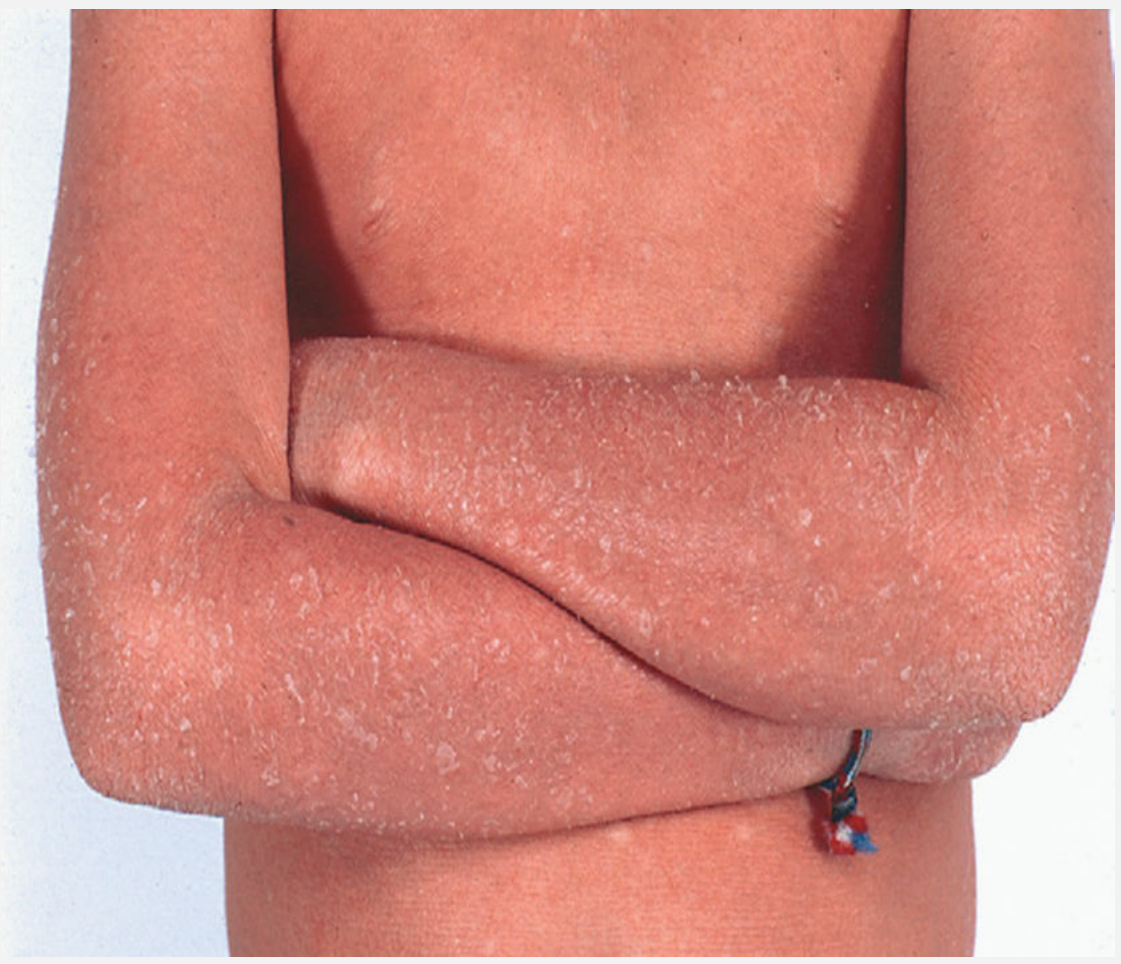
HARLEQUIN ICHTHYOSIS

- Premature delivery
- Neonatal death common : sepsis or respiratory insufficiency
- Survivors develop severe CIE-like phenotype

- Early administration of acitretin (1 mg/kg/day)

CONGENITAL ICHTHYOSIFORM ERYTHRODERMA (CIE)

- Autosomal recessive
- Age of onset : Birth
- Frequently collodion membrane at birth;
- Generalized fine, white scale with a “powdery” consistency
- erythroderma
- palms and soles - diffuse, fissuring keratoderma
- Asso. Features
  - Heat intolerance;
  - variable scarring
    - alopecia,
    - Ectropion
- Treatment
  - increased intake of fluids, calories, iron and protein



Congenital ichthyosiform erythroderma.
Intense redness and fine, flaky, white scale on the trunk and arms.
Close-up of fine white coarser yellowish scale in a background of prominent erythema.

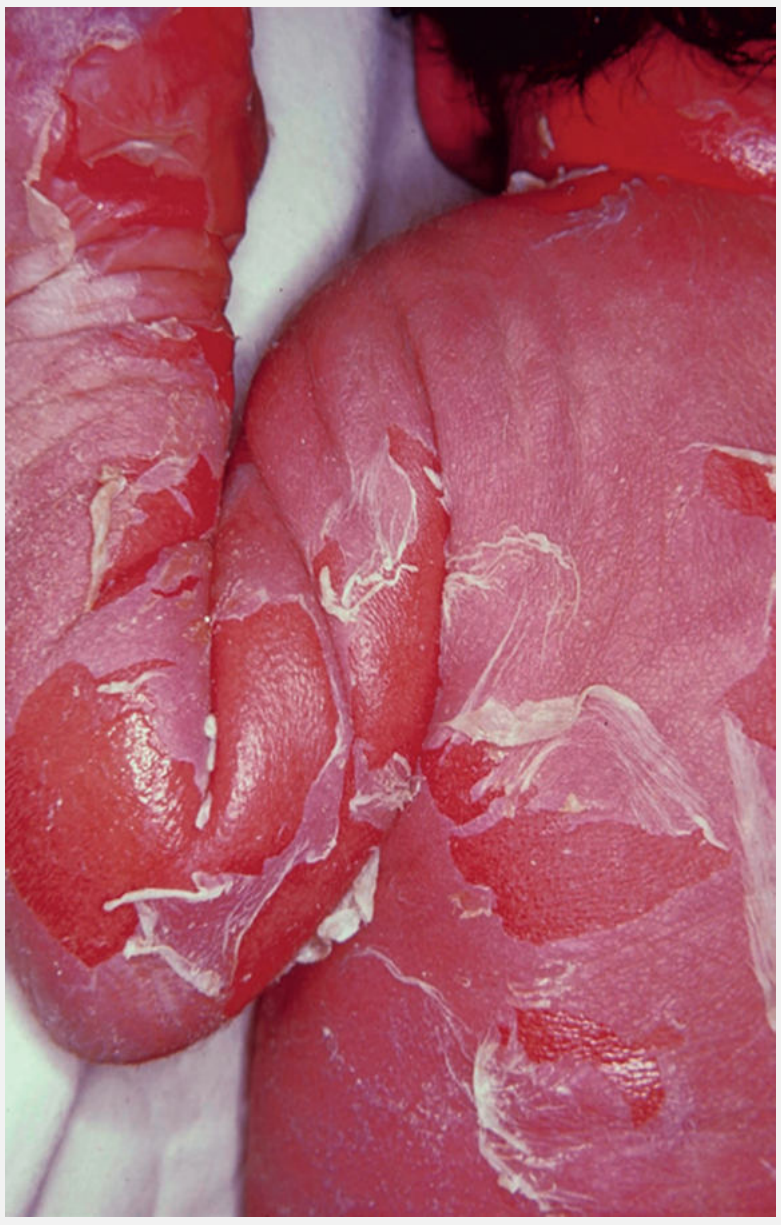
BULLOUS CONGENITAL ICHTHYOSIFORM ERYTHRODERMA

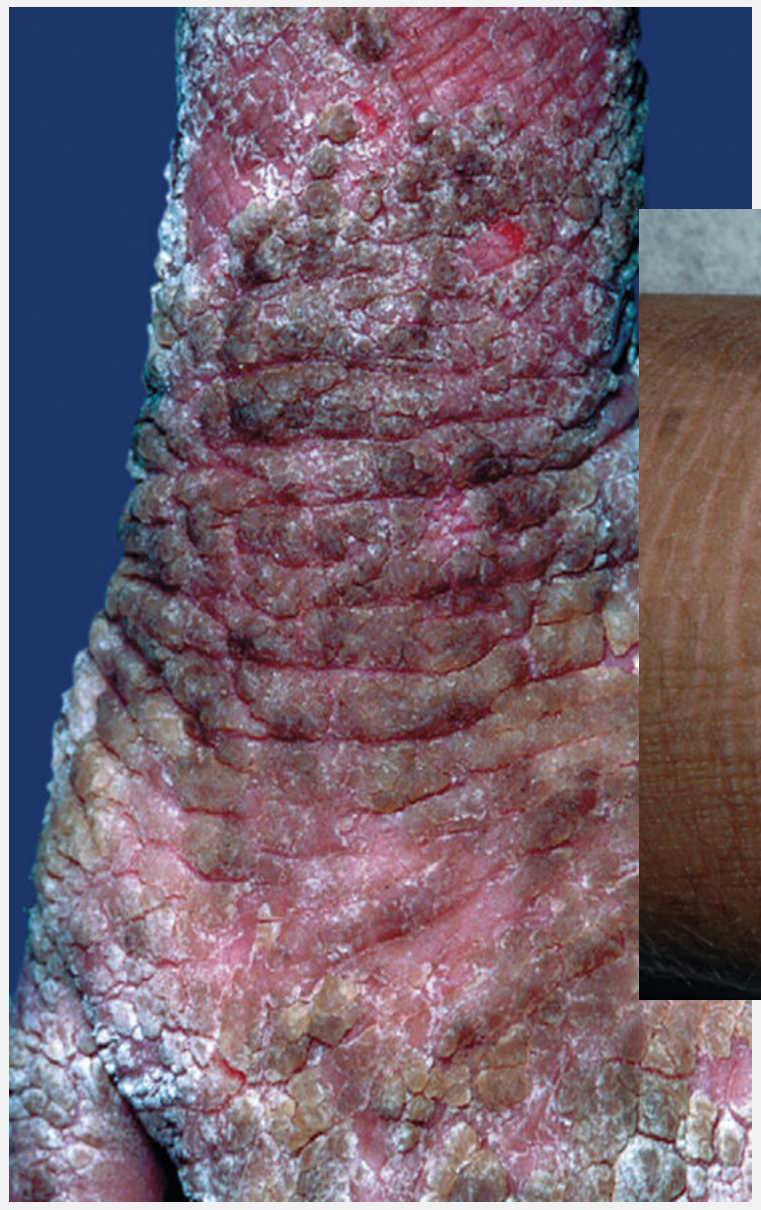
- Autosomal dominant
- Heterozygous mutations in the genes encoding keratin 1 (KRT1) and keratin 10 (KRT10)
- These keratins expressed in spinous and granular layers
- Age of onset : birth
- Erythroderma, peeling, erosions and widespread denuded areas
- Skin fragility, blistering and erythema decrease over time
- Hyperkeratosis develops later during infancy and prevails
- Sepsis, fluid and electrolyte imbalance in the neonatal period : life threatening

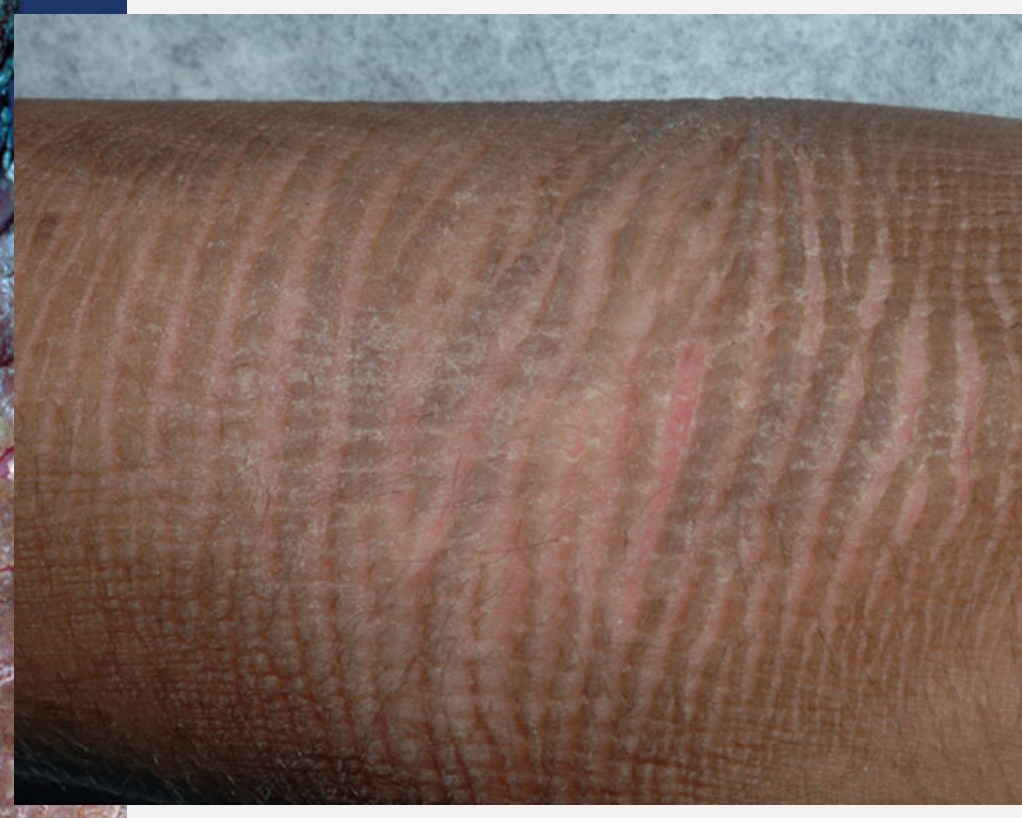
BULLOUS CONGENITAL ICHTHYOSIFORM ERYTHRODERMA

- Episodes of blistering and secondary skin infections problematic later in life
- Accompanied by a pungent body odor,
- Associated posture and gait abnormalities.
- Tremendous impact on patients' quality of life

- Management in NICU as colloidion baby
- Later – hyperkeratosis
  - Keratolytic creams
  - emollients and humectants
- antibacterial soaps, chlorhexidine or dilute sodium hypochlorite baths to control bacterial colonization
- Low doses systemic Retinoids

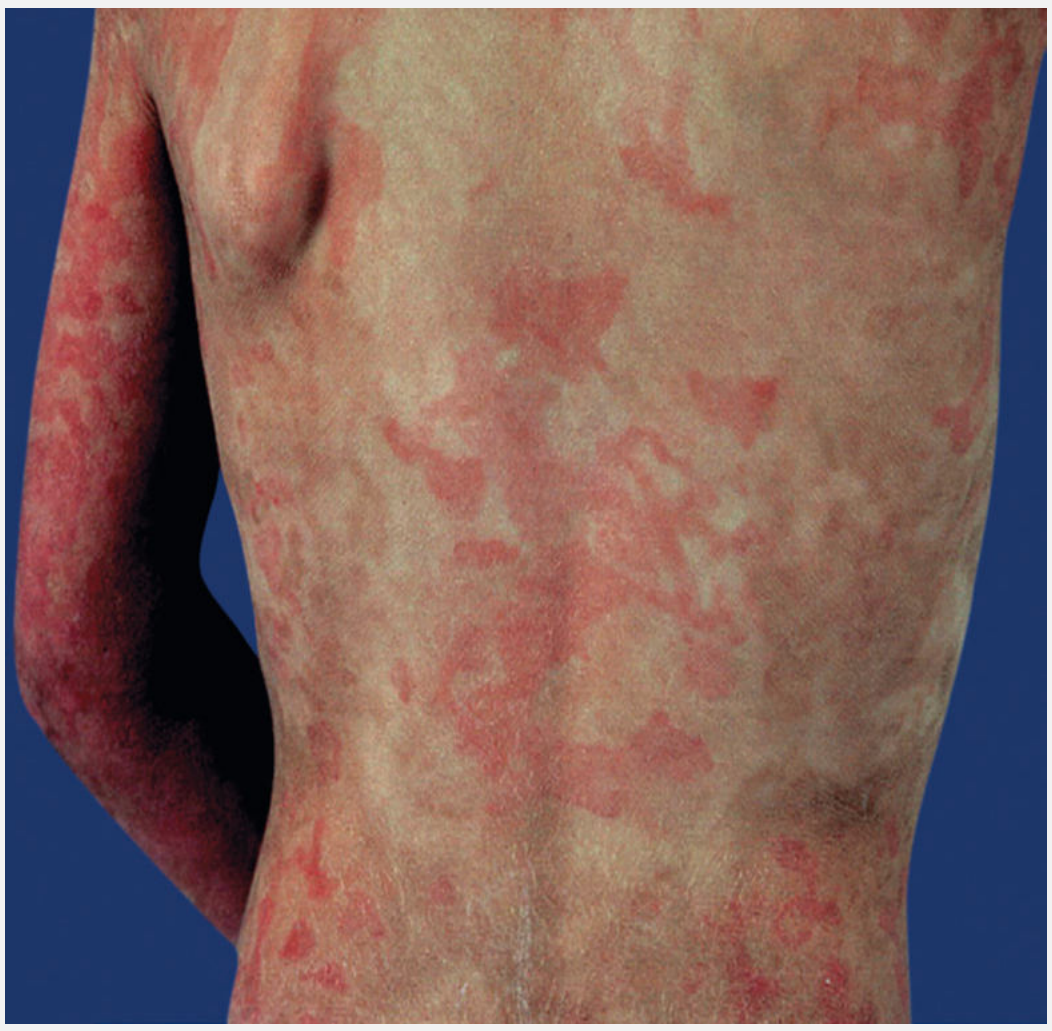


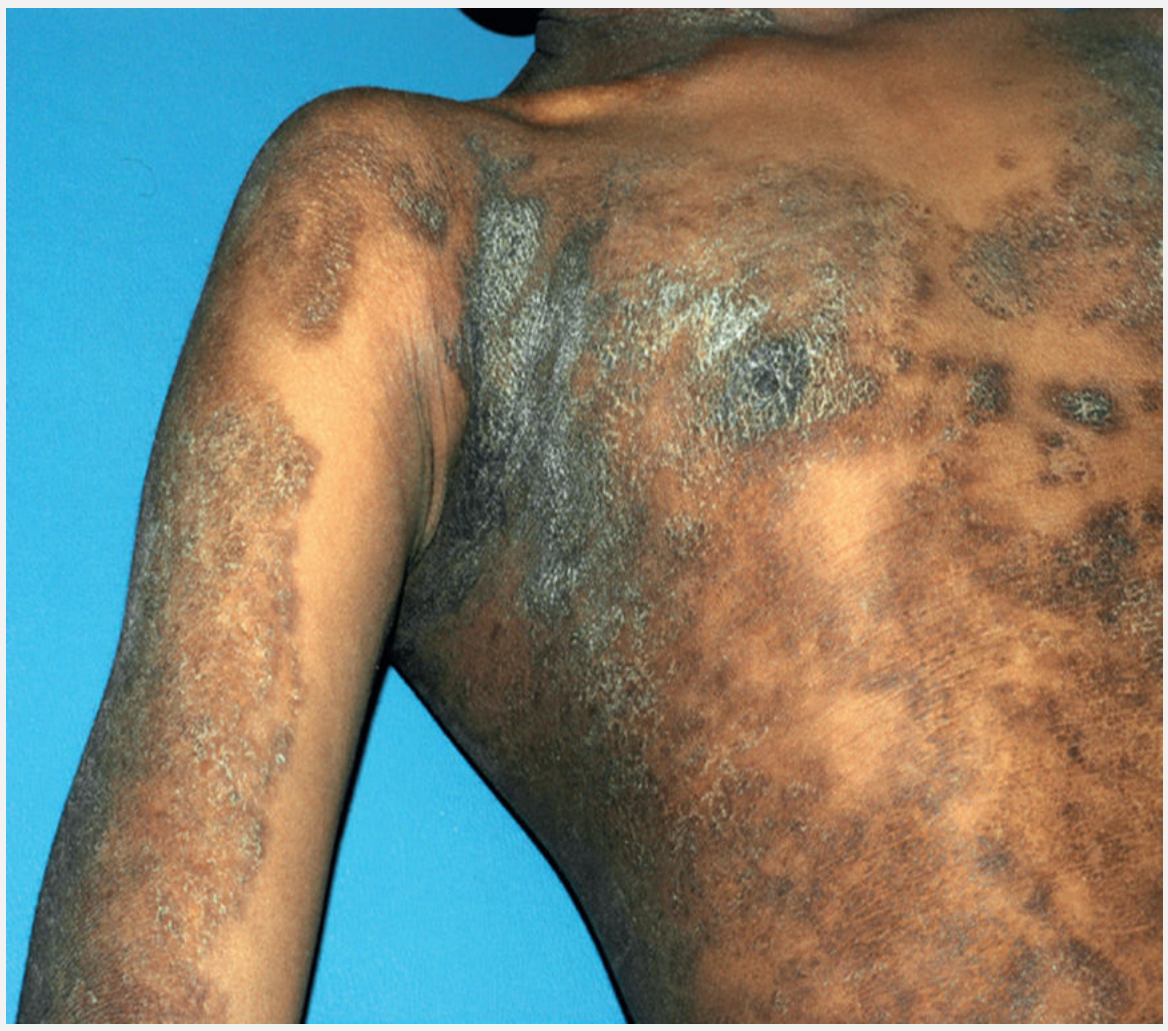




ERYTHROKERATODERMA VARIABILIS

- Autosomal dominant
- Coexistence of transient erythematous patches and stable hyperkeratotic plaques
- One of these features frequently predominates
- Age of onset : at birth or within first year
- Erythema - variable intensity, persist for min to hours
- Sharply demarcated, yellow– brown thickened hyperkeratotic plaques with geographic borders on limbs, buttocks, trunk
- Symmetric distribution
- Topicals for mild diseases
  - Keratolytics (e.G. Lactic acid, urea, α-hydroxy acids)
  - Retinoids (e.G. Tretinoin, tazarotene)
- Systemic retinoids – acitretin, isotretinoin for more extensive disease





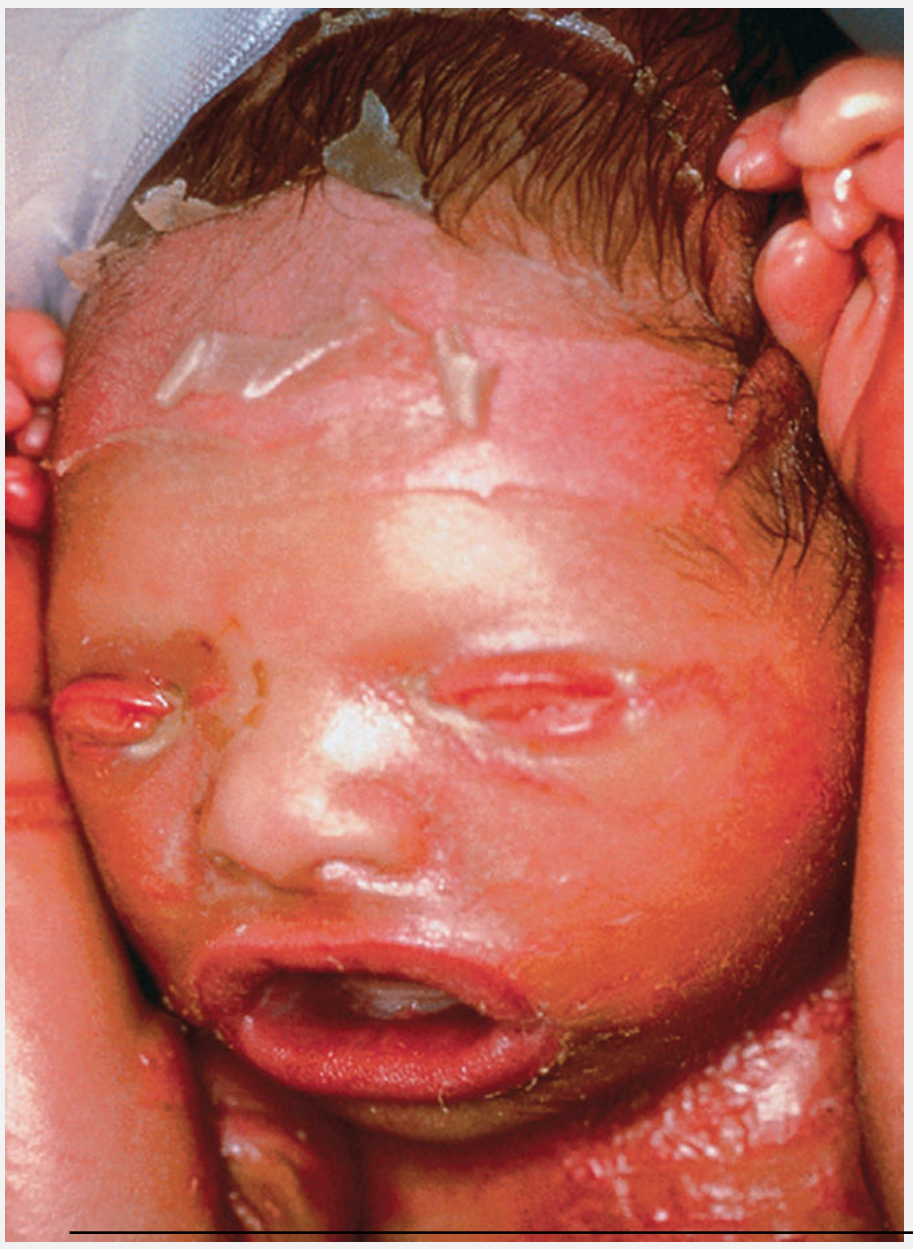
COLLODION BABY

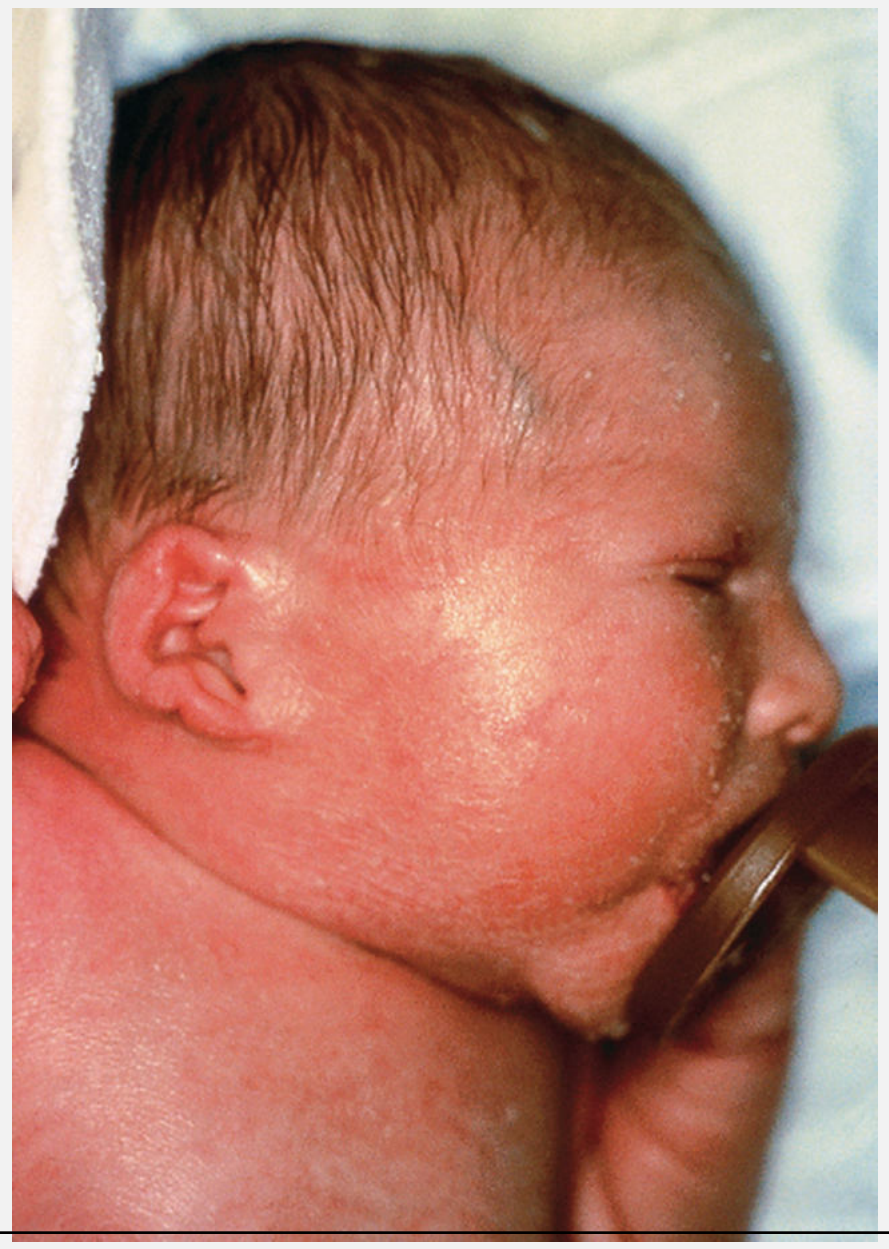
- At birth, neonate is covered with a taut, shiny, transparent membrane, resembling a plastic wrap
- Membrane formed by thickened stratum
- Tautness of the membrane leads to
  - ectropion,
  - eclabium
  - hypoplasia of nasal,auricular cartilage.
- At risk for
  - thermoinstability,
  - hypernatremic dehydration,
  - skin infections and sepsis.
  - Pneumonia due to restricted ventilation and aspiration of amniotic fluid containing scales.
  - Sucking impaired- malnutrition

COLLODION BABY

- After birth, membrane dries, cracks and breaks
- Fissures –
  - impair epidermal barrier function,
  - percutaneous loss of water - fluid and electrolyte imbalances,
  - entry of microorganisms; skin infections, sepsis
- Circular bands of hardened skin - vascular constriction, distal edema.

- In two weeks, membrane peels off in sheets
- Transition to the underlying disease phenotype takes place





Collodion baby.

Day 1: eclabium.

Day 8 : erythema, diffuse mild scaling, misshapen ears.

COLLODION BABY

Colloidon baby seen in

- Lamellar ichthyosis
- Congenital ichthyosiform erythroderma
- Self-healing/self-improving collodion baby

Treatment – NICU- humidified incubator

- protective isolation to prevent infection
- prevent or treat dehydration, electrolyte imbalance
- protective padding and lubricants
- wet compresses,light emollients on skin
- Prevent dehydration , dyselectrolytemia, hypothermia, infection

A NEONATE/YOUNG INFANT WITH A COLLODION MEMBRANE/ICHTHYOSIS

- Complete blood count, electrolytes, hepatic panel, immunoglobulin levels (including IgE)
- Peripheral blood smear
- Light microscopic examination of clipped hairs (including eyebrow hairs)
- Hearing screen
- Ophthalmologic examination
- Consider skin biopsy,
- Consider X-rays to evaluate epiphyses, especially if ichthyosiform lesions are in a mosaic distribution pattern