DISEASES OF SKIN

DEPARTMENT OF ORAL AND MAXILLOFACIAL
PATHOLOGY & ORAL MICROBIOLOGY

CONTENTS

ECTODERMAL DYSPLASIA

- Represents a group of inherited diseases in which two or more ectodermally derived structure does not develop
- it can be autosomal dominant, autosomal recessive and X-linked
- it shows X-linked inheritance

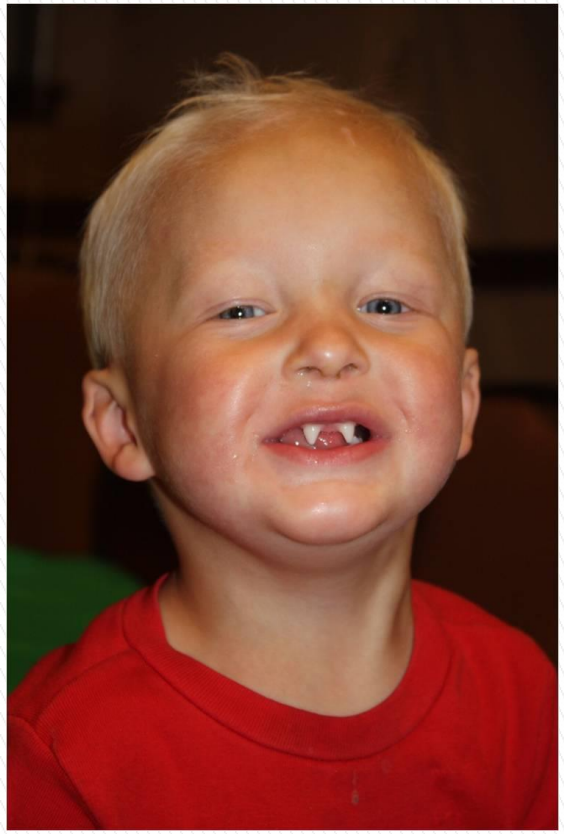
Clinical features

• It is characterised by hypotrichosis, hypohydrosis and anhidrosis with saddle nose appearance .

• the hair and scalp and eyebrows tend to be fine, scanty and blond

• supraorbital and frontal bosses are pronounced .

• there is heat intolerance. Such patients cannot perspire and they usually suffer from hyperpyrexia and inability to endure warm temperature.



Radiological features

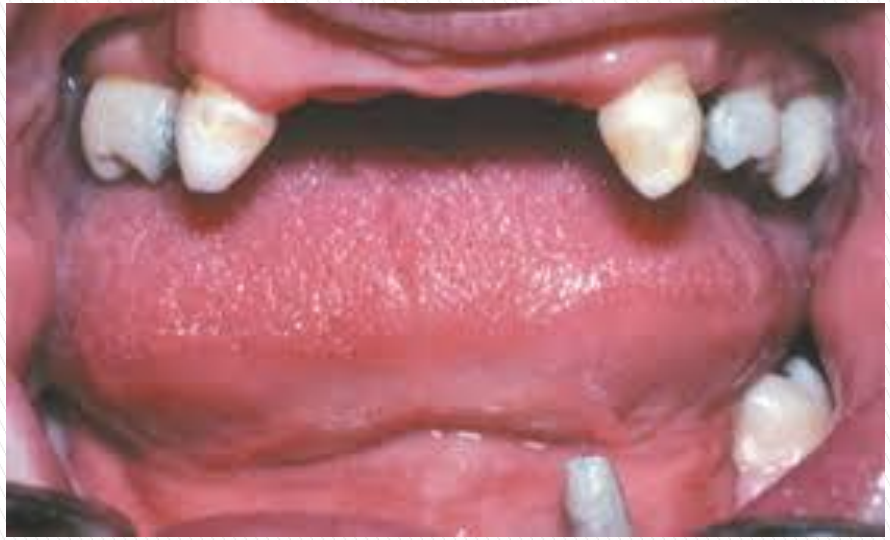
- Since the alveolar process does not develop in the absence of teeth, they appear as thin and less vertical dimension
- Radiograph will show absence of teeth

Oral manifestations

• Oligodontia or anodontia

• The palatal arch is frequently high and the cleft palate may be present

• Midfacial hypoplasia – there is protuberant lip of the patient



Diagnosis

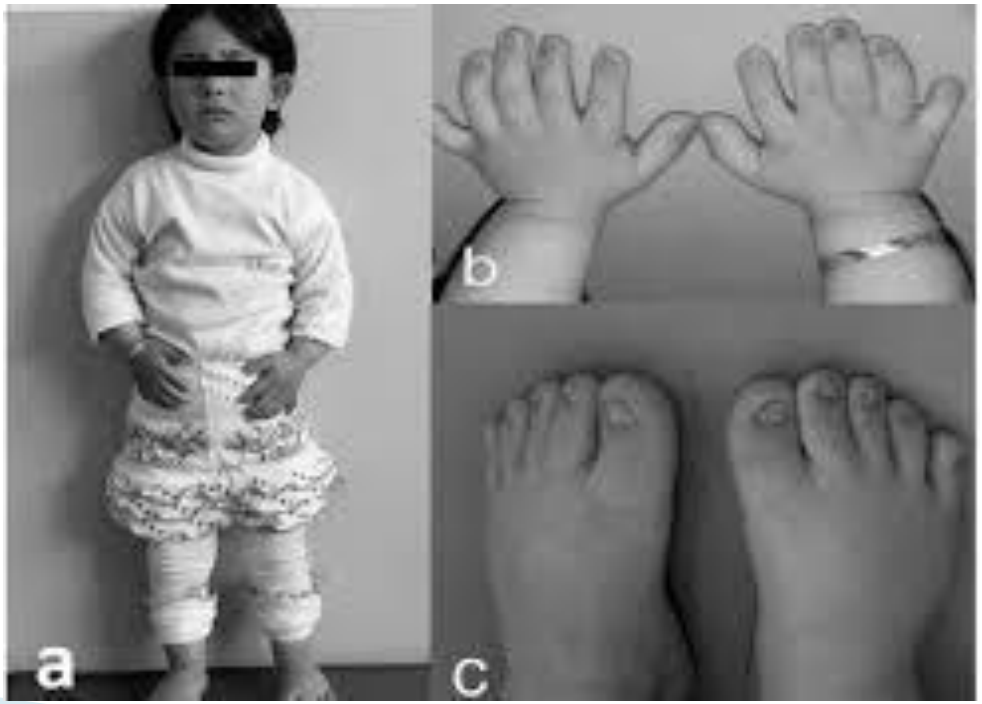
- Absence of teeth with dry skin is valuable aid in the diagnosis of ectodermal dysplasia
- Reduction in the height of alveolar bone

Management

- Partial and complete denture should be constructed for both functional and cosmetic purpose
- Endogenous dental implant

CHONDROECTODERMAL DYSPLASIA

- It is also called as Ellis van Creveld disease
- It is congenital and the patient present evidence of chondrodysplasia, ectodermal dysplasia, polydactylism and congenital morbus Cordis



Clinical features

• Congenital heart defects may include hypoplasia of aorta, atrial and ventricular septal defects and a single atrium

• there is absence of nails and the hair tend to be fine and sparse

• there may be deformity of the knees that frequently progresses and causes a significant malalignment

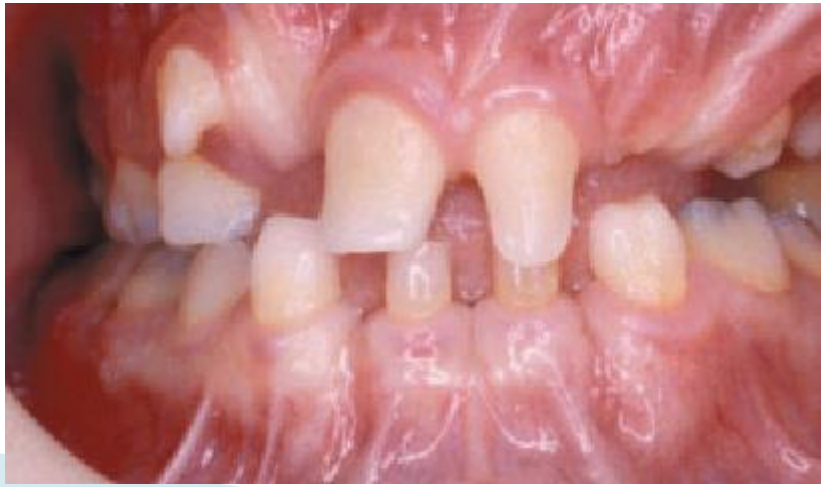
• there is bone dysplasia

Oral manifestations

• Teeth are deficient in number and those which develop are small, rudimentary, conical, spaced and irregular in position. The teeth are affected, with eruption occurring at birth or shortly thereafter

• The deciduous molars present a crenate occlusal surface. Eruption is delayed

• the lip deformity often referred to as partial harelip results from an abnormally short upper lip, which may also be sunken secondary to hyperplasia of maxilla



Radiographic features

- As mentioned previously, there are shorter distal and middle phalangeal, in relation to the proximal phalangeal segments
- deciduous molars show typical shape

Management

- Management of harelip is done by surgical method for cosmetic reason

ORAL LICHEN PLANUS

- Common chronic immunological mucocutaneous disorder that varies in appearance from keratotic to erythematous and ulcerative.

- Wilson 1896

• Lichen planus is a relatively common disorder of the startified squamous epithelia.

-Duske and Frick 1982

EPIDEMIOLOGY

- Different prevelance figures for OLP have been reported.
- • They vary from 0.6% to 2.2%.
- • In Indians it was approx 1.5% (3.7% in tobacco users and 0.3% in non users of tobacco)
- • Females are affected more compared to males.

• Mean age at time of diagnosis is approx 55 years.

ETIOPATHOGENESIS

- The specific etiology is unknown.
- It is generally considered to be an immunologically mediated process, which could be as a result of interaction/interplay between multiple factors.
- There is also no definitive immunogenetic basis yet established for LP and familial cases are rare.
- Believed to be related to psychological stress, which affects severity

Pathologically, there is a local cell-mediated immunological response characterized by a dense T lymphocyte inflammatory cell infiltrate in the upper lamina propria causing cell death (apoptosis) in the basal epithelium.

- This is probably caused by the production of cytokines such as
  - tumor-necrosis factor alpha (TNF∝)
  - interferon gamma (IFN-γ)
  - Interleukin -1

CLINICAL FEATURES

OLP may be associated with pain or discomfort, which interferes with function and with quality of life.

- Symptoms vary from mucosal sensitivity to continuous debilitating pain
- The buccal mucosa is the most commonly affected site.
- Other sites include the tongue and the gingivae
- OLP lesions usually persist for many years with periods of exacerbation and quiescence.

- Exacerbation of OLP has been linked to periods of psychological stress and anxiety.
- • Periods of exacerbation characterized increased erythema or ulceration increased pain and sensitivity.
- • Quiescence period is associated with decrease of erythema or ulceration decreased pain and sensitivity.
- • Quiescent OLP typically as faint white striations, papules or plaques which patient may not be aware of

DERMATOLOGICAL MANIFESTATIONS

- Purple, pruritic and polygonal papules.
- May be discreet or gradually coalesce into plaques each covered by fine glistering scale.
- Bilaterally symmetrical.
- Increase in size if subjected to any irritation.
- Usually self limiting unlike the oral lesions lasting only one year or less. Initially red > purple or violet hue > a dirty brownish color
- Periods of regression and recurrence
- “Koebner’s phenomenon”- skin lesions extend along the areas of injury or irritation
- Most often on wrist, forearms, knees, thighs and trunk
- Face remains uninvolved

CLASSIFICATION

ANDREASEN classified oral lichen planus into 6 types

I. Reticular

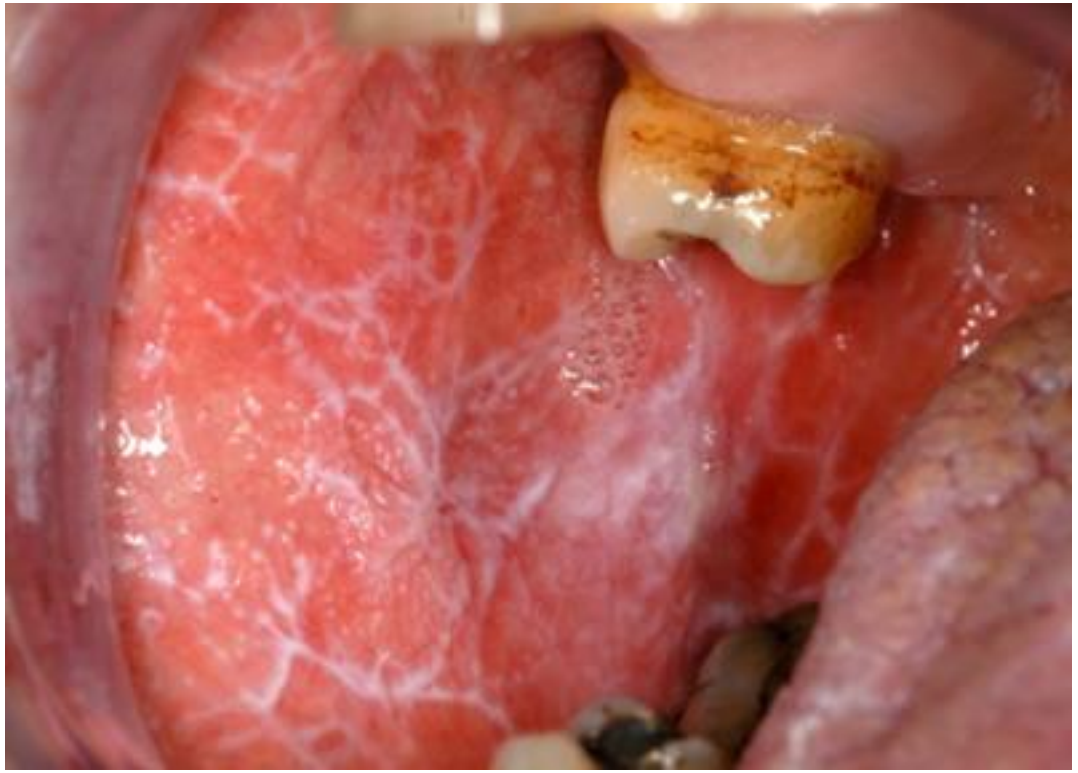
II. Papular

III. Plaque

IV. Atrophic

V. Erosive

VI. Bullous



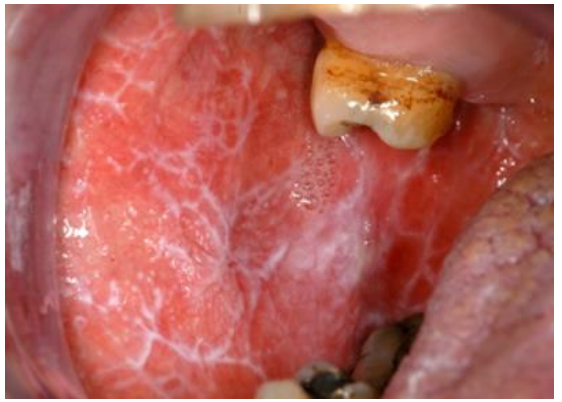
RETICULAR TYPE

- Most common and most readily recognized form.
- Characterized by numerous interlacing white keratotic lines or striae – called Wickham's striae that produce an annular or lacy pattern.
- The buccal mucosa is the site most commonly involved- bilaterally.
- This form generally presents with minimal clinical symptoms and is often an incidental discovery.
- It is the baseline presentation found in almost all OLP patients.
- They may also be seen on the lateral

border of tongue and less often on the gingiva and the lips.

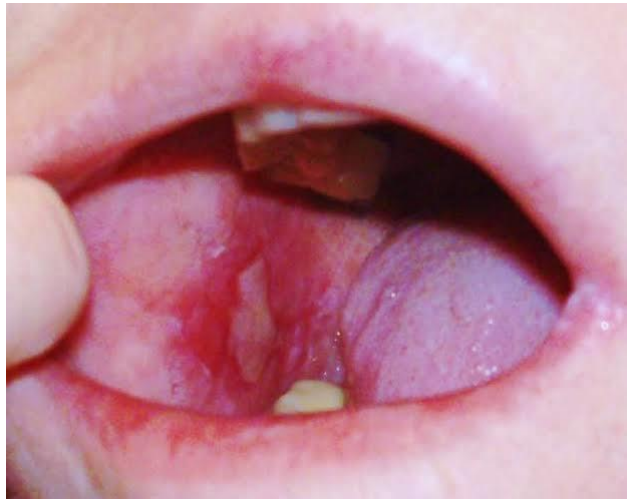
- Reticular lichen planus is likely to resolve in

41 % of cases



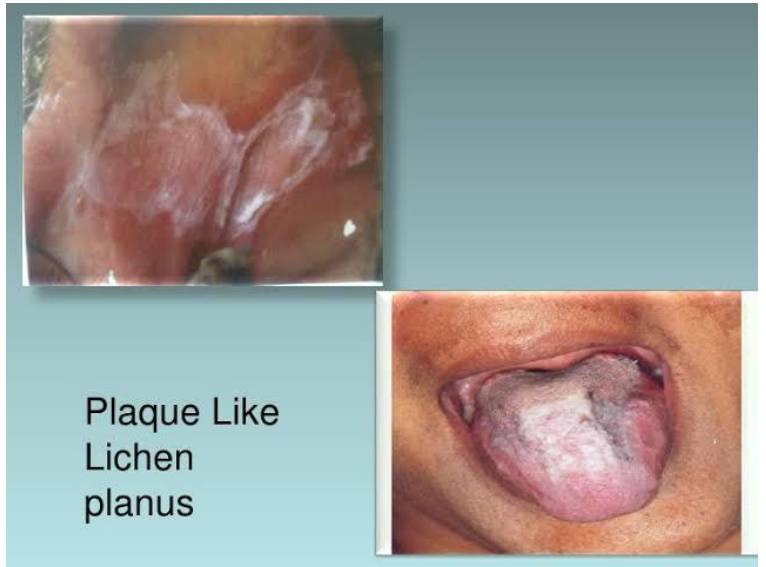
EROSIVE TYPE

- The 2nd most common type after the reticular type.
- The lesion consist of mixture of erythematous and ulcerated areas surrounded by radiating keratotic striae.
- It has a similar appearance to candidiasis and pemphigus.
- Lesions tend to migrate and are often multifocal.
- Mostly affect the buccal mucosa and vestibule
- It is usually symptomatic, characterized by: Sore mouth sensitive to heat, cold, spices, and alcohol Pain and bleeding on touch
- Commonly on buccal mucosa and vestibule
- More dysplasia and malignant transformation



PLAQUE TYPE

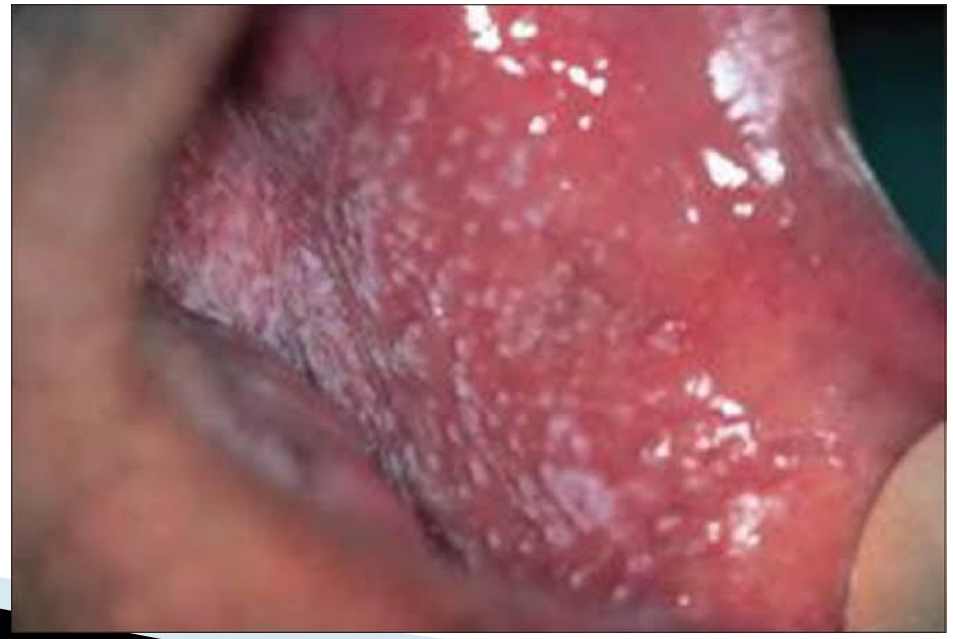
- Tends to resemble leukoplakia clinically but has a multifocal distribution.
- The plaques generally range from slightly elevated to smooth and flat.
- The primary sites for this variant are the dorsum of the tongue buccal mucosa.
- Resolves in only 7% of cases.
- This form is significantly more common among tobacco smokers.



PAPULAR TYPE

The papular type of OLP is usually present in the initial phase of the disease

- This form presents as small white pinpoint papules about 0.5 mm in site.
- It is rarely seen and being small, it is possible to overlook them during a routine oral examination.

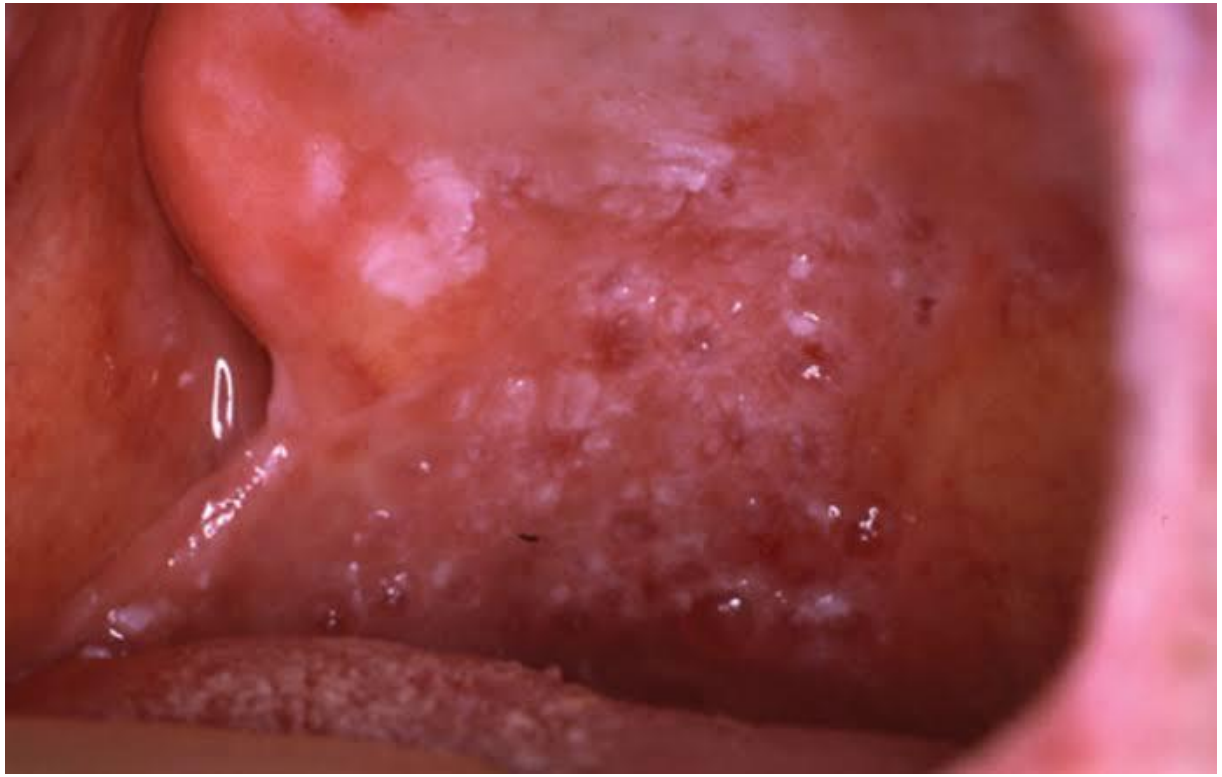


BULLOUS TYPE

Appear as small bullae or vesicles that tend to rupture easily.

- The bullae or vesicles range from a few millimeters to several centimeters in diameter which when ruptured leave an ulcerated, painful surface.
- This form is rarer than the other forms of oral lichen planus.
- Usually present in combination with reticular or erosive pattern.
- The bullous form is commonly seen on the buccal mucosa, particularly in the postero-inferior areas adjacent to the second or third molar teeth.
- The next most common site is the lateral margin of the tongue.
- The lesions are rarely seen on the gingiva or inner aspect of the lips

Bullous type



ATROPHIC TYPE

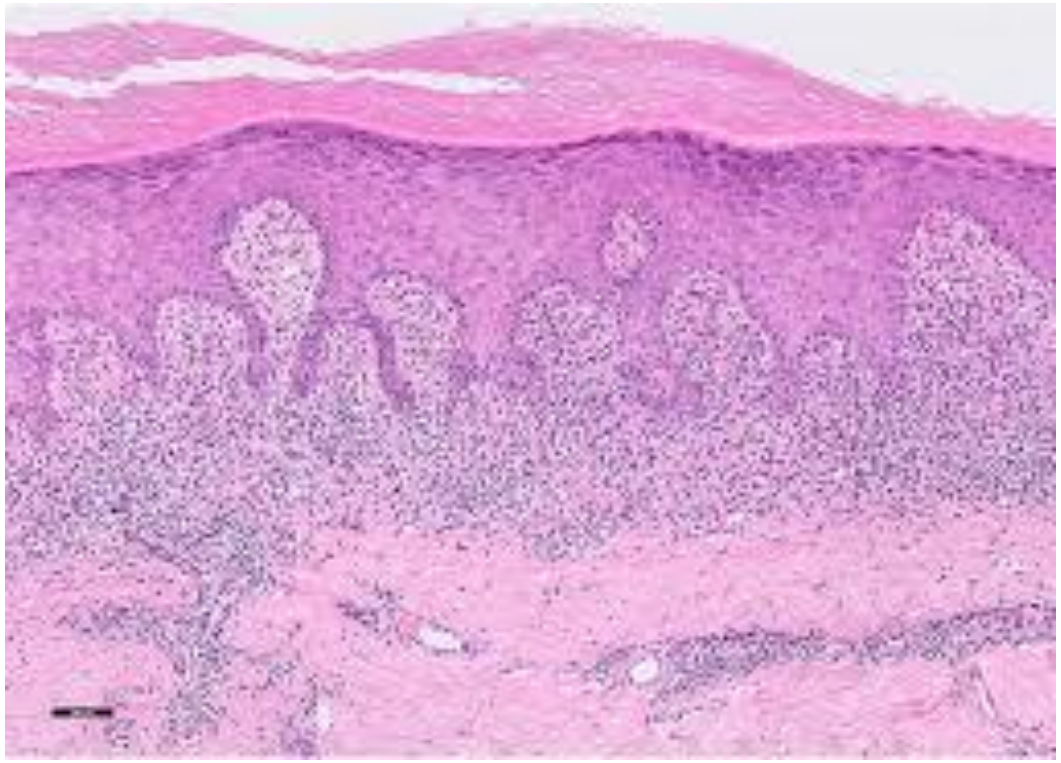
- The atrophic type is diffuse, red and there are usually white striae within the lesion.
- Striae that radiate peripherally are usually evident at the margins of the atrophic zones of the lesion.
- The attached gingiva is often involved and the condition is commonly referred to as `chronic desquamative gingivitis'.
- The lingual gingiva is usually less severely involved.
- This condition can cause a burning sensation particularly when in contact with certain foods.
- Patients may complain of burning, sensitivity, and generalized discomfort
- About 12% of the atrophic lesions will resolve spontaneously.


Reticular lichen planus
Erosive lichen planus
Lichen planus plaque

- Diagnosis is based on:
- Clinical Presentation. E.g. Reticular lichen planus with characteristic appearance of Wickham’s striae.
- Histological Examination requires biopsy.
- Direct Immunofluorescent Examinations requires biopsy.

HISTOLOGICAL FEATURES

- The following histologic features are essential for the diagnosis of lichen planus :
- Areas of hyperparakeratosis or hyperorthokeratosis.
- The spinous cell layer may be thickened (acanthosis) with shortened and pointed rete pegs.
- The thickened areas are seen clinically as Wickham's striae.
- Liquefaction degeneration or necrosis of the basal cell layer- Max Joseph spaces which is often replaced by an eosinophilic band.
- There is also dense subepithelial band of lymphocytes.
- Isolated epithelial cells, shrunken with eosinophilic cytoplasm and one or more pyknotic nuclear fragments-Civatte bodies.
- Often scattered within the epithelium and superficial lamina propria.
- These represent cells that have undergone apoptosis




Saw tooth appearance of rete ridges
A band like lymphocytic infiltrate
fills the papillary dermis. The
infiltrate press against the
undersurface of the epidermis

MANAGEMENT

- No treatment for OLP is curative
- Goal:

Reduce painful symptoms

Resolution of oral mucosal lesions

Reduce risk of oral squamous cell carcinoma

Improve oral hygiene

- Eliminate exacerbating factors Repair defective restorations or prosthesis Remove offending material causing allergy
- Diet Eliminate smoking and alcohol consumption Eat fresh fruit and vegetables (but avoid tomatoes and nuts)

Reduce Stress

- Topical corticosteroids
- 0.05% clobetasol proprionate gel
- 0.1% or 0.05% betamethasone valerate gel
- 0.05% fluocinonide gel
- 0.05% clobetasol butyrate ointment
- 0.1% triamcinolone acetonide ointment Can be applied directly or mixed with Orabase

- Systemic Steroid Therapy
- Prednisone (for 70kg adult) :10-20mg/day for moderately severe cases -As high as 35 mg/day for severe cases
- Should be taken in the morning to avoid insomnia
- Should be taken with food to avoid peptic ulceration
- Azathioprine – Inhibits synthesis of DNA :1mg/kg/d for 6-8 weeks
- Methylprednisolone : to reduce pain and inflammation

PSORIASIS

- Psoriasis is a chronic non-infectious, inflammatory disease of the skin in which epidermal cells are produced at a rate that is about six to nine times faster than normal.

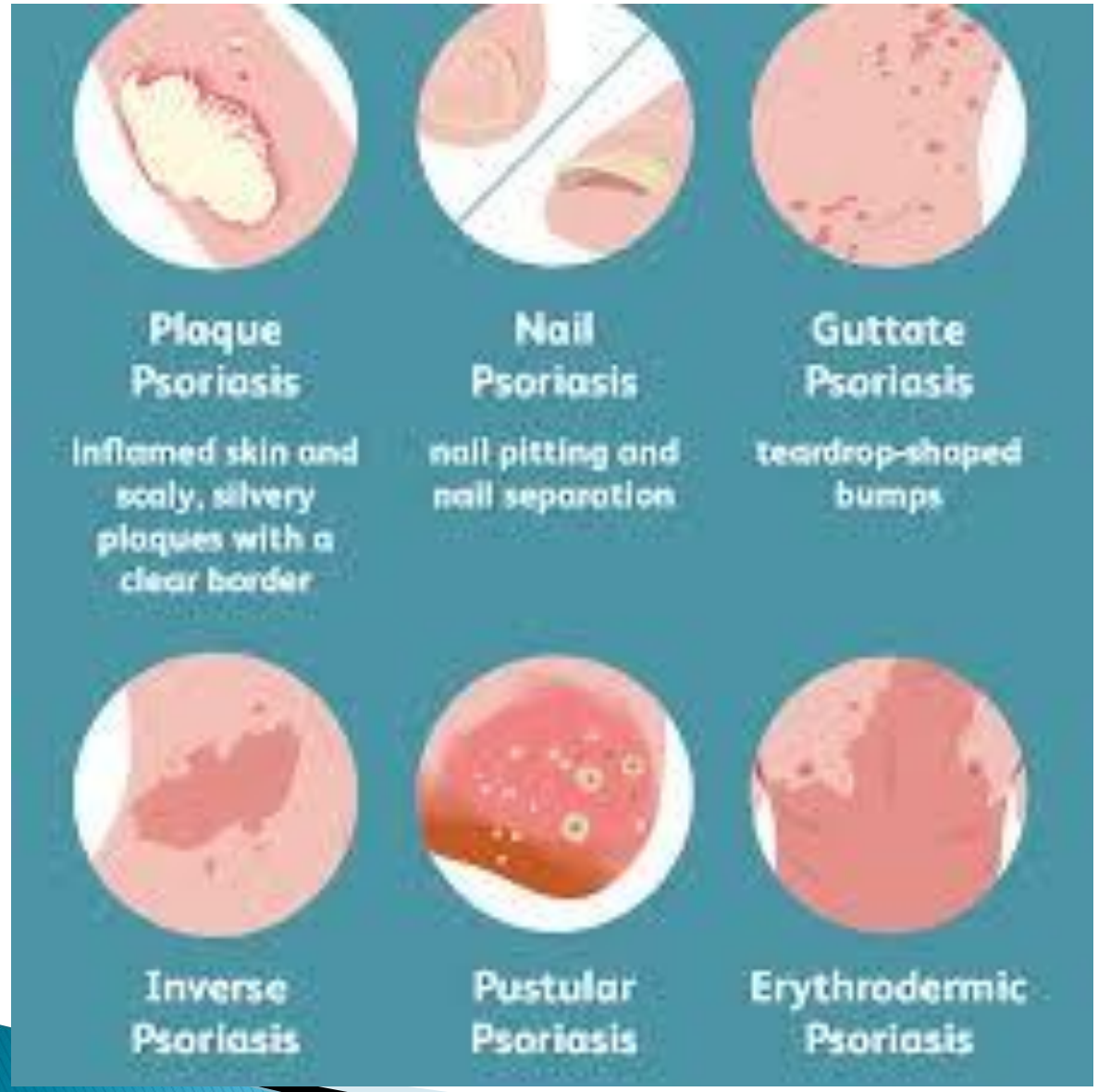
ETIOLOGY

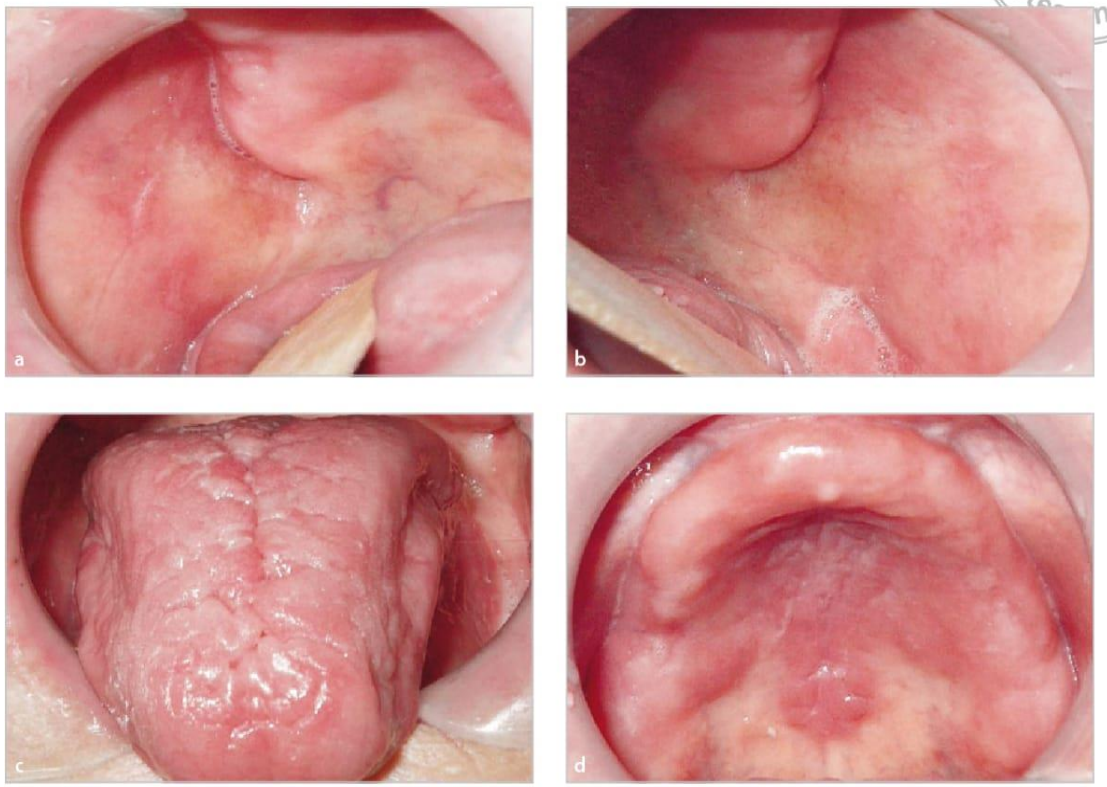
- Idiopathic cause
- Some of the factors that may trigger psoriasis are:
- Genetic
- Autoimmune reaction
- Infection
- Injury to skin
- Changes in climate
- Medications: Lithium, Antimalarial Medications, Propronalol , Indomethacin
- Stress
- Obesity
- Smoking

CLINICAL FEATURES

- Initially the first sign of psoriasis is often red spots on the body.
- Dry, swollen and inflamed patches
- Patches Covered with silver white flakes
- Raised and thick skin
- Other symptoms of psoriasis includes :

Pain, itching and burning sensation Restricted joint motion or pain Cracked and bleeding skin Dandruff on scalp Pus filled blisters Genital lesions in males. Pitting, small depression on the surface of the nail Yellow, discolored nail Arthritis


Plaque Psoriasis
Inflamed skin and scaly, silvery plaques with a clear border
Nail Psoriasis
nail pitting and nail separation
Guttate Psoriasis
teardrop-shaped bumps
Inverse Psoriasis
Pustular Psoriasis
Erythrodermic Psoriasis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- The lesions are most abundant over the scalp, the extensor surface of the elbows and knees, the lower part of the back, and the genitalia.
- Bilateral symmetry is a feature of psoriasis.
- In approximately one fourth to one half of patients, the nails are involved, with pitting, discoloration, crumbling beneath the free edges, and separation of the nail plate.
- When psoriasis occurs on the palms and soles, it can cause pustular lesions called palmar pustular psoriasis.

Histological Features

- Parakeratosis
- Diminished granular layer
- Elongation of rete pegs
- Tortuous papilary dermal vessels
- Neutrophil Excess in epidermis

MANAGEMENT

There is no known cure.

- First, avoid any precipitating or aggravating factors .
- An assessment is made of lifestyle, because psoriasis is significantly affected by stress. The standard treatment modalities includes:
- Topical therapy
- Intralesional therapy
- Systemic therapy
- photochemotherapy

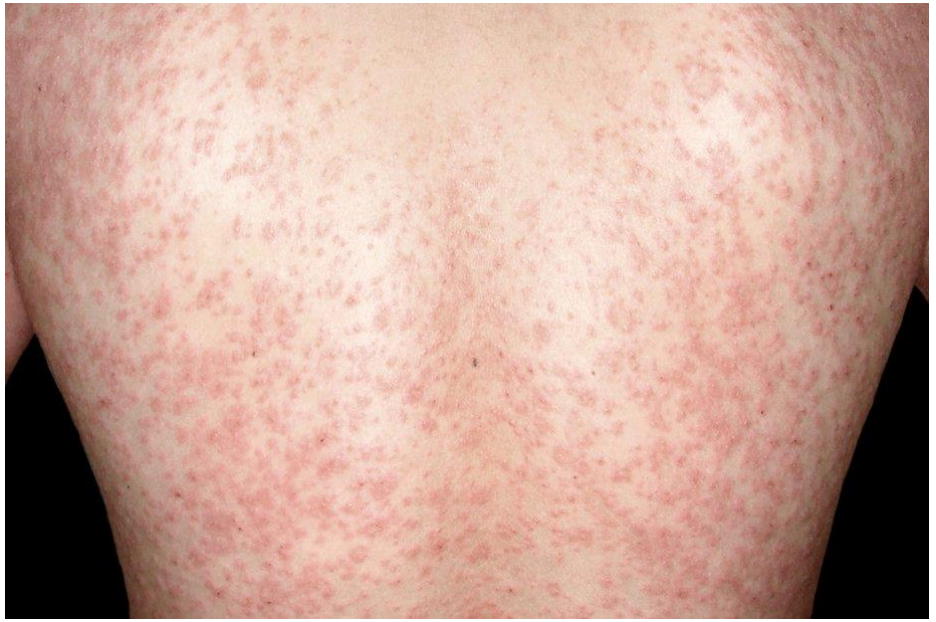
PITYRIASIS ROSEA

• Pityriasis rosea (pit-uh-rahy-uh-sis row-zee-ah) is a common skin disease that causes a rash.

• This rash usually disappears on its own without treatment.

• You can expect to see the rash for about 6 to 8 weeks. Sometimes the rash lasts much longer.

• This rash can be very itchy



Causes

- May be due to a viral infection.
- This skin disease does not seem to be contagious. It does not seem to spread from one person to another.
- People of all ages and skin colors get pityriasis rosea, but this skin disease is more likely to occur:
    - Between 10 and 35 years of age.
    - During pregnancy

Signs and Symptoms

• The symptoms of this condition include:

• An upper respiratory tract infection may precede all other symptoms in as many as 69% of patients.

• A single, 2- to (rarely) 10-cm oval red "herald" patch appears, classically on the abdomen.

• Occasionally, the "herald" patch may occur in a 'hidden' position (in the armpit, for example) and not be noticed immediately.

• The "herald" patch may also appear as a cluster of smaller oval spots, and be mistaken for acne. Rarely, it does not become present at all.

• 7–14 days after the herald patch, many small (5– 10 mm) patches of pink or red, flaky, oval-shaped rash appear on the torso

• In 6% of cases an inverse distribution may occur, with rash mostly on the extremities. The more numerous oval patches generally spread widely across the chest first, following the rib-line in a characteristic "christmas-tree" distribution.

• Small, circular patches may appear on the back and neck several days later. It is unusual for lesions to form on the face, but they may appear on the cheeks or at the hairline.

• About one in four people with PR suffer from mild to severe symptomatic itching. (Moderate itching due to skin over-dryness is much more common, especially if soap is used to cleanse the affected areas.)

Diagnosis

- Discrete circular or oval lesions,
- Scaling on most lesions, and
- Peripheral collarette scaling with central clearance on at least two lesions.
- Truncal and proximal limb distribution, with less than 10% of lesions distal to mid-upper-arm and mid-thigh,
- Orientation of most lesions along skin cleavage lines, and
- A herald patch (not necessarily the largest) appearing at least two days before eruption of other lesions, from history of the patient or from clinical observation.

Epidemiology

• The overall prevalence of PR in the United States has been estimated to be 0.13% in men and 0.14% in women. It most commonly occurs between the ages of 10 and 35.

• It is more common in spring.

• PR is not viewed as contagious though there have been reports of small epidemics in fraternity houses and military bases, schools and gyms.

Management

• No treatment is usually required.

• Oral antihistamines or topical steroids may be used to decrease itching

• Direct sunlight makes the lesions resolve more quickly.

• According to this principle, medical treatment with ultraviolet light has been used to hasten resolution, though studies disagree whether it decreases itching or not. UV therapy is most beneficial in the first week of the eruption.

• Oral erythromycin was effective in treating patients.

• Human Herpes Virus 6 or Human Herpes Virus 7 has been hypothesized to be the cause.

• The antiviral drug Acyclovir can reduce length of duration and severity

ERYTHEMA MULTIFORME

- Stevens-Johnson syndrome,
- erythema multiforme major,
- Erythema multiforme minor,
- herpes-induced EM major,
- herpes-associated erythema multiforme,
- drug-induced Stevens-Johnson syndrome

• An acute self-limiting dermatitis characterized by a distinctive clinical eruption manifested as the iris or target lesion.

• Types of EM:

– EM minor: localized eruption of the skin with mild or no mucosal involvement

– EM major : skin and mucosal erosions of raised atypical target lesions, • usually located on the extremities and/or on the face.

– Stevens-Johnson syndrome (SJS) : skin and mucosal erosions plus widespread distribution of flat atypical targets or purpuric macules, • may be present on the trunk, the face, and on the extremities.

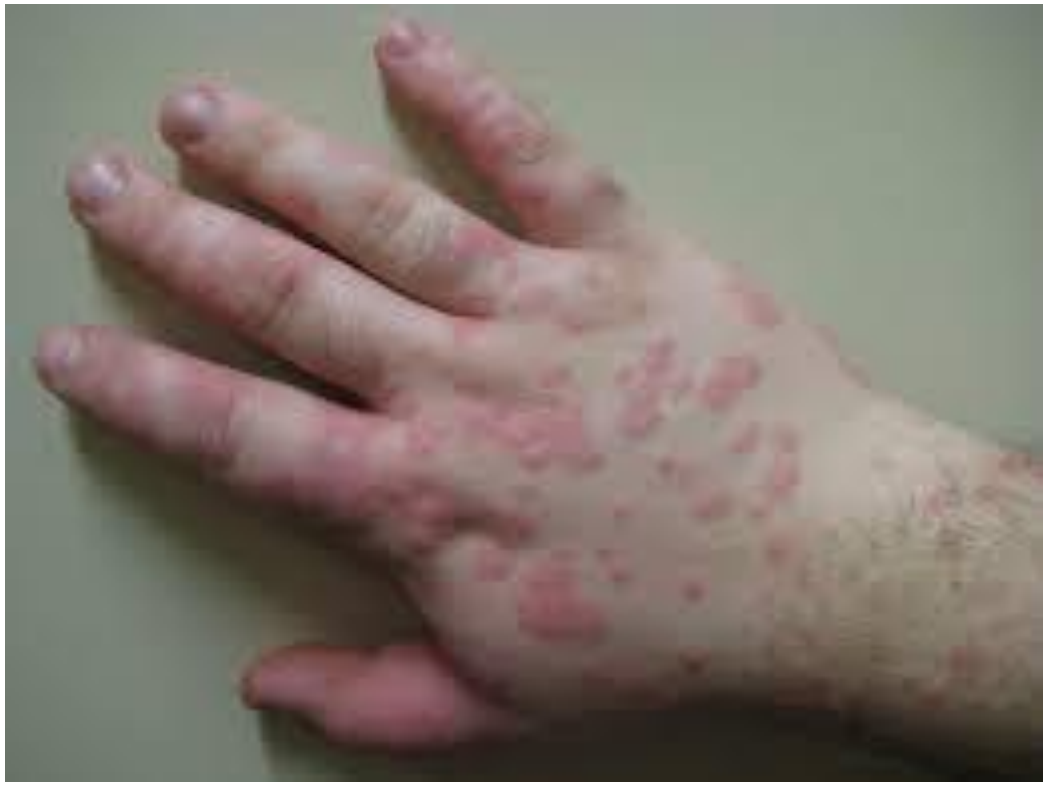
ETIOLOGY

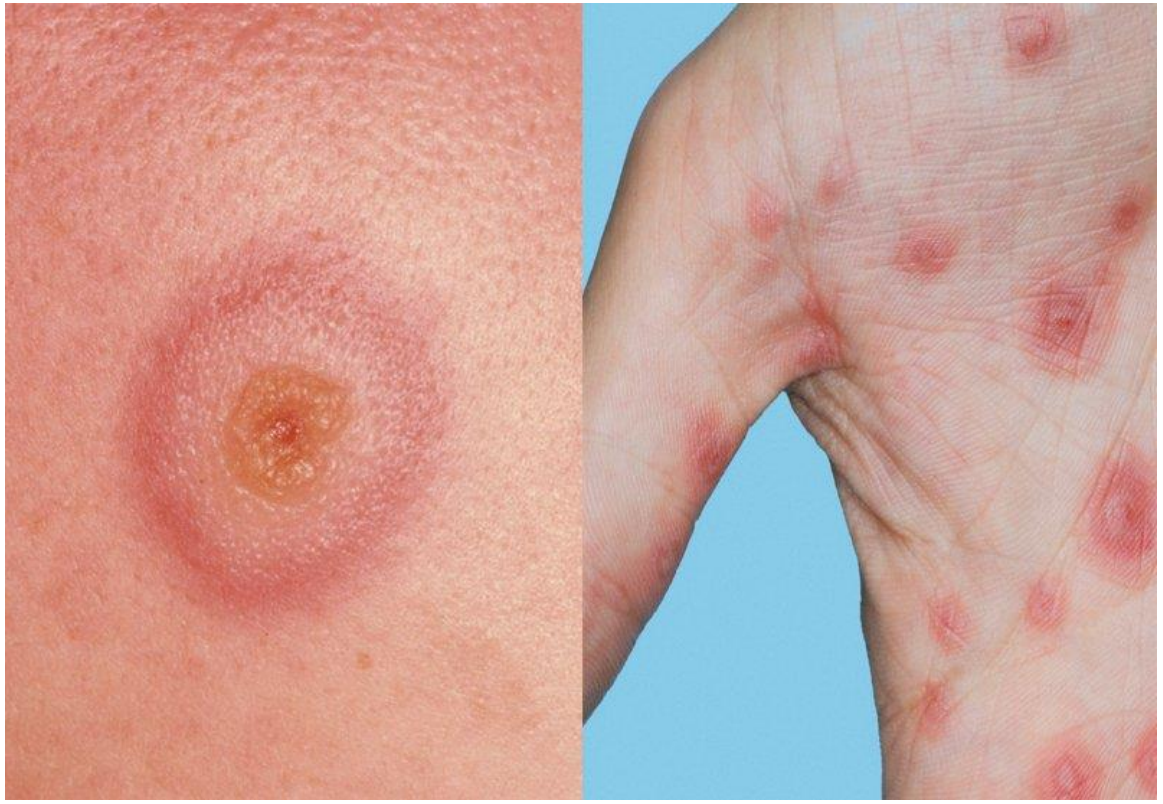
- EM and SJS are both caused by drugs, but infectious agents are considered to be the major cause of EM.
- History of HSV infection 1 to 3 weeks before onset of EM
- EM minor: triggered by HSV in nearly 100% of cases
- EM major: herpetic etiology also accounts for 55% of cases of, other infections, Mycoplasma
- SJS and EM major: Drugs are found to be major cause, antibacterial sulfonamides, anticonvulsants, oxicam, NSAIDs, and allopurinol

CLINICAL FEATURES

- Chiefly in young adults, although it may develop at any age,
- Second to fourth decades of life
- Affects males more frequently than females.
- Characterized by the occurrence of – asymptomatic, – vividly erythematous discrete macules, papules – occasionally vesicles and bullae distributed in a rather symmetrical pattern most commonly over the hands and arms, feet and legs, face and neck.
- Variable size, but are generally only a few centimeters or less in diameter.

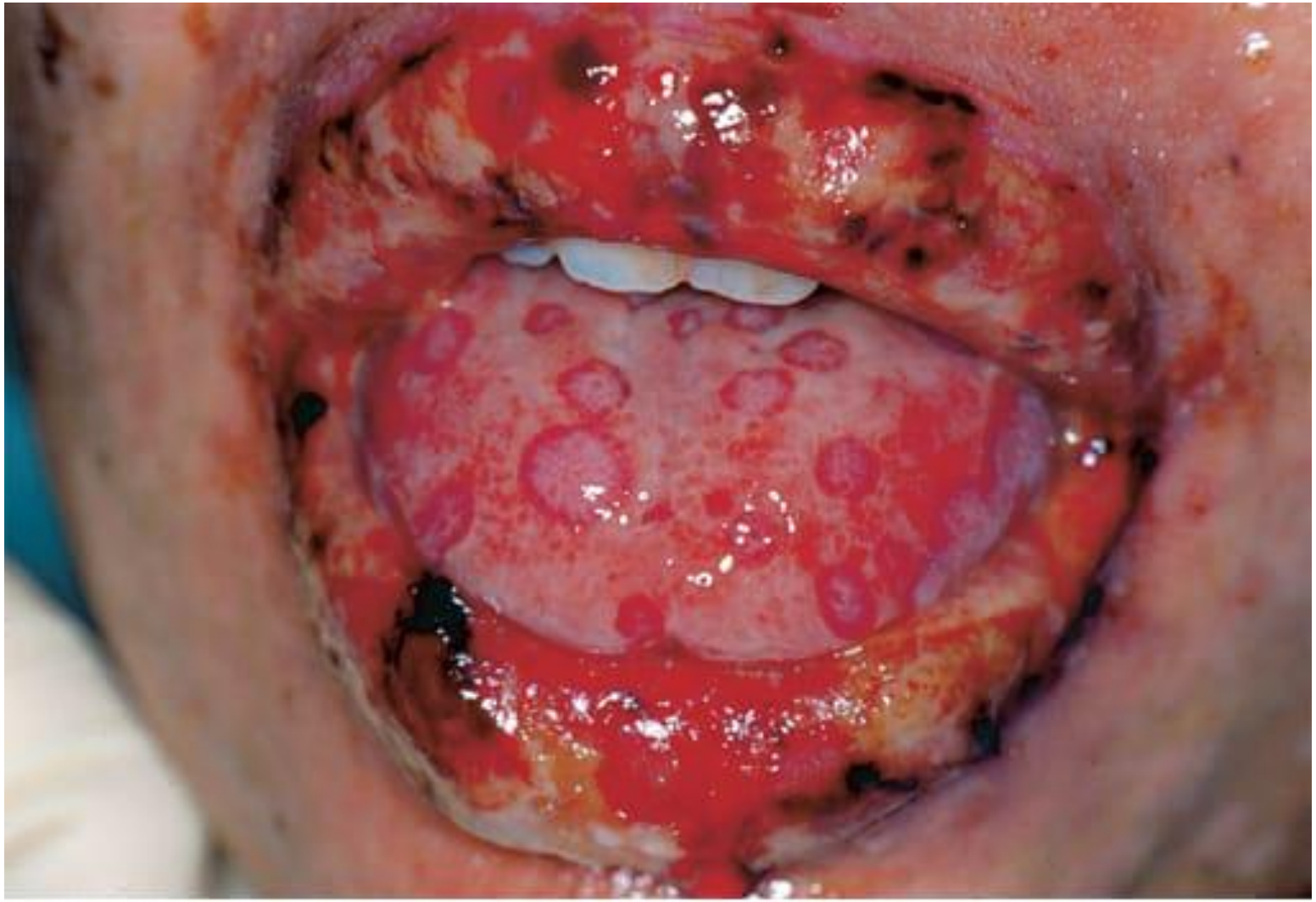
- A concentric ring like appearance of the lesions, resulting from the varying shades of erythema, occurs in some cases and has given rise to the terms ‘target’, ‘iris’, or ‘bull’s eye’
- Most common on the hands, wrists and ankles.
- Mucous membrane involvement, including the oral cavity, is common.
- The lesions make their appearance rapidly,
- usually within a day or two, and persist from several days to a few weeks,
- gradually fading and eventually clearing.
- Recurrence of the disease over a period of years is common





ORAL MANIFESTATIONS

- Usually not a significant feature of the disease
- Except for the pain and discomfort they cause.
- Hyperemic macules, papules or vesicles may become eroded or ulcerated and bleed freely.
- The tongue, palate, buccal mucosa and gingiva are commonly diffusely involved
- Occasionally, mucous membrane lesions occur before the cutaneous manifestations, but oral involvement without dermal lesions has been questioned



Source: J.L. Jameson, A.S. Fauci, D.L. Kasper, S.L. Hauser, D.L. Longo, J. Loscalzo: Harrison's Principles of Internal Medicine, 20th Edition

STEVENS JOHNSON SYNDROME

- At one time considered to be a separate disease
- Mucocutaneous-ocular disease
- Now recognized as simply a very severe bullous form of EM
- With widespread involvement typically including the skin, oral cavity, eyes and genitalia.
- It commences with the abrupt occurrence of fever, malaise, photophobia, and eruptions of the oral mucosa, genitalia and skin.
- The cutaneous lesions in this are similar to those of EM, #
- Although they are commonly hemorrhagic and are often vesicular or bullous

CLINICAL FEATURES

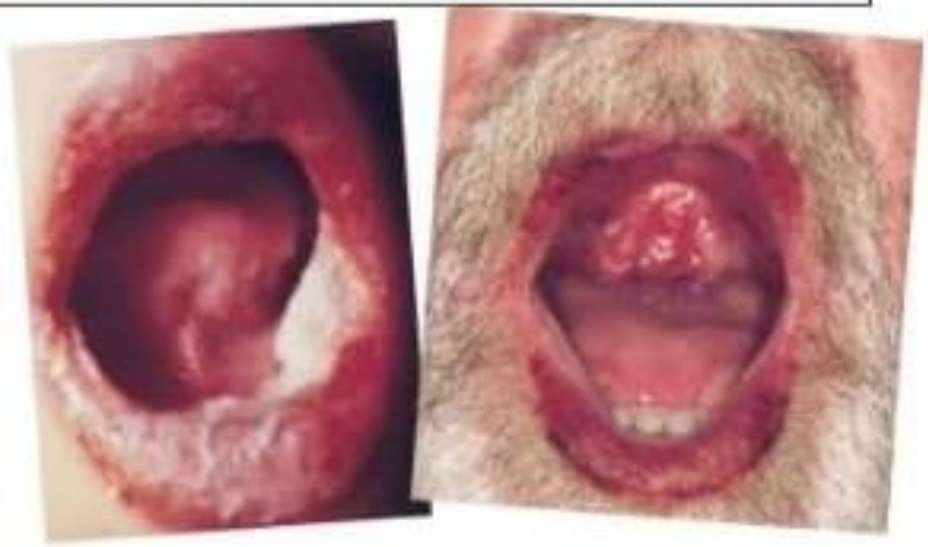
- Eye lesions
  – photophobia, a characteristic of the disease referable to the conjunctivitis,
  – corneal ulceration and panophthalmitis which may occur
  – Keratoconjunctivitis sicca also has been described.
  – Blindness may result chiefly from intercurrent bacterial infection.
- Genital lesions
  – Nonspecific urethritis,
  – balanitis and/or vaginal ulcers
- Other reported complications are related to respiratory tract involvement such as tracheobronchial ulceration and pneumonia.
- The patients usually recover unless they succumb to a secondary infection.

ORAL MANIFESTATIONS OF SJS

- Extremely severe and so painful that mastication is impossible.
- Mucosal vesicles or bullae occur which rupture and leave surfaces covered with a thick white or yellow exudate.
- Erosions of the pharynx are also common
- The lips may exhibit ulceration with bloody crusting and are painful.
- May be the chief complaint of the patient, and understandably, have been mistaken for acute necrotizing ulcerative gingivostomatitis.
- Interestingly, however, it has been reported that the organisms of Vincent's infection are scarce in patients with this disease.
- The mucosal involvement in SJS is more severe and extensive than in EM major

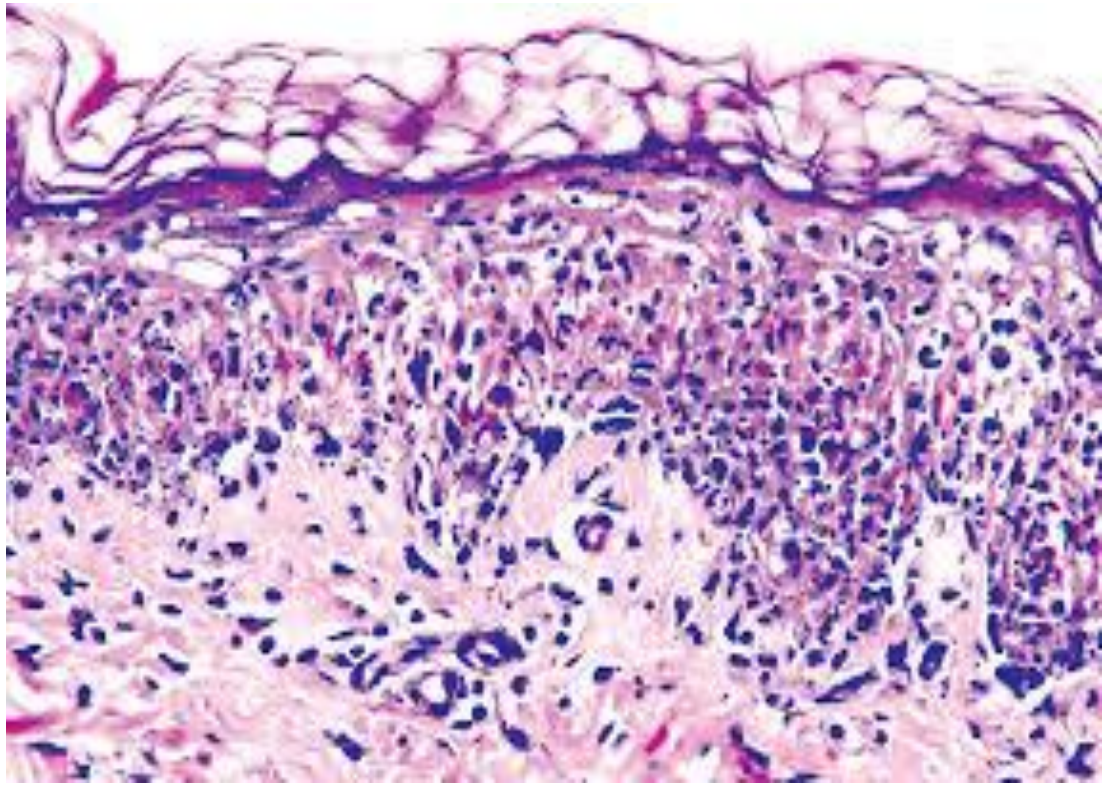
Oral manifestations SJS

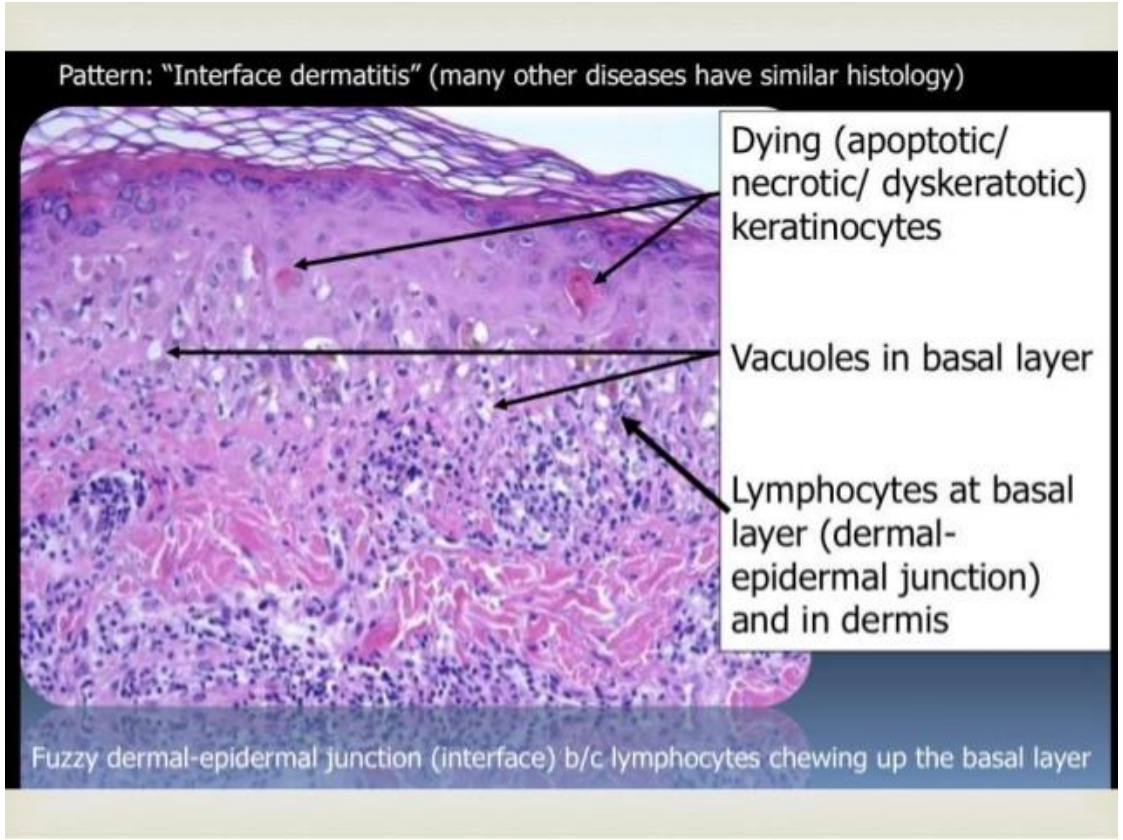
- Severe and painful making mastication impossible
- Mucosal vesicles or bullae occur which ruptures leaving it covered with a ***thick white or yellow exudate***
- ***Lips*** exhibit ulceration with bloody crusting and are painful
- Erosions of ***pharynx*** also common
- Mistaken for *ANUG*



HISTOLOGICAL FEATURES

- Not of diagnostic importance.
- Although considerable variation occurs, corresponding to the variation in clinical appearance
- Cutaneous or mucosal lesions generally exhibit intracellular edema of the spinous layer of epithelium
- Edema of the superficial connective tissue which may actually produce a subepidermal vesicle.
- Zone of severe liquefaction degeneration - upper layers of the epithelium,
- Intraepithelial vesicle formation and thinning with frequent absence of the basement membrane.
- Dilatation of the superficial capillaries and lymphatic vessels
- Varying degree of inflammatory cell infiltration, chiefly lymphocytes, but often neutrophils and eosinophils, is also present




Pattern: "Interface dermatitis" (many other diseases have similar histology)
Dying (apoptotic/ necrotic/ dyskeratotic) keratinocytes
Vacuoles in basal layer
Lymphocytes at basal layer (dermal-epidermal junction) and in dermis
Fuzzy dermal-epidermal junction (interface) b/c lymphocytes chewing up the basal layer

TOXIC EPIDERMAL NECROLYSIS (TENS)

- very serious, often fatal, bullous drug eruption
- So severe that large sheets of skin peel off - appearance of a widespread scalding burn.
- Oral erosions may also occur
- Considered to be a confluent form of StevensJohnson syndrome.
- TENS must be differentiated from the Staphylococcal Scalded Skin Syndrome (SSS) – clinically similar even though the latter is a milder disease with a better prognosis

MANAGEMENT

• Identification of the cause should be made if possible.
• If a drug is suspected, it must be withdrawn.
• Infections should be appropriately treated after cultures and/or serologic tests
• For all forms of EM
– symptomatic treatment
– Oral antihistamines, analgesics,
– Local skin care: liquid antiseptics 0.05% chlorhexidine
– soothing mouthwashes
– Topical steroids may be considered.
– Oral antacids

- discrete oral ulcers.
- • SJS and TENS: treatment is generally with high doses of systemic corticosteroids, intravenous immunoglobulin, and thalidomide

KAWASAKI DISEASE

- Also known as mucocutaneous lymph node syndrome and infantile polyarteritis nodosa.
- It is an acute febrile illness of childhood seen worldwide with the highest incidence occurring in Asian children.
- KD is a vasculitis affecting medium size vessels
- Coronary artery aneurysms or ectasia develop in 20% to 25% of untreated children & can lead to 1) ischemic heart disease or 2)sudden death

EPIDEMIOLOGY

- More prevalent in Japan and in children of Japanese ancestry (annual incidence of 112 cases per 100 000 children < 5 yrs old.

ETIOLOGY & PATHOGENESIS

- Etiology of KD remains unknown, (although clinical and epidemiological features strongly suggest an infectious cause)
- Hypothesis - KD is caused by a ubiquitous infectious agent that produces clinically apparent disease only in certain genetically predisposed individuals, particularly Asians

• KD is a vasculitis that predominantly affects the medium-size arteries.

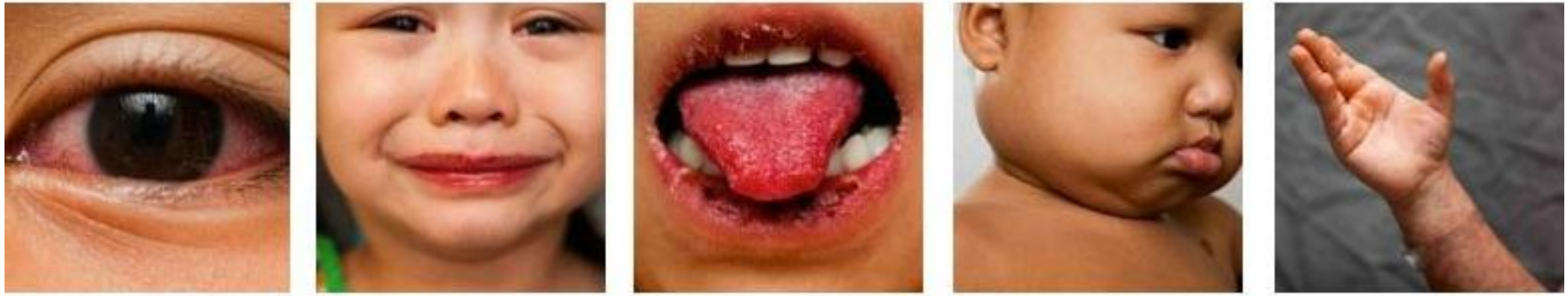
• The coronary arteries are the most commonly involved, although other arteries, such as the popliteal and brachial arteries, can also develop dilation.

• A 3-phase process to the arteriopathy of KD are
• 1 st phase - neutrophilic necrotizing arteritis in the 1st 2 wk of illness Saccular aneurysms may form from this arteritis.
• 2 nd phase – subacute/chronic vasculitis driven by lymphocytes, plasma cells, and eosinophils, which may last weeks to years and results in fusiform aneurysms.
• 3 rd phase - smooth muscle cell myofibroblasts develop which causes progressive stenosis. Thrombi may form in the lumen and obstruct blood flow.

Clinical Features

- Prolonged fever – usually > 5 days duration
- At least 4 of the following 5 features
- 1 – bilateral nonexudative conjunctival injection with limbal sparing
- 2 – erythema of the oral and pharyngeal mucosa with strawberry tongue and red, cracked lips
- 3 – edema and erythema of the hands and feet
- 4 – polymorphic rash usually truncal
- 5 – nonsuppurative cervical lymphadenopathy, usually unilateral, with node size >1.5 cm

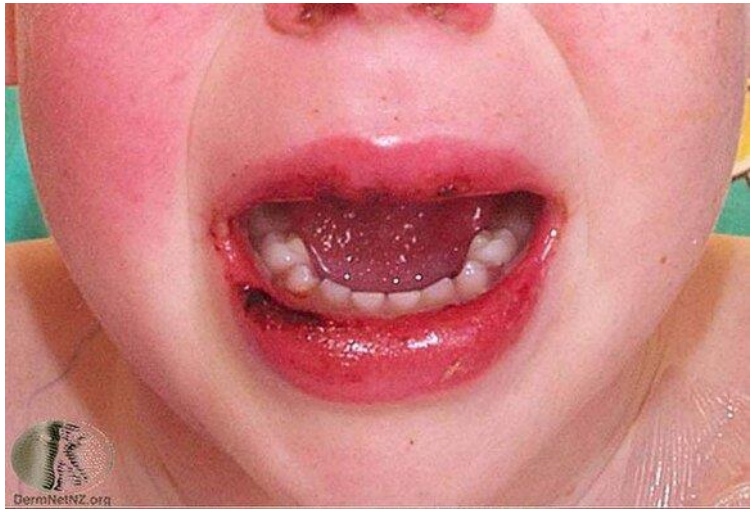
Signs & Symptoms of Kawasaki Disease



Images courtesy of the Kawasaki Foundation

Oropharyngeal changes

- Erythema, dryness, swelling and peeling of lips - lipstick sign
- Lips may bleed
- Erythema of oropharyngeal mucosa
- Strawberry tongue
- No Koplik’s spots or oral ulceration or exudates in KD



MANAGEMENT

- Acute stage : - IVIG 2 gm/kg over 10 – 12 hrs - Aspirin 80-100 mg/kg/day divided every 6 hr orally until patient is afebrile for at least 48 hr
- Convalescent Stage : - Aspirin 3-5 mg/kg/day once daily orally until 6- 8 wk after illness onset if normal coronary findings throughout course

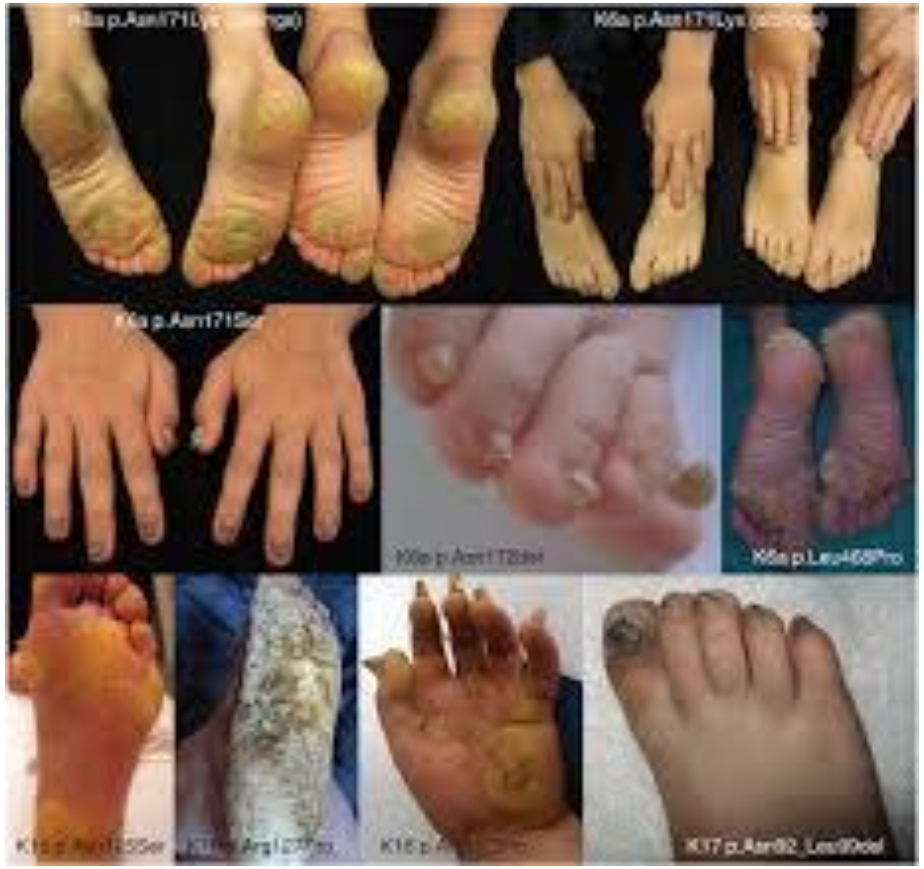
PACHYONICHIA CONGENITA

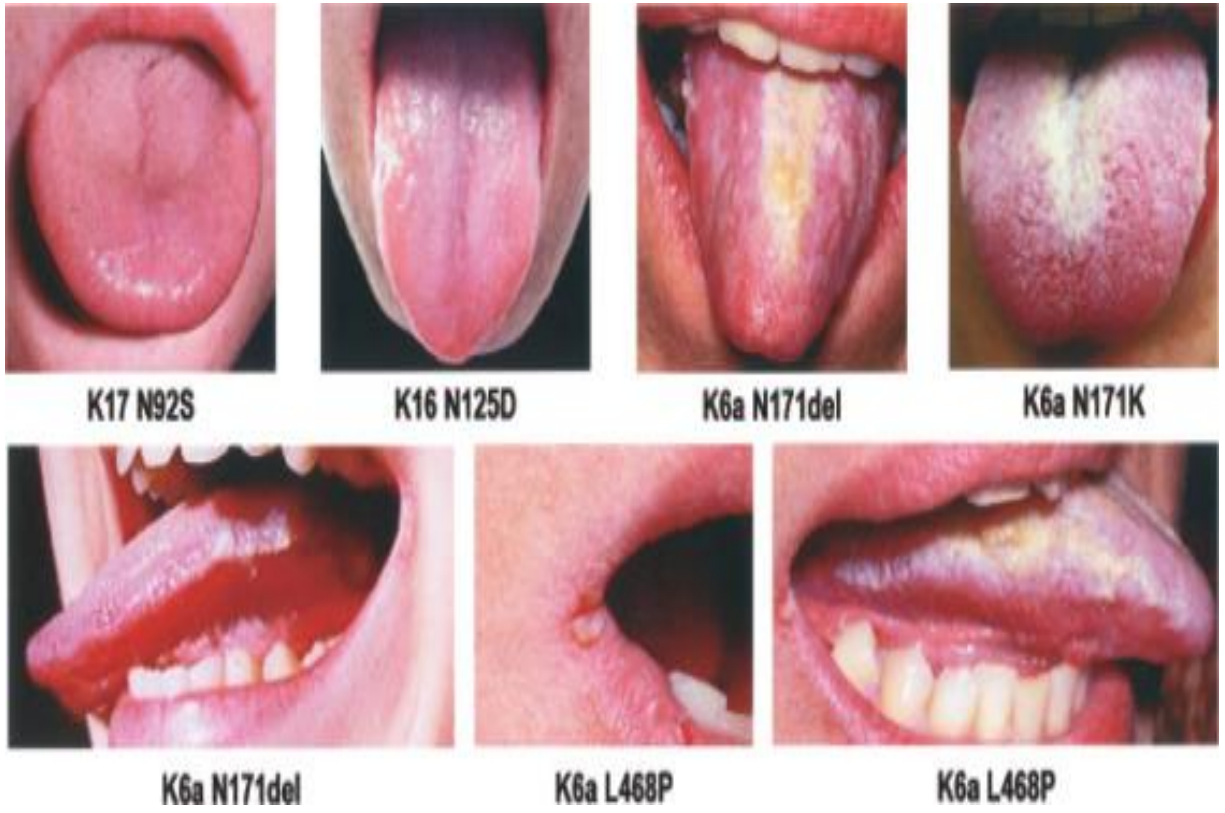
- An autosomal dominant condition that primarily affects the nails & skin.
- Signs & symptoms usually become apparent within few months of life
- Pachyonichia congenita tarda – a rare form of the condition – Appears in adolescence or early adulthood.
- This disorder does not affect lifespan, but patients do experience constant pain.

CLINICAL MANIFESTATIONS

- Hypertrophic nail dystrophy: A hallmark of this disorder is thickened & abnormally shaped fingernails & toenails.
- Painful calluses & blisters: Palms & the feet will have painful blisters & calluses : makes it almost impossible for the child to walk.
- Palmoplantar keratoderma
- Follicular hyperkeratosis: Bumps around hair follicles in areas that get lots of friction: waist, hips, knees, elbows
- Oral leukokeratosis: thick, white patches inside the mouth and on the tongue
- Palmoplantar hyperhidrosis: excessive sweating on the palms & soles

- Angular cheilitis: sores at the corner of the mouth
- Teeth at or before birth
- Laryngeal involvement with a white keratin film on the larynx causing hoarseness or breathing difficulties
- Intense pain on first bite: The pain is near the jaw or ears & lasts 15-25 seconds when beginning to eat or swallow – More common in younger children & often confused with ear problems.




K17 N92S
K16 N125D
K6a N171del
K6a N171K
K6a N171del
K6a L468P
K6a L468P

Diagnosis

- Usually by its clinical appearance
- Skin biopsy of affected tissue show only nonspecific changes
- Molecular genetic testing – to detect the affected keratin genes

MANAGEMENT

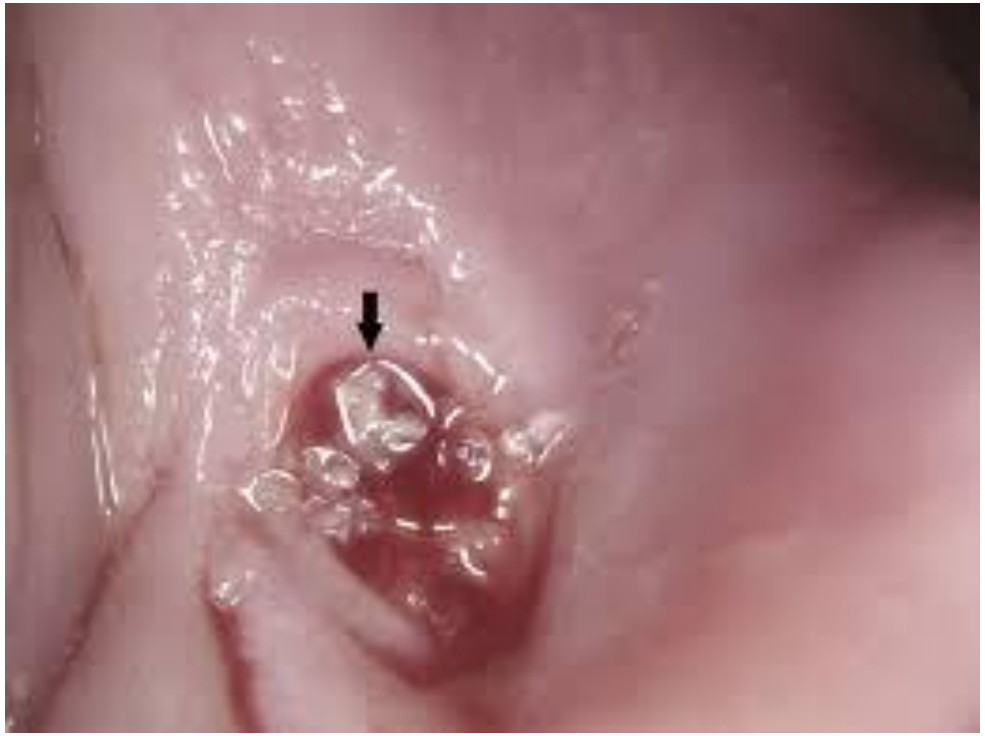
Effectiveness of treatment depends on the severity of the specific problem .

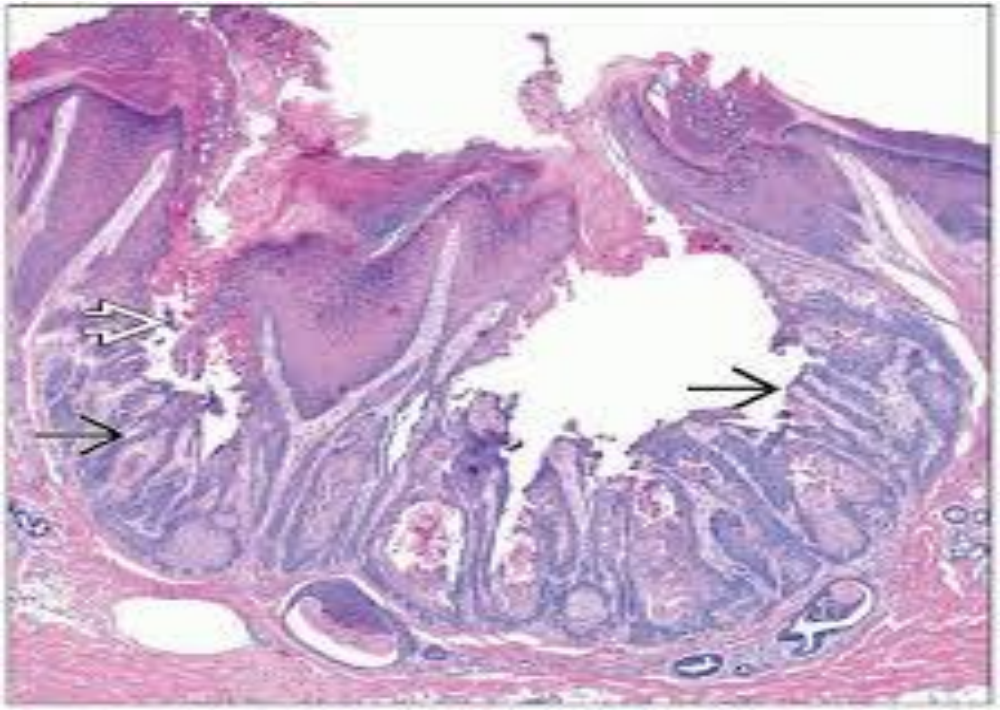
- Treatment might include:
  - Emollients – soften the skin
  - Keratolytics eg salicylic acid ointment (for treatment of scaly skin diseases), urea cream (a natural moisturizing factor) or other forms of heel balm
  - Mechanical debridement – Pain relief
  - Oral retinoids eg acitretin

WARTY DYSKERATOMA

- Warty dyskeratoma, also known as an Isolated dyskeratosis follicularis, is a benign epidermal proliferation with distinctive histologic findings that may mimic invasive squamous cell carcinoma and commonly manifests as an umbilicated (Having a central mark or depression resembling a navel) lesion with a keratotic plug, WD have some histopathologic similarities to viral warts but it's not caused by HPV and the majority of these lesions display overall histopathologic features consistent with a follicular adnexal neoplasm.
- Usually limited to the head, neck, scalp or face and vulva.

- Oral Lesions particularly the hard palate, and genital involvement have been reported.
- it can also be thought of as one of the manifestations of focal acantholytic dyskeratosis, an epidermal reaction pattern that can be seen in several disorders, including Darier's disease and Grover's disease.
- But the main Difference between Darier disease and Warty dyskeratoma, is that Darier disease inherited dermatosis (autosomal dominant) consisting of multiple keratotic papules on the face, trunk, and extremities, while WD occurs as an isolated, noninherited, single keratotic nodule mainly confined to the head and neck as mentioned earlier.

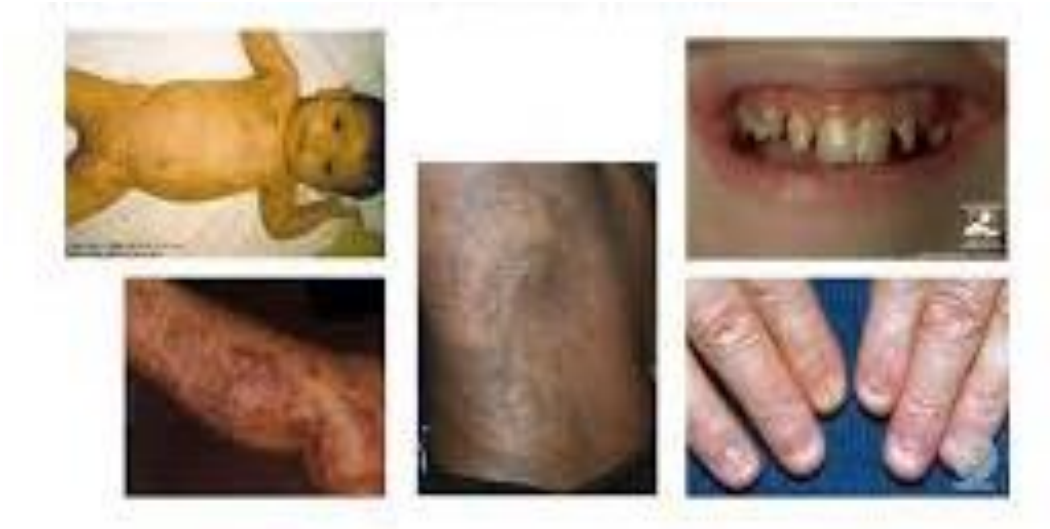


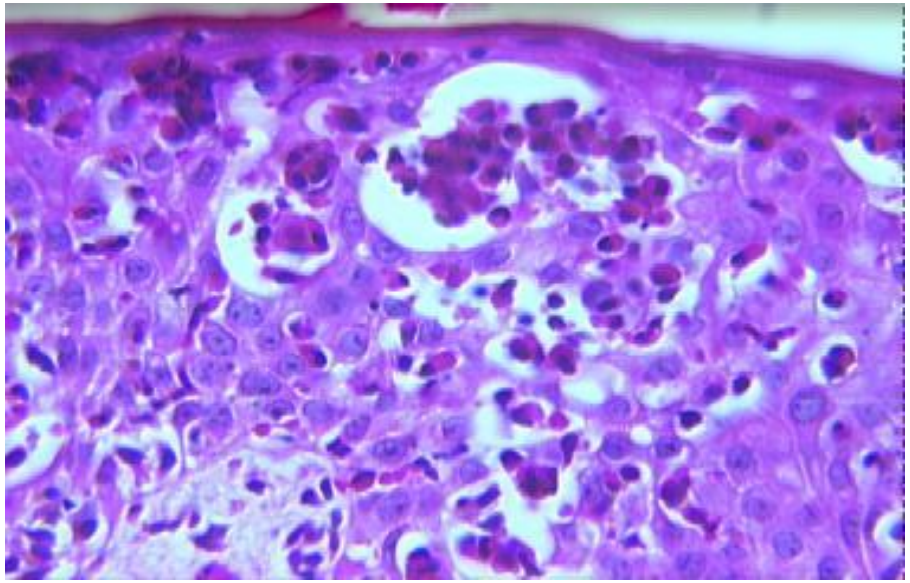


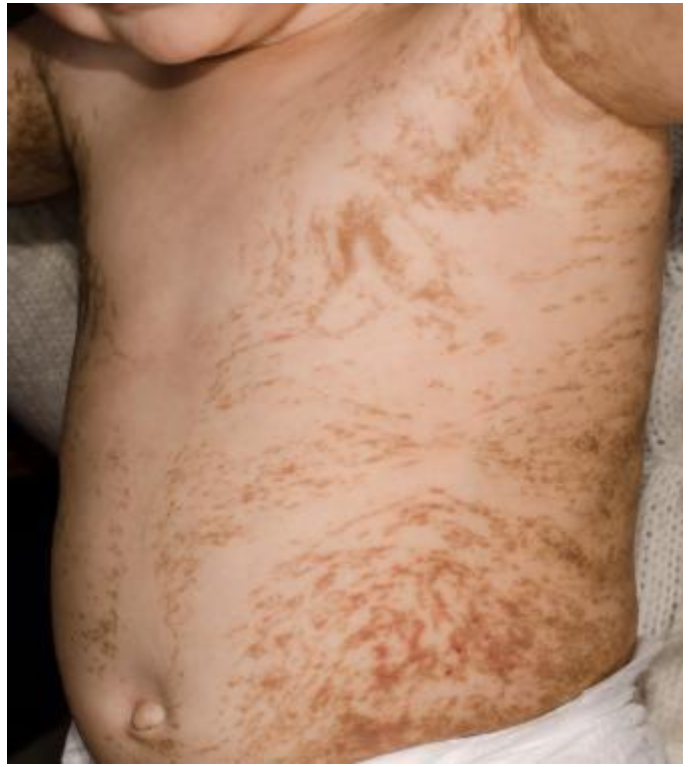
INCONTINENTIA PIGMENTI

- Incontinentia pigmenti (IP) is a rare X-linked dominant genetic disorder that affects the skin, hair, teeth, nails and central nervous system. It is named from its appearance under a microscope.
- The disease is characterized by skin abnormalities that begin in childhood, usually a blistering rash which heals, followed by the development of harder skin growths.
- The skin may develop grey or brown patches which fade with time. Other symptoms can include hair loss, dental abnormalities, eye abnormalities that can lead to vision loss and lined or pitted fingernails and toenails.

- Associated problems can include delayed development, intellectual disability, seizures and other neurological problems.
- Most males with the disease do not survive to childbirth
- Incontinentia pigmenti is caused by a mutation in the IKBKG gene, which encodes the NEMO protein, which serves to protect cells against TNF-alpha-induced apoptosis. A lack of IKBKG therefore makes cells more prone to apoptosis.
- There is no specific treatment; individual conditions must be managed by specialists.

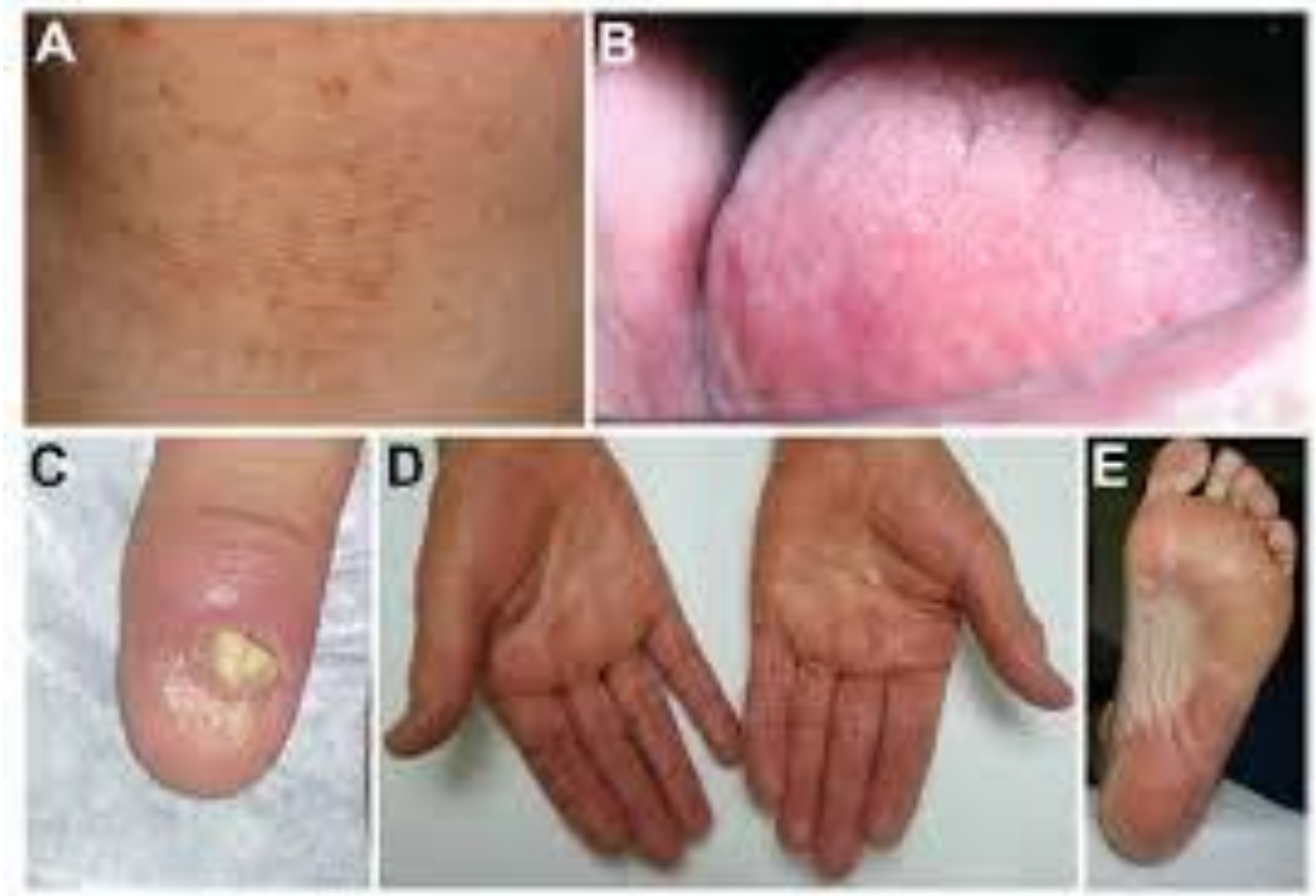






DYSKERATOSIS COGENITA

- Dyskeratosis congenita (DKC), is a rare progressive congenital disorder with a highly variable phenotype.
- DKC can be characterized by cutaneous pigmentation, premature graying, dystrophy of the nails, leukoplakia of the oral mucosa, continuous lacrimation due to atresia of the lacrimal ducts, often thrombocytopenia, anemia, testicular atrophy in the male carriers, and predisposition to cancer. Many of these symptoms are characteristic of geriatrics, and those carrying the more serious forms of the disease often have significantly shortened lifespans.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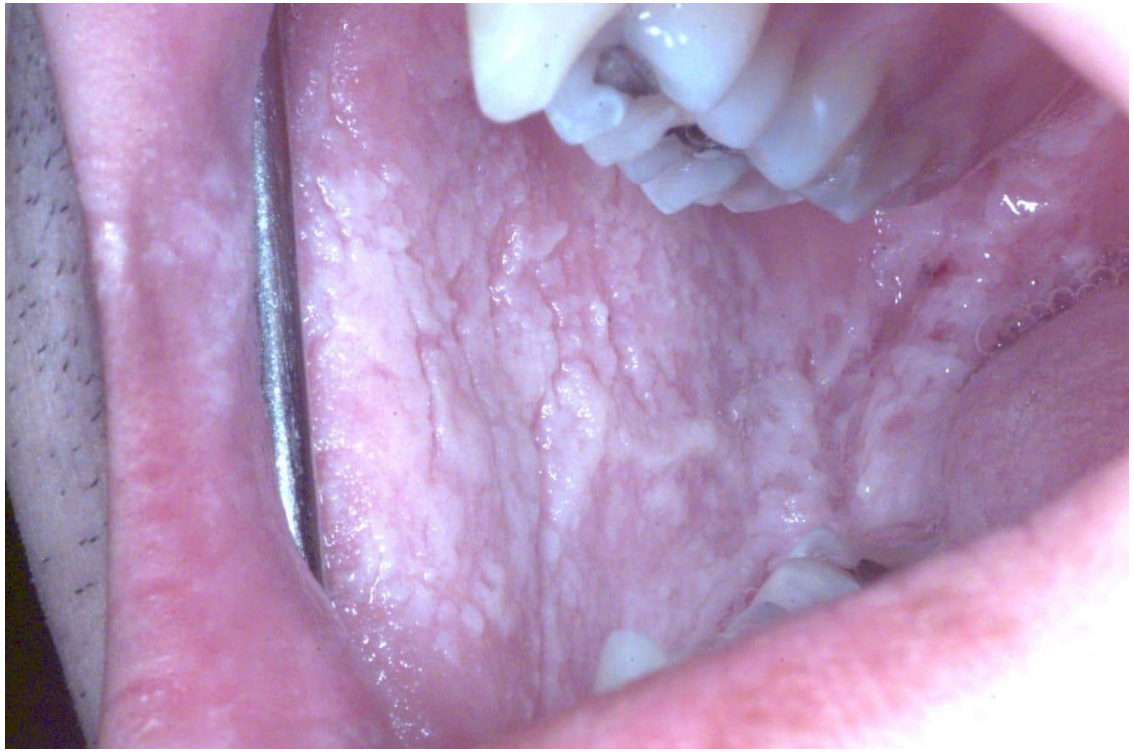
WHITE SPONGE NEVUS

- Familial white folded dysplasia appearing to follow a hereditary pattern as an autosomal dominant trait.
- Other names
- Cannons disease
- Familial white folded dysplasia
- Hereditary mucosal leukokeratosis
- Nevus of cannon
- White folded gingivostomatitis
- Exfoliative leukoedema

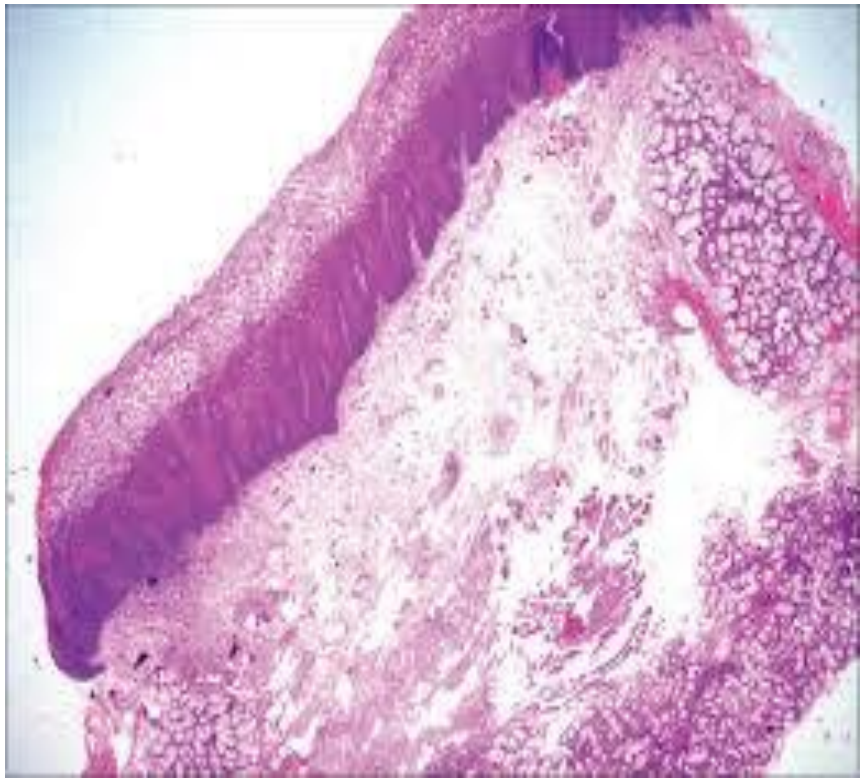
Clinical features

- Below 20years of age or in early childhood
- No gender predilection
- Oral lesions widespread involving cheeks palate gingiva floor of mouth and portions of tongue . Mucosa appears thickened or folded or corrugated with a soft spongy texture and peculiar white opalescent hue.Sometimes minimal folding present.
- White ragged areas sometimes present can be removed by gentle rubbing without ensuing bleeding. Lesions themselves asymptomatic.
- Sometimes oral lesions accompanied by similar lesions of other mucosal surfaces like vagina, labia, anus, rectum and nasal cavity.



Histologic features

- Epithelium generally thickened showing both hyperparakeratosis and acanthosis and the basal layer is intact.
- Cells of entire spinous layer continuing to very surface show intracellular edema.
- Vacuollated cells may show pyknotic nuclei
- Parakeratin plugs running deep into spinous layer



Treatment and prognosis

- No treatment.
- Benign condition.
- Excellent prognosis.

PEMPHIGUS

CLASSIFICATION

- • PEMPHIGUS VULGARIS
- – GENERALIZED
- – LOCALISED
- – DRUG INDUCED
- • PEMPHIGUS FOLIACEUS
- – GENERALIZED
- – LOCALIZED
- – FOGO SELVAGEM: ENDEMIC
- – DRUG INDUCED
- • PARANEOPLASTIC PEMPHIGUS
- • IgA PEMPHIGUS

EPIDEMIOLOGY

- • PV
- – RARE
- – JEWS, MEDITERRANEAN, INDIAN SUBCONTINENTS
- • PF
- – RARE
- – ENDEMIC:-
- FOGO SELVAGEM
- • AGE:- 40-60YRS
- • SEX:- MALE=FEMALE

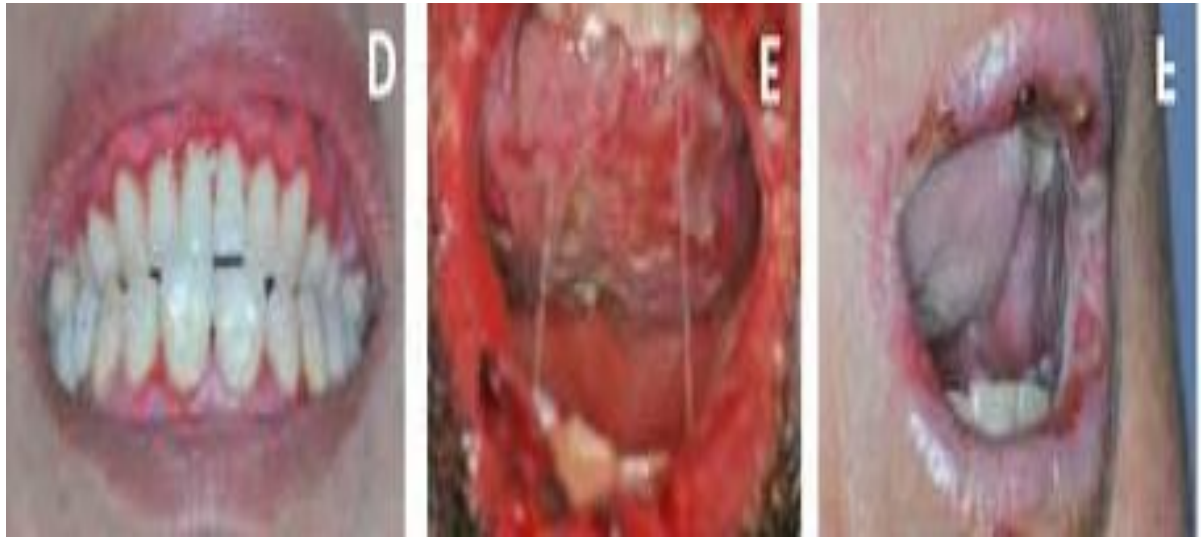
ETIOLOGY

- AUTOIMMUNE
- ACANTHOLYSIS – IgG AB AGAINST DESMOGELIN – PV:-DESMOGELIN 3 & 1 – PF:-DESMOGELIN 1 • INTERFERE CALCIUM-SENSITIVE ADHESION FUNCTION

CLINICAL FEATURES

PEMPHIGUS VULGARIS

- • start in oral mucosa
- • initially locallised
- • painful & tender
- • round oval vessicle& bullae with serous content
- • flacid, easily ruptured
- • localized or generalized
- • nicolsky sign
- • sites:- scalp face chest axilla
- • mucous membrane



PEMPHIGUS FOLIACEOUS

- • seborrheic area
- • scaly, crustederythematous

PEMPHIGUS VEGETANS

- • intertriginous region, perioral area, neck and scalp drug induced pv • captopril, • d-penicillamine

PARANEOPLASTIC PEMPHIGUS

- • mucous membrane primarily
- • combine features of pemphigus vulgaris & erythema multiforme


Mucous membrane pemphigoid
Pemphigus vulgaris
Paraneoplastic pemphigus
Stevens-Johnson syndrome
Oral lichen planus
A
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DIAGNOSIS

- DIFFICULT – FROM ALL FORM OF ACQUIRED BULLUS DISEASE
- BIOPSY
- MICROSCOPY
- AB DIRECTION IN SERUM
- DIRECT IF
- ELISA

MANAGEMENT

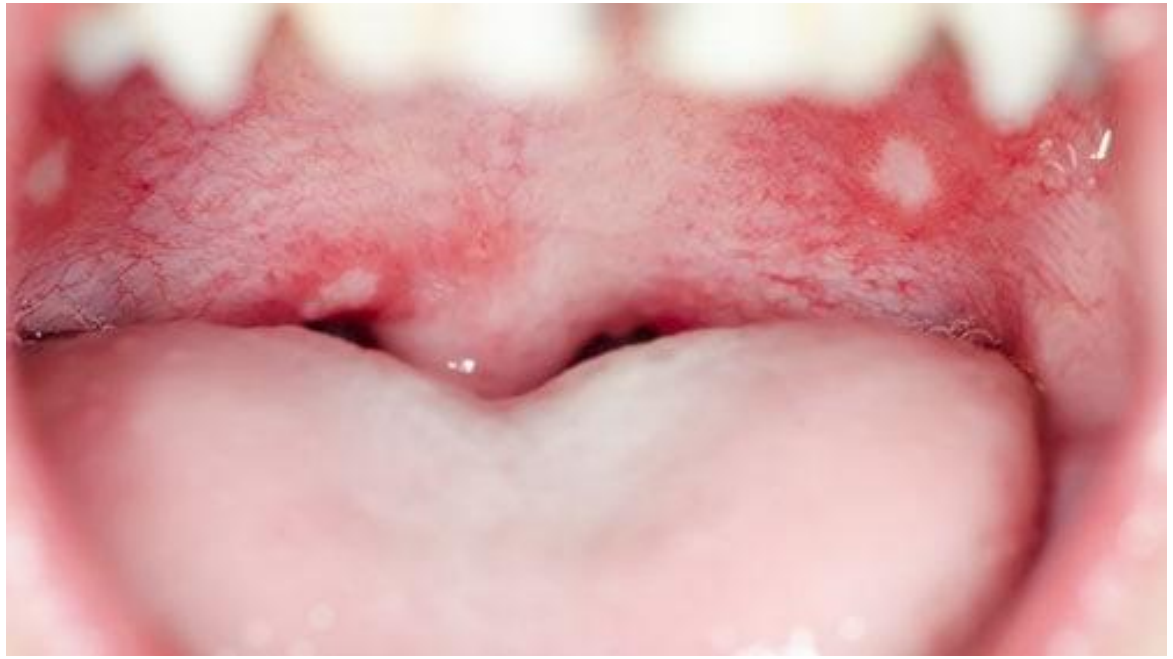
- • GLUCOCORTICOIDS – PREDNISOLONE (60-100mg)
- • CONCOMITANT IMMUNOSUPPRESSIVE THERRAPY
  – AZATHIOPRINE (2-3mg/kg)
  – MYCOPHENOLATE MOFETIL(1g tds) – IV-Ig (2g/kg every 3-4weeks) – RITUXIMAB
- • OTHER MEASURES
- – CLEANSING BATH
- – WET DRESSING
- – ANTIMICROBIAL
- – FLUID & ELECTOLYTE CORRECTION
- • MONITORING

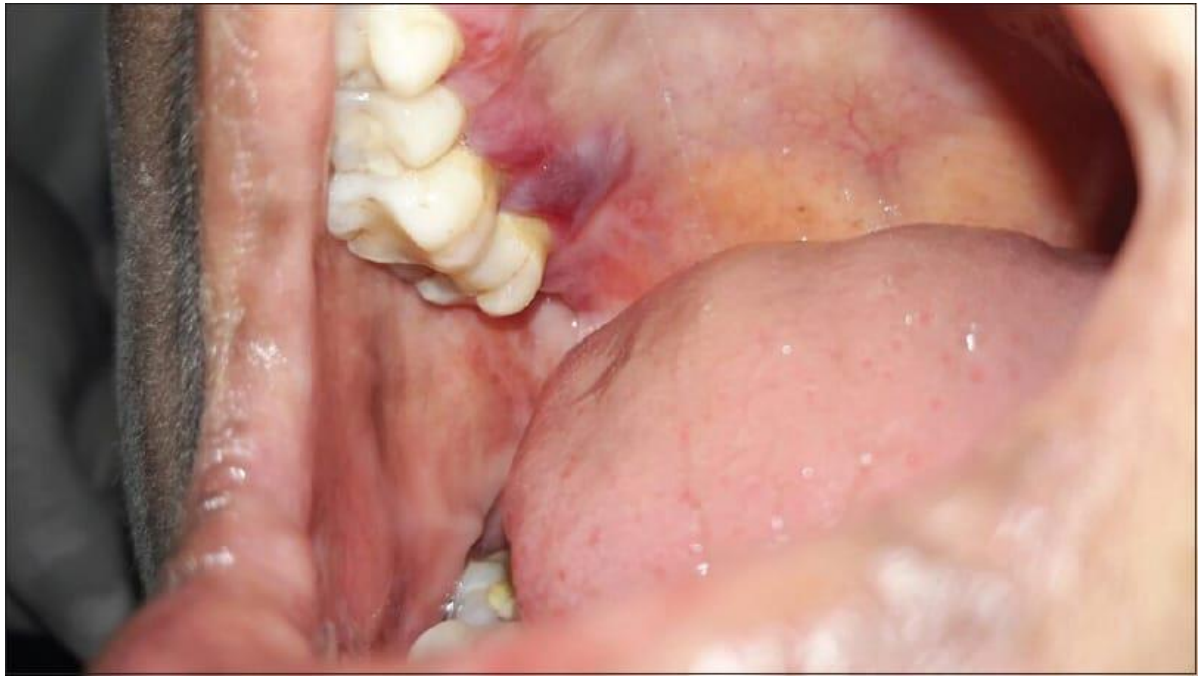
BULLOUS PEMPHIGOID

- Common subepidermal blistering autoimmune disease of skin due to igG antibodies to the hemidesmosomal antigens bullous pemphigoid antigen 1 and 2
- Affects elderly patients and presents as large tense bulla involving trunk extremities and intertriginous areas
- Nikolsky sign negative
- Oral lesions present in 1/3rd of patients

Clinical features

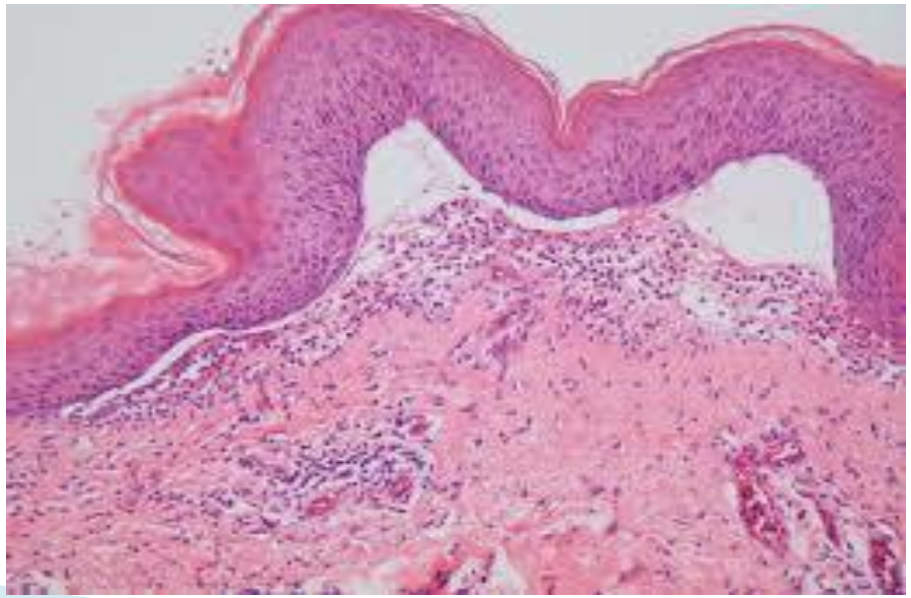
- Autoimmune blistering disorder
- Common in elderly
- Split is at the basement membrane zone (deeper than p vulgaris)
- Crops of tense fluid filled blisters often with surrounding erythema
- Itchy
- Can be localised or widespread
- Oral mucosal involvement less frequent than p vulgaris
- Usually less severe





Histologic features

- Subepidermal vessicle often filled with eosinophils
- Superficial perivascular mixed inflammatory cell infiltrate rich in eosinophils
- In the cell poor variant only scant inflammatory cell infiltrate is present
- Early lesions present with spongiosis and infiltrate of eosinophils



Treatment

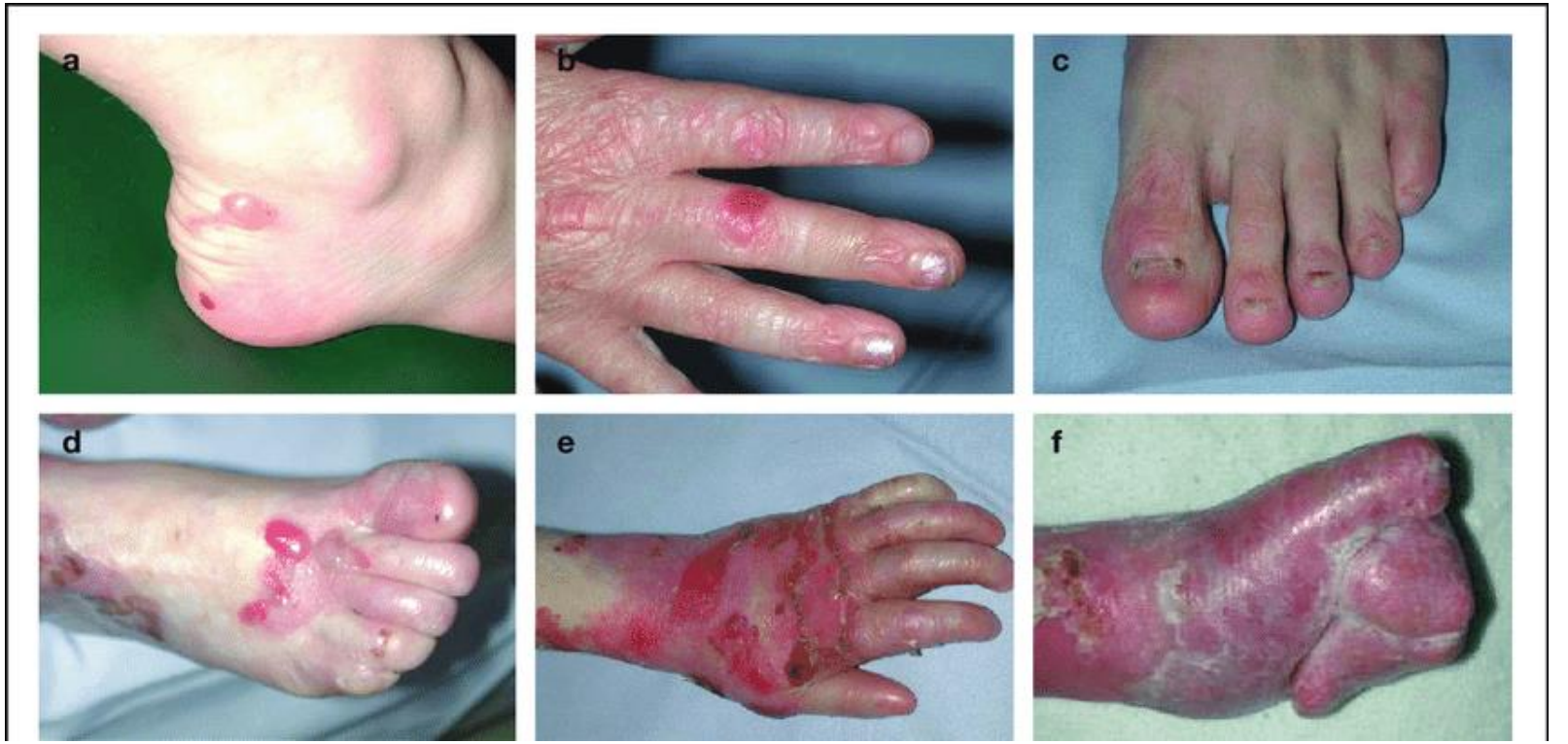
- Systemic steroids
- Immunosuppresants
- Other: Rituximab, dapsone

EPIDERMOLYSIS BULLOSA

- An inherited disorder that causes painful fluid filled blisters to form on the skin
- There is no cure but there’s treatment to help the pain.

TYPES OF EB

1) EB simplex (intraepidermal skin separation)
2) Junctional EB (skin separation in lamina lucida or central basement membrane zone)
3) Dystrophic EB (sublamina densa basement membrane zone separation)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CAUSES

- Genetic factor (both dominant and recessive)
- More severe when attained recessively.

Clinical features

Blistering of skin
Tooth decay
Deformed or absent nails
Internal blistering in throat stomach and intestine
Scalp blistering
Loss of hair
Excessive sweating
Hardening of skin on feet or hands
Thin skin appearance and white bumps
Difficulty in swallowing

Management

- Dental care
- Skin care
- Physical therapy
- Steroids

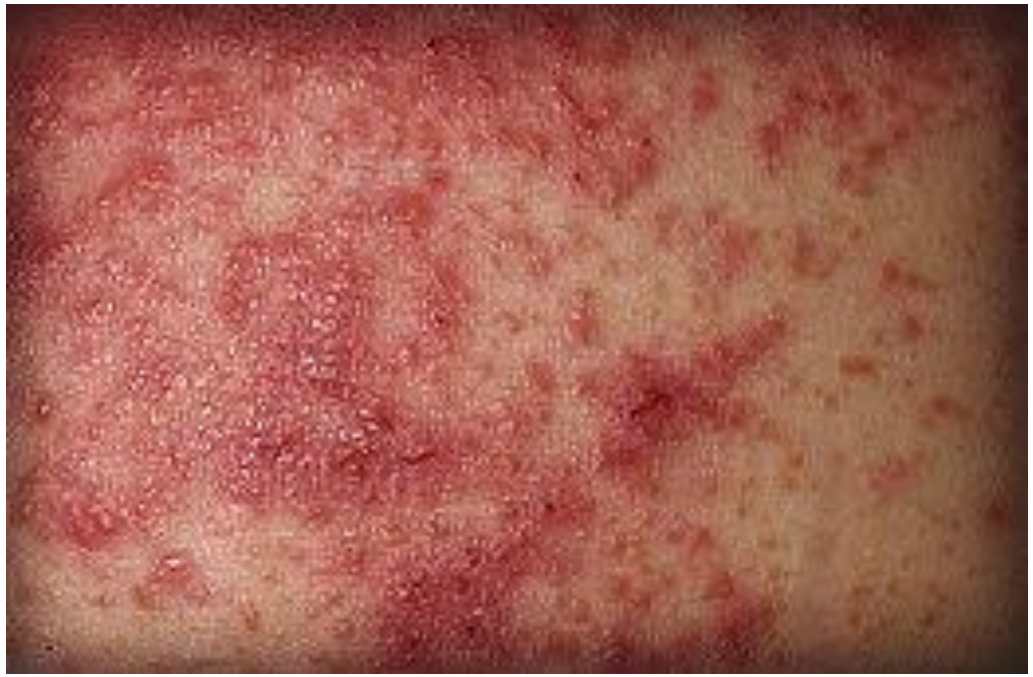
DERMATITIS HERPETIFORMIS

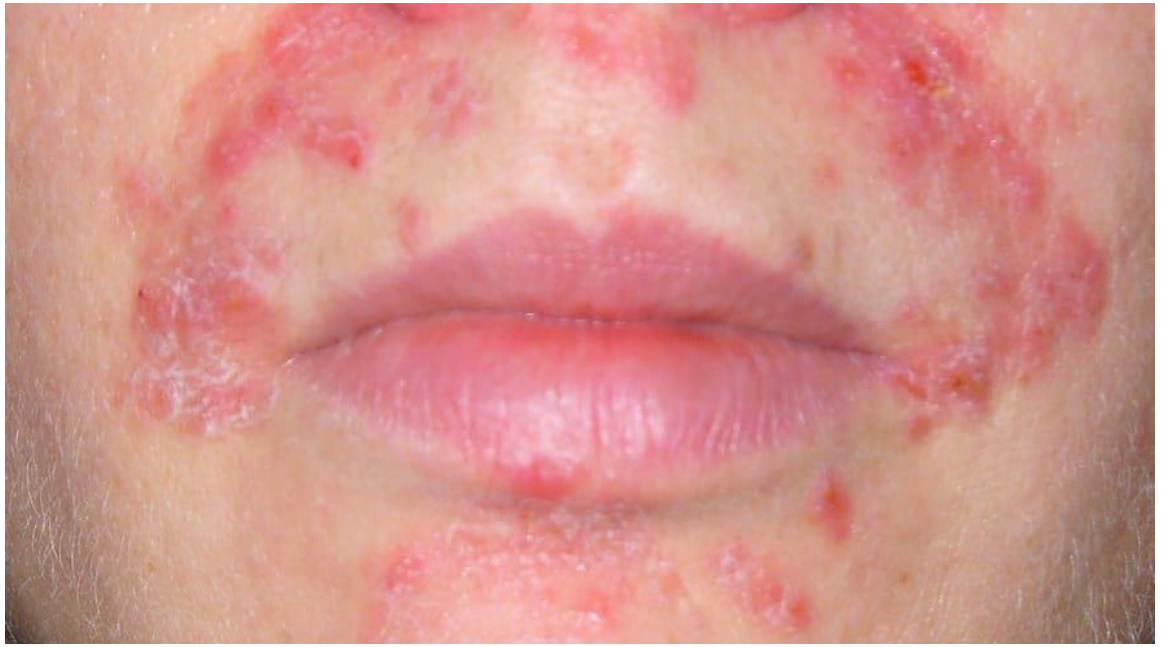
- Dermatitis herpetiformis (DH) is a chronic autoimmune blistering skin condition,[3] characterised by blisters filled with a watery fluid[4] that is intensely itchy.
- DH is a cutaneous manifestation of Coeliac disease.[5] Despite its name, DH is neither related to nor caused by herpes virus: the name means that it is a skin inflammation having an appearance similar to herpes.

- The signs and symptoms of DH typically appear around 30 to 40 years of age, although all ages may be affected.
- Although the first signs and symptoms of dermatitis herpetiformis are intense itching and burning, the first visible signs are the small papules or vesicles that usually look like red bumps or blisters.
- The rash rarely occurs on other mucous membranes, excepting the mouth or lips.

- The symptoms range in severity from mild to serious, but they are likely to disappear if gluten ingestion is avoided and appropriate treatment is administered. The rash caused by dermatitis herpetiformis forms and disappears in three stages.
- In the first stage, the patient may notice a slight discoloration of the skin at the site where the lesions appear.

- In the next stage, the skin lesions transform into obvious vesicles and papules that are likely to occur in groups.
- Healing of the lesions is the last stage of the development of the symptoms, usually characterized by a change in the skin color. This may result in areas of the skin turning darker or lighter than the color of the skin on the rest of the body. Because of the intense itching, patients usually scratch, which may lead to the formation of crusts.





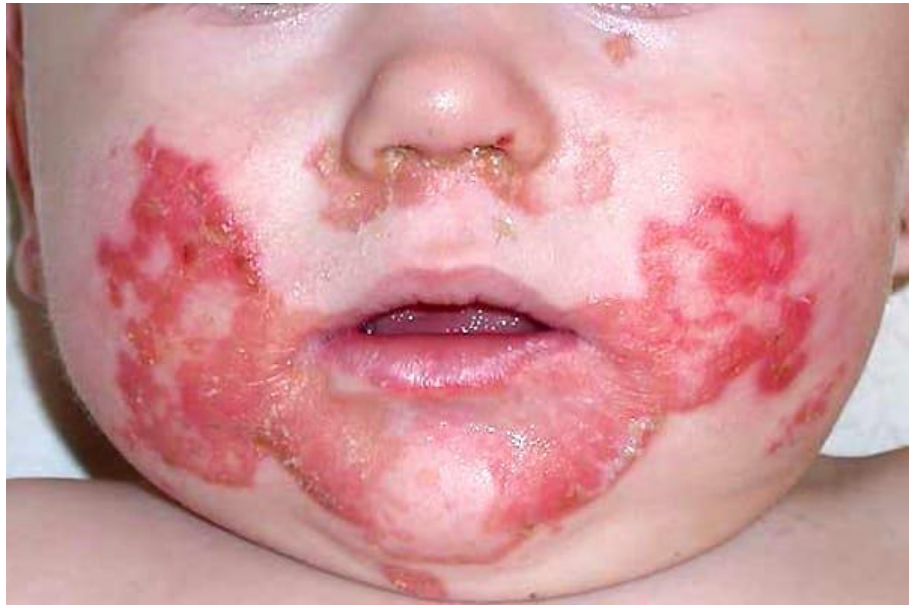
Treatment

- 1. Gluten free diet
- 2. Dapsone - drug of choice.

ACRODERMATITIS ENTEROPATHICA

- Acrodermatitis enteropathica is an autosomal recessive metabolic disorder affecting the uptake of zinc through the inner lining of the bowel, the mucous membrane.
- Individuals with acrodermatitis enteropathica may present with the following:
- Blistering of skin
- Dry skin
- Emotional lability

- Glossitis
- Pustule
- Alopecia (loss of hair from the scalp, eyebrows, and eyelashes) may occur.
- Skin lesions may be secondarily infected by bacteria such as Staphylococcus aureus or fungi such as Candida albicans. These skin lesions are accompanied by diarrhea.



SYSTEMIC LUPUS ERYTEMATOSUS

- SLE is a complex autoimmune disease.
- Chronic multisystem inflammatory disease associated with abnormalities of immune system.

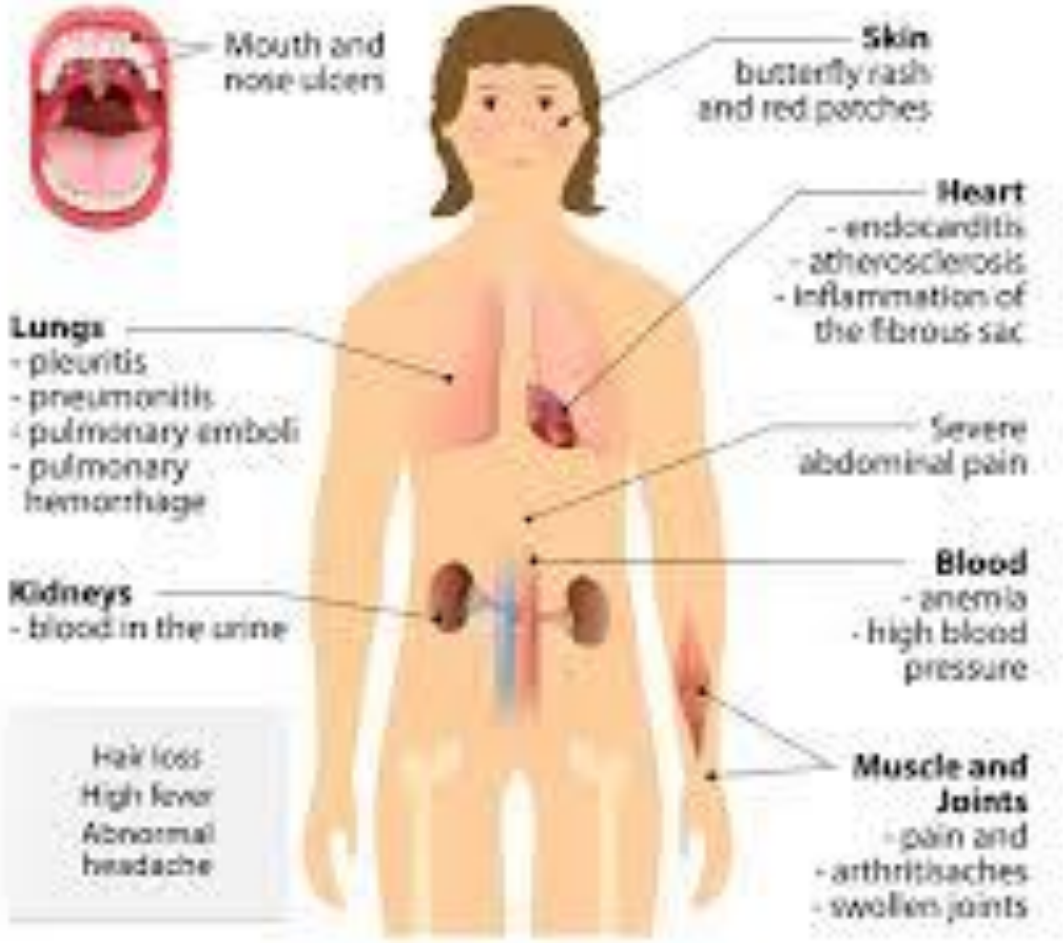
ETIOLOGY

- Unknown
- Propable causes
- Genetic
- Hormonal
- Enviromental
- Certain medications

CLINICAL FEATURES

- Commonly affects : skin, muscle, lining if lungs, heart, nervous tissue & kidneys.
- Skin manifestations :
- oropharyngeal ulcers,
- cutaneous vascular lesions,
- alopecia,
- butterfly rash.

Systemic lupus erythematosus



ORAL MANIFESTATIONS

- Oral lesions similar to DLE, except hyperemia, edema, and extension of lesions are more pronounced.
- Greater tendency for bleeding,petechiae, and superficial ulceratiobs surrounded by red halo.
- Superimposed oral moniliases.
- xerostomia

MANAGEMENT

- NSAIDS: acetaminophen
- CORTICOSTERIODS: Prednisolone
- ANTI-MALARIAL : Hydroxychloroquine

SYSTEMIC SCLEROSIS

- Multisystem disorder
- Unknown etiology
- Thickening of skin caused by accumulation of connective tissue (collagen types I and III)
- Involvement of visceral organs

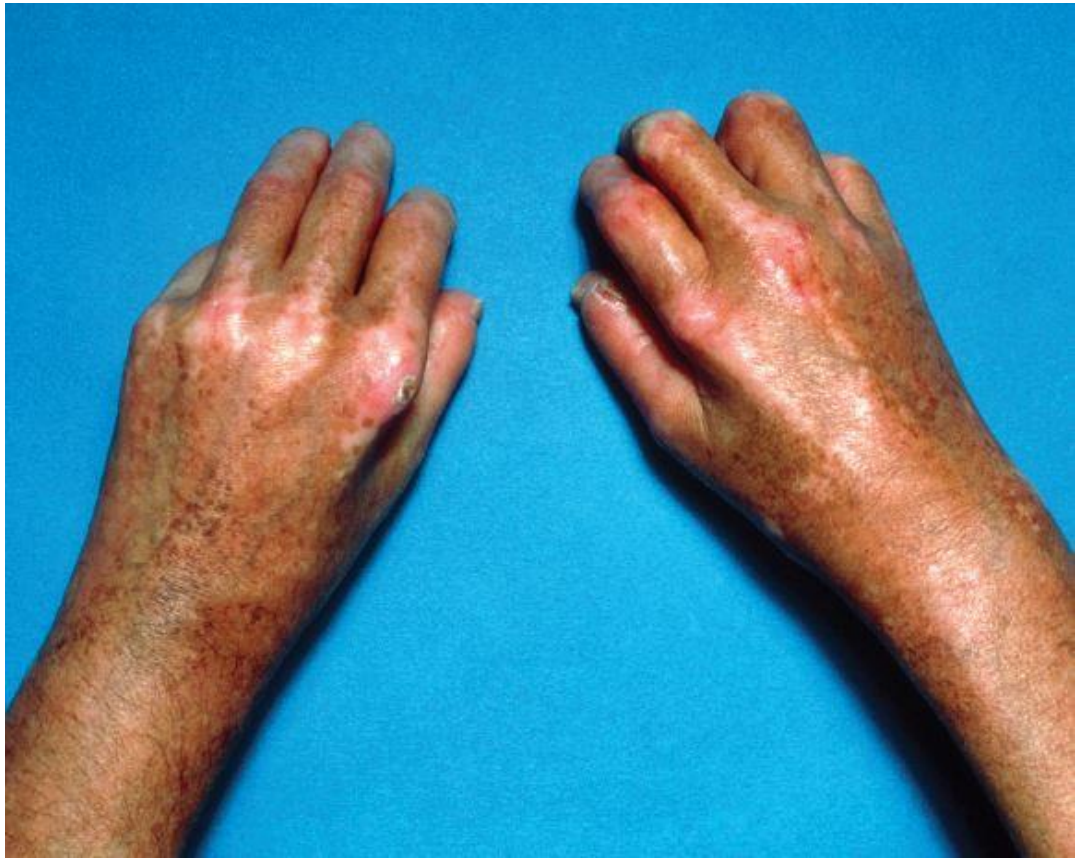
ETIOLOGY

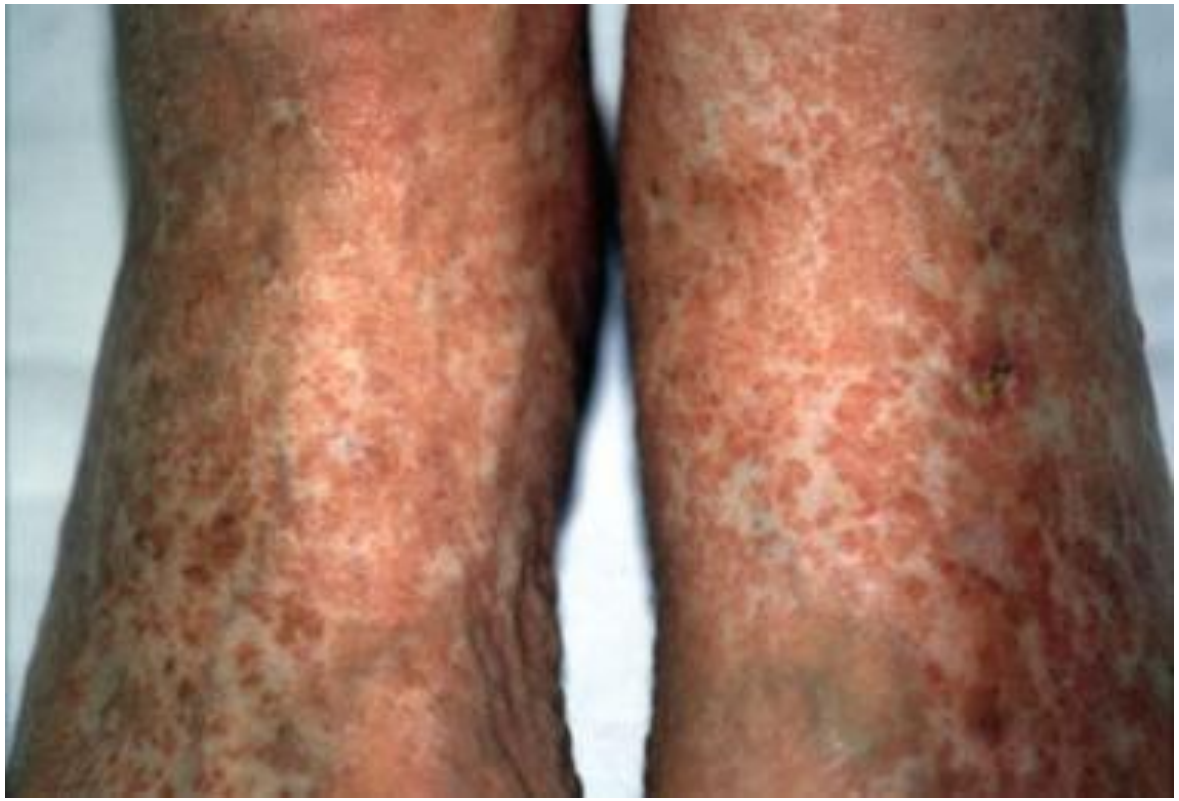
- Unknown
- Environmental Exposures
- Silica exposure in men conferred increased risk
- Silicone breast implants: no definite risk identified
- Aniline laced Contaminated rapseed oil in Spain
- Vinyl chloride exposure increased risk of SSc like disorder: Eosinophilic Fasciitis
- bleomycin
- L-tryptophan: Eosinophilia Myalgia syndrome
- Genetic factors

SKIN MANIFESTATIONS

- Early stages:

• Perivascular infiltrate which are primarily T cells.
• Skin swelling which eventually becomes skin thickening.
• Involves the hands and/or feet (distal).
• Late Stages:
• Finger-like projections of collagen extend from the dermis to the subcutaneous tissue to anchor skin deeper.
• Skin becomes firm, thick and tight.
• Skin thickening moves proximally.
• Fibroblasts and collagen deposition.
• Hair and wrinkles overlying area of skin thickening disappears.





MANAGEMENT

Stable disease: no treatment

- Advancing diffuse skin involvement:
- Methotrexate
- Mycophenolate
- Current trial with Tocilizumab (Actemra)
- D-penicillamine 125 mg/day.
- Research on various anti-fibrosis therapies is being performed (imatinib, Gleevac).

EHLERS-DANLOS SYNDROME

- Hereditary connective tissue disorder (autosomal dominant or recessive traits)
- Prevalence: 1/10000 to 1/25000 20,000-50,000 EDS patients in the US
- Signs/Symptoms
- Joint hypermobility
- Skin hyperextensibility
- Tissue fragility Pain

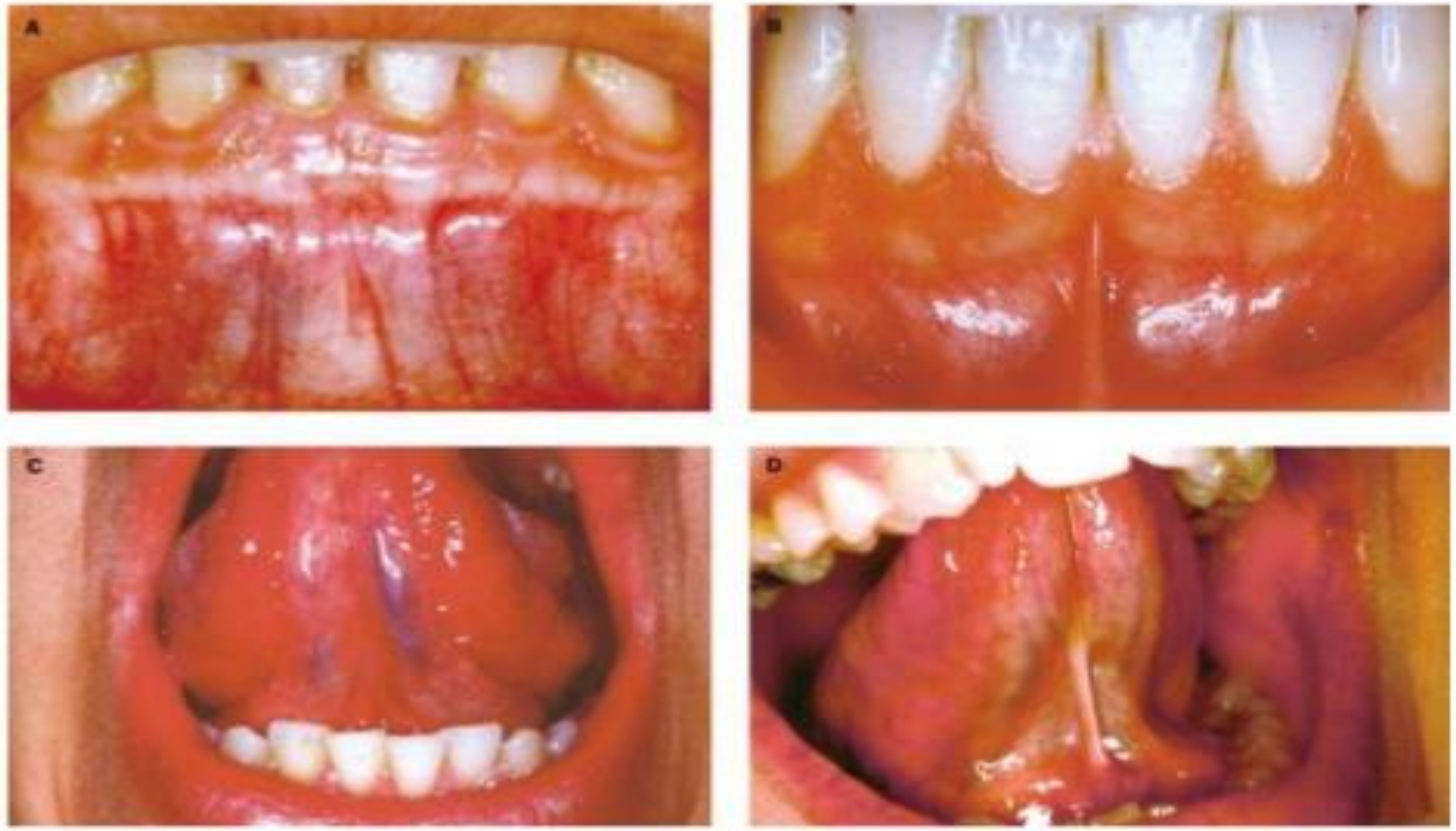
CLASSIFICATION

- Six subtypes of EDS
  - Classical
  - Vascular
  - Hypermobility
  - Kyphoscoliotic
- Arthrochalasia
  - Dermatosparaxis

ORAL MANIFESTATIONS

- Oral mucosa of normal color but excessively fragile and brusied easily.
- Slightly retarded wound healing
- Gingival bleeding after tooth brushing
- Gingival hyperplasia
- Fibrous nodules
- Hypermobility of TMJ causing repeated dislocations
- Alteration in structure of teeth
- Extensive periodontal destruction.

EHLER DANLOS SYNDROME ORAL MANIFESTATION



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MANAGEMENT

- Patient education
- Prevention and early recognition of injuries/complications
- Monitoring & Interventions
- Particular manifestations
- Complications with each forms of EDS
- Medical alert device.
- No medical treatments

FOCAL DERMAL HYPOPLASIA SYNDROME/ GOLTZ SYNDROME

- Uncommon genetic disorder characterized by distinctive skin abnormalities and a wide variety of defects affecting eyes, teeth, skeletal, urinary, gastrointestinal, cardiovascular and central nervous system.
- Etiology

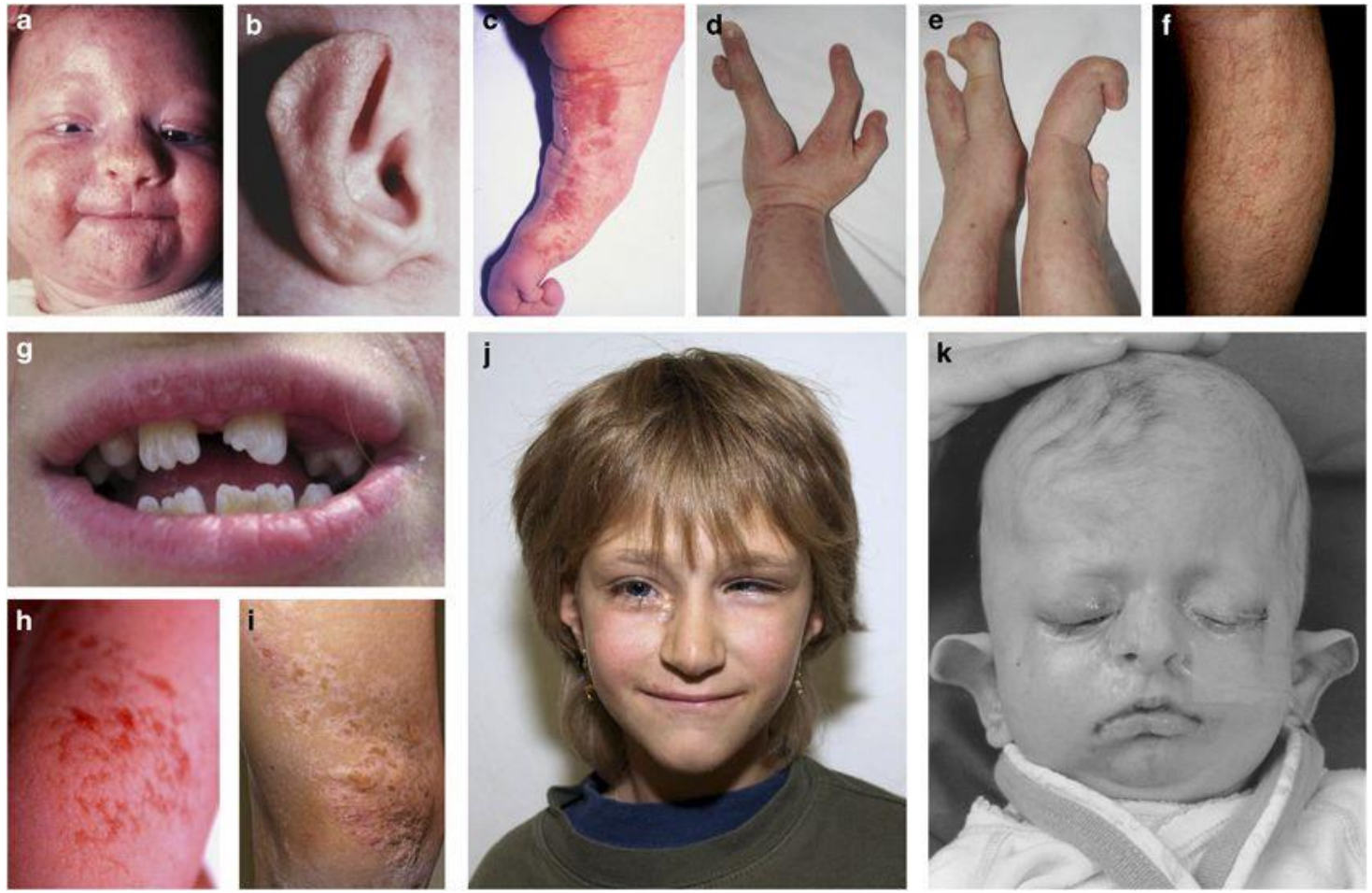
Xlinked dominant inheritance pattern and lethal in males.

Clinical features

- It is a multisystem disorder characterized primarily by skin manifestations to the atrophic and hypoplastic areas of skin which are present at birth.
- These defects manifest as yellow-pink bumps on the skin and pigmentation changes.
- The disorder is also associated with shortness of stature and some evidence suggests that it can cause epilepsy.

Oral manifestation

- Papillomas of lips, buccal mucosa, gingiva.
- Teeth are defective in size shape or structure.
- Microdontia
- Enamel hypoplasia
- Cleft lips and palate occasionally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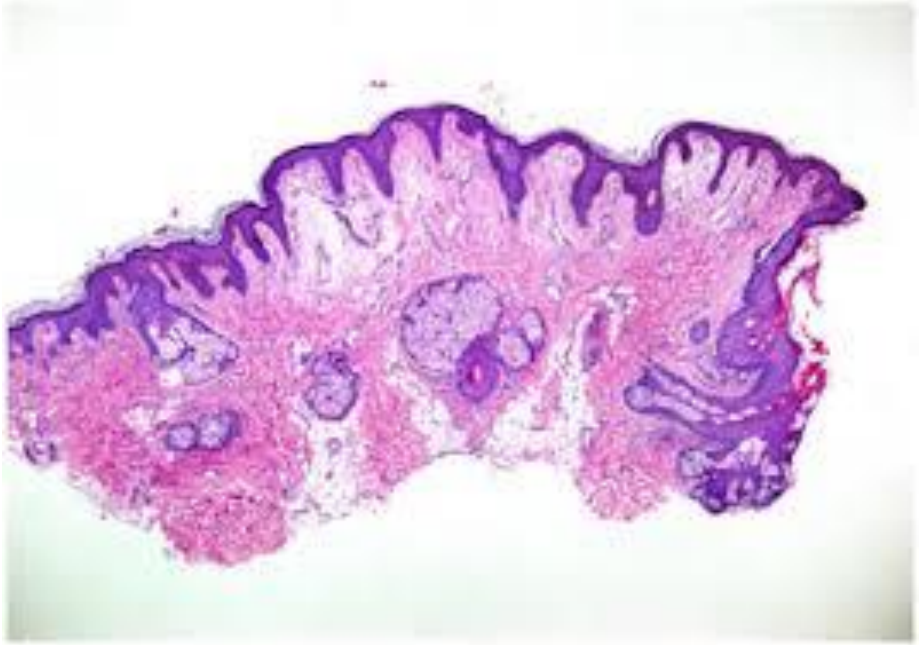
Histological features

- A range of histopathologic features, including some underreported findings (increased papillary dermal blood vessels, decreased thickness of the dermis, and adipocytes high in the dermis), are reproducible and can strongly point to the correct diagnosis of Goltz syndrome.
- Treatment: Symptomatic



THANK YOU