Developmental disorders of orofacial structures

- ***Congenital :*** Existing since before birth or at birth; dating from birth. Congenital malformations are anatomical or structural abnormalities that are present at birth, though they may not be diagnosed until later.

- ***Hereditary:*** Transmitted from ancestors or parents to a child. Same as genetic.

- ***Genetic :*** Transmitted from one generation to another through genes, same as hereditary.

- ***Autosomal :*** A trait transmitted by a gene carried on an autosome (any member of the 22 paired chromosomes other than the X and Y sex chromosomes).

- ***Sex-liked:*** **A trait transmitted by a gene carried on one of the sex chromosomes.**

- ***Dominant :*** **A dominant gene is one which will produce its effect when present in either heterozygous or homozygous condition (when only one or both alleles are affected).**

- ***Recessive :*** **A recessive gene is one which will produce its effect only when present in a homozygous condition (only when both alleles are affected).**

- ***Developmental :*** Characterized by , or belonging to the process of development. Used here to designate a condition or disease caused by some non-hereditary error in the process of development.

- ***Acquired :*** A term used to describe a condition, habit or other characteristic which is not present at birth , but which develop in the individual by reaction to some environmental factor (to acquire is to obtain).

- ***Translocation :*** Transfer of chromosomal material between chromosomes. This involves breakage of both chromosomes with repair in an abnormal arrangement.

- ***Inversion :*** This arises through two chromosomal breaks with inversion through 180 of the segment between the breaks.

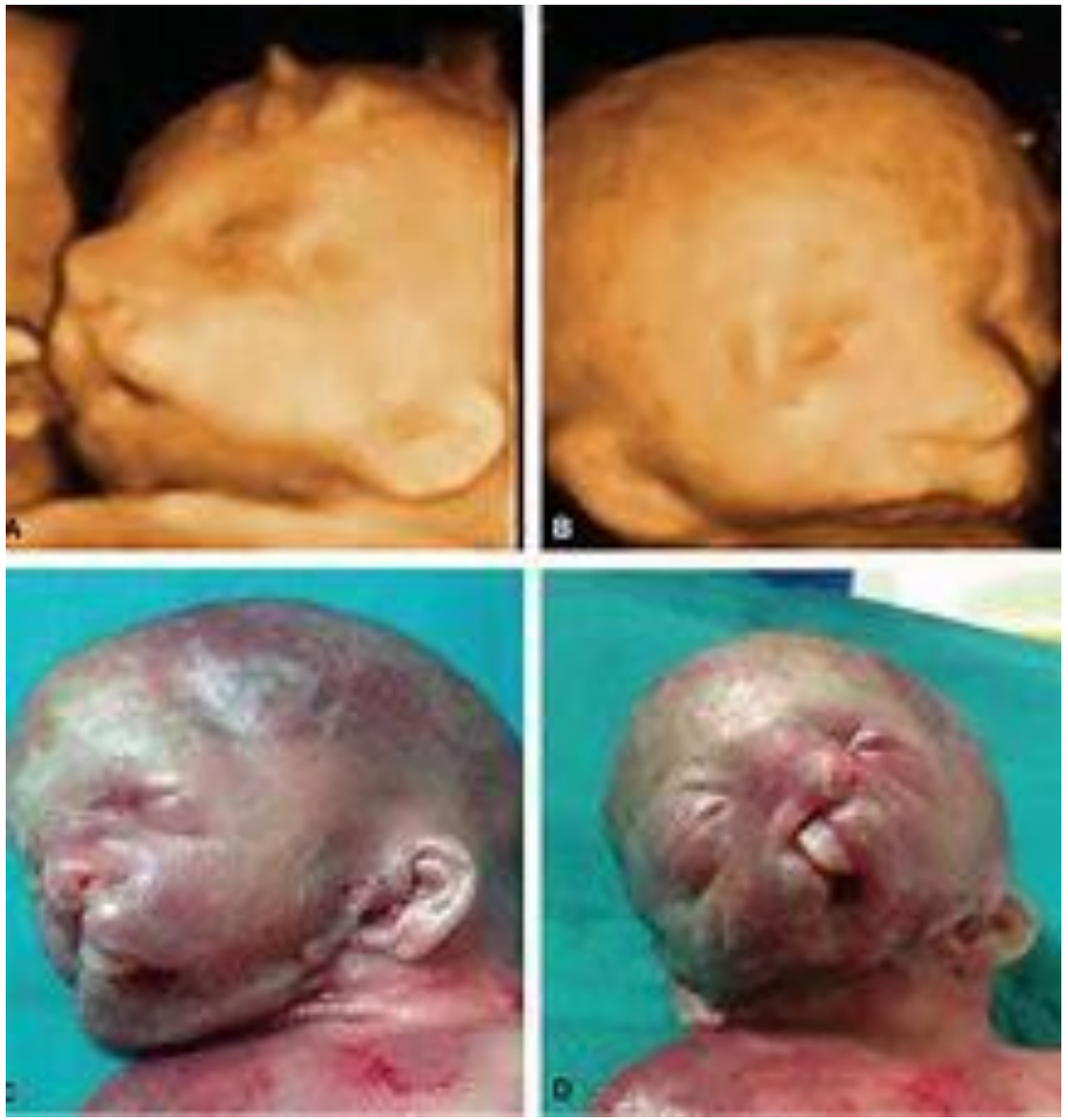
- ***Deletion :*** **This arises from loss of a portion of a chromosome between two break points or as a result of a parental translocation.**

- ***Teratogen :*** **Any agent that can induce or increase the incidence of a congenital malformation. An agent capable of causing abnormal development prior to birth .**

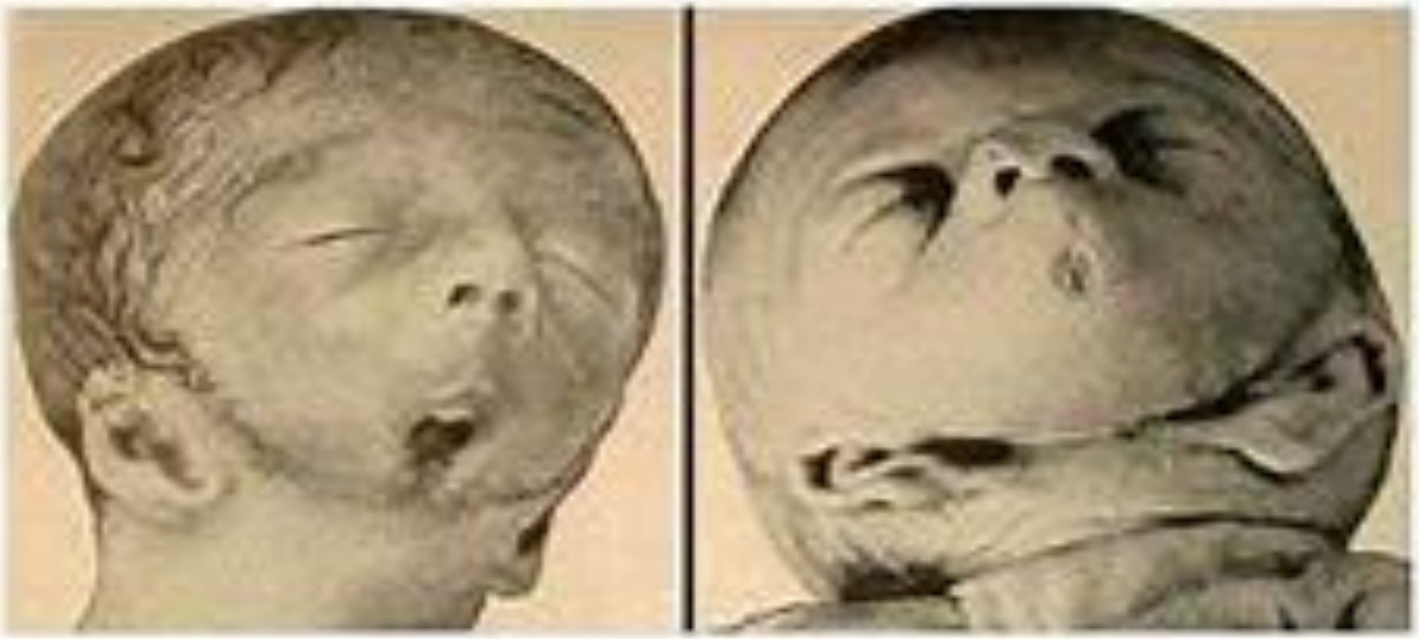
Developmental Disturbances of the jaws

Agnathia

- ** Very rare congenital defect.
- ** it means complete absence of mandible or maxilla.
- **it is more common in mandible (mandibular agnathia.).
- **In maxilla:- the maxillary process or the premaxilla may be abscent.

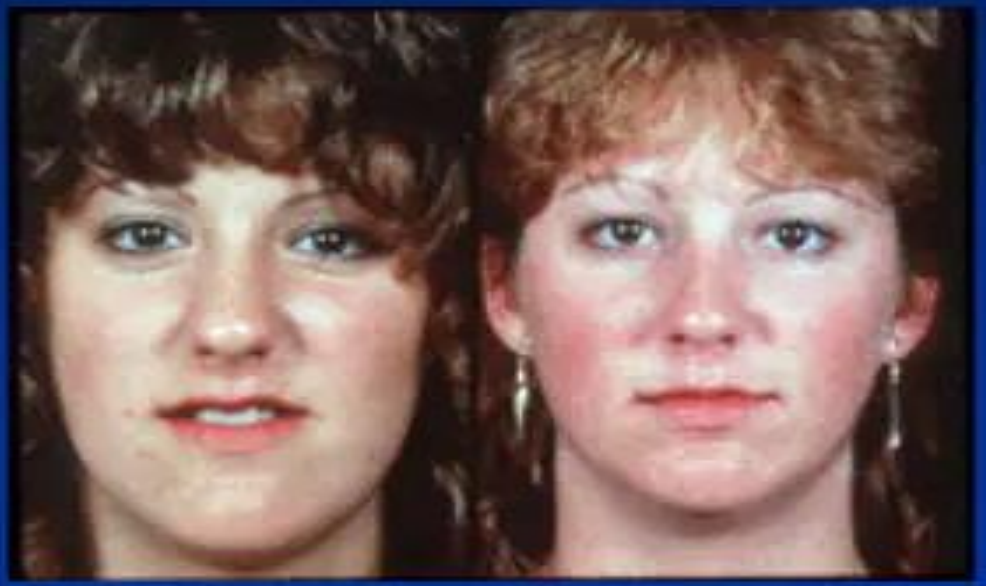


Agnathia: Agnathia (also termed hypognathous] is absence of a portion or the entirety one or both jaws.It is a very rare condition. deficiency of neural crest tissue in lower part of face.



Macrognathia

- The condition of abnormally large jaw.
- It may be associated with:-
  **Some diseases of bone as Paget`s disease.
  **Hormonal disturbances as acromegally.



Orofacial Clefts

- Cleft Lip (CL)
- Cleft Palate (CP)
- Cleft Lip with Cleft Palate (CL + CP)
- Lateral Facial Cleft
- Oblique Facial Cleft
- Median Cleft of upper lip
- Median Maxillary anterior alveolar cleft.

Etiology

- Genetic abnormalities
- Inherited
- Spontaneous mutation
- Environmental factors
- Nutritional Deficiency
- Cigarette smoking
- Drugs, radiation.
- Amniotic bands

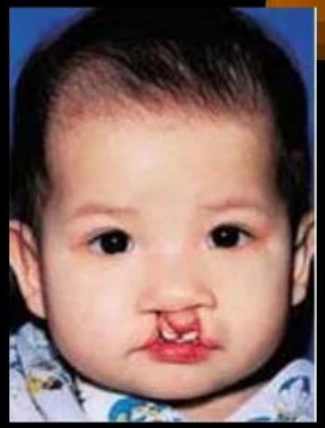
Cleft Lip and Palate

- Most common among the facial clefts.
- Cleft is a division or separation of parts of the lip or palate that is formed during the early months of development of the fetus.
- Clefts may be unilateral or bilateral.
- They can vary in severity.
- 3-8% of clefts are associated with syndromes

Unilateral incomplete cleft lip

Unilateral Complete Cleft lip

Bilateral Complete Cleft Lip



Frequency of occurrence

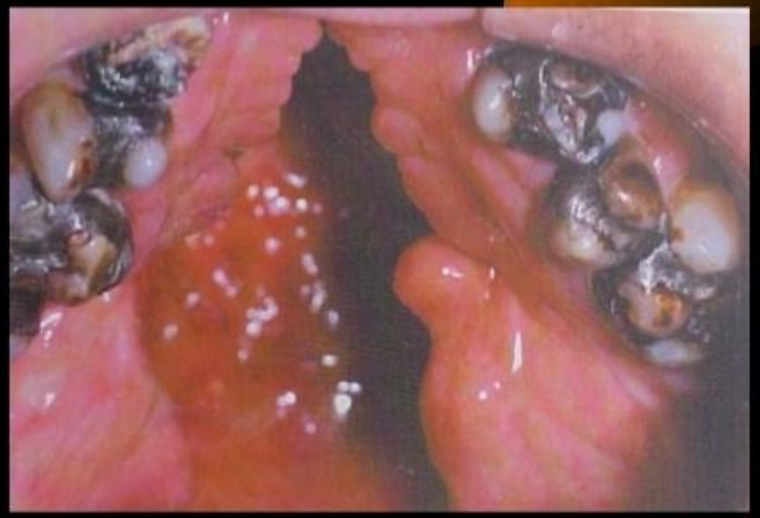
- Isolated Cleft Lip : 25%
- Isolated Cleft Palate : 30%
- Cleft Lip + Cleft Palate : 45%
- CL ± CP : more common in males.
- For isolated CL , M : F = 1.5 : 1
- For CL + CP, M : F = 2 : 1
- Isolated CP is more common in females.

Location

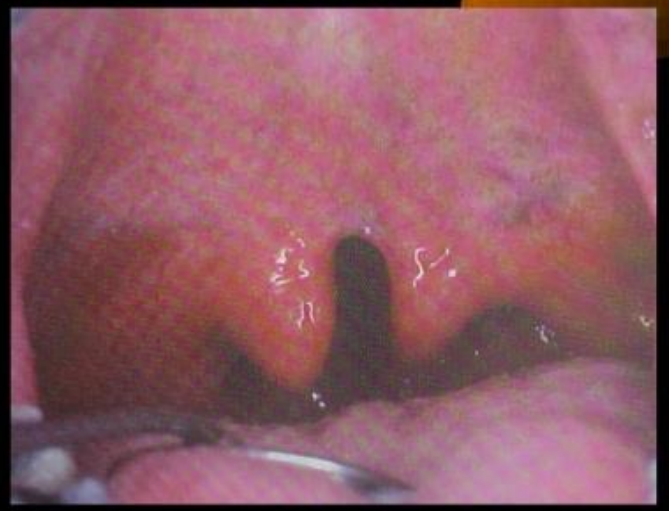
- Cleft Lip is more commonly unilateral (80%)
- 70% of cleft lips on left side.
- Complete CL extends upward into nostril.
- CP may involve hard and soft palate or soft palate alone.
- Cleft or Bifid Uvula is more common.

- **Bifid Uvula**:
- Whites: 1 in 80
- Asian and Native American: 1 in 10
- African Americans: 1 in 250
- **Submucous palatal cleft** : surface mucosa is intact but underlying musculature of soft palate is defective.

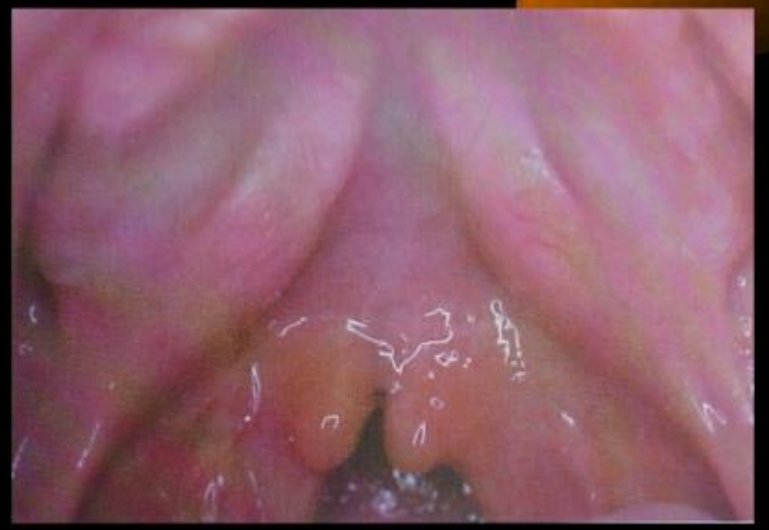
Complete cleft palate



Bifid uvula



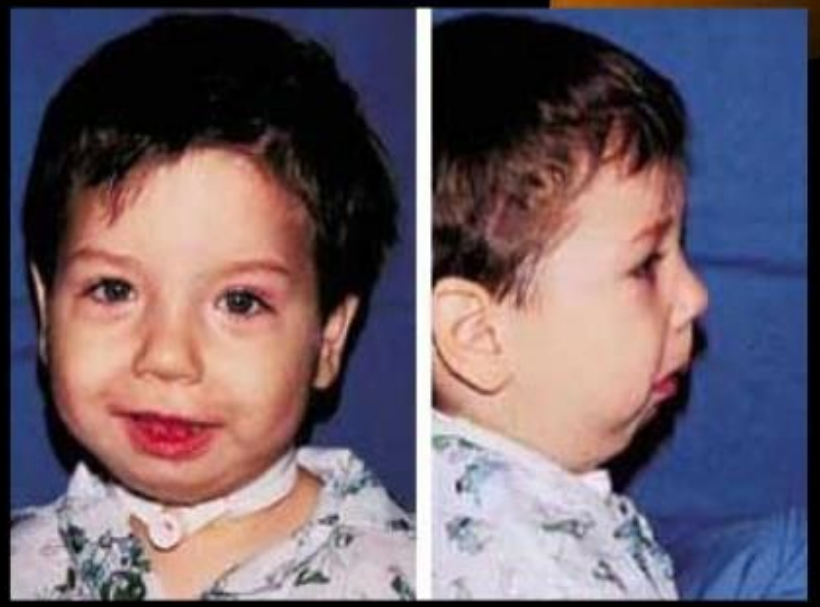
Submucous palatal cleft



Pierre Robin Syndrome

- Triad of :
- Mandibular micrognathia
- Glossoptosis
- Cleft Palate.

Bird facies



Problems associated with clefts

- Esthetic disfigurement
- Difficulty in breathing ,feeding and speech
- Malocclusion.
- Psychosocial problems.
- Recurrent upper respiratory tract infections.

Treatment

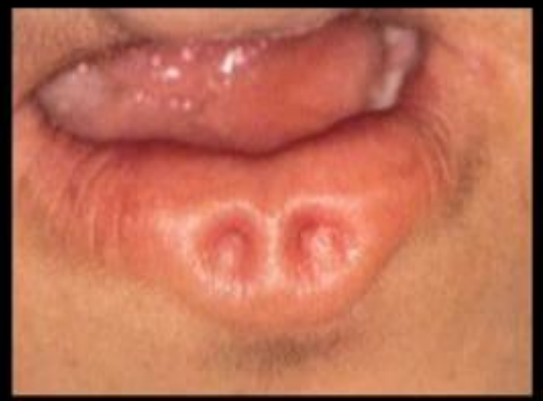
- Multidisciplinary approach
- Plastic surgery, Dentistry,Otolaryngology,Audiology,speech pathology,Genetics and Pediatrics

- SURGICAL REPAIR.

Developmental disorders of Lips

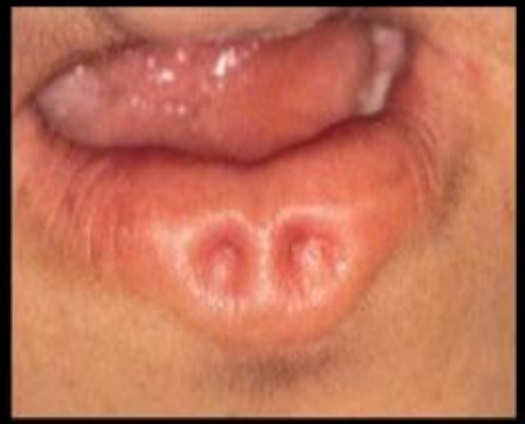
- **Lip Pits:**
- Para median Lip Pits
- Commissural Lip Pits
- **Double Lip**

Paramedian Lip Pits



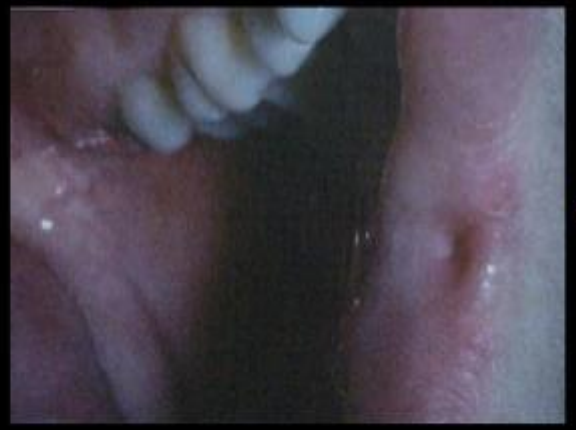
- Rare
- Autosomal dominant inheritance
- Persistence of lateral sulci on embryonic mandibular arch
- Bilateral symmetric fistulas on either side of the midline

Paramedian Lip Pits



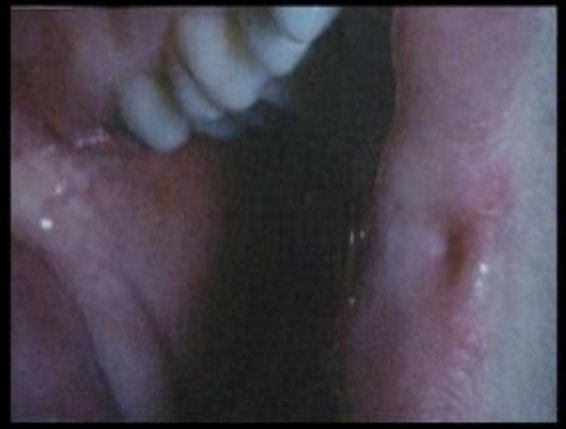
- Appearance varies from subtle depressions to prominent humps.
- Pits can extend to a depth of 1.5 cm and may express salivary secretions
- Seen in Van der Woude syndrome with CL±CP
- Surgical excision for cosmetic reasons

Commissural Lip Pit



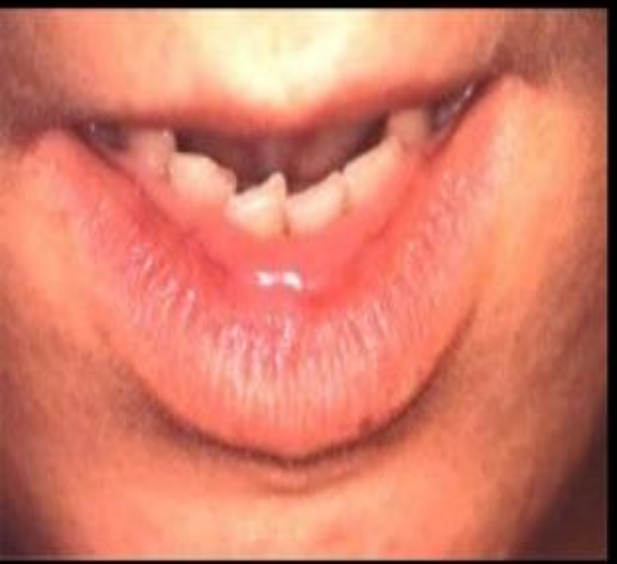
- Small mucosal invaginations at corners of the mouth on vermilion border.
- Failure in the normal fusion of maxillary and mandibular processes during development

Commissural Lip Pit



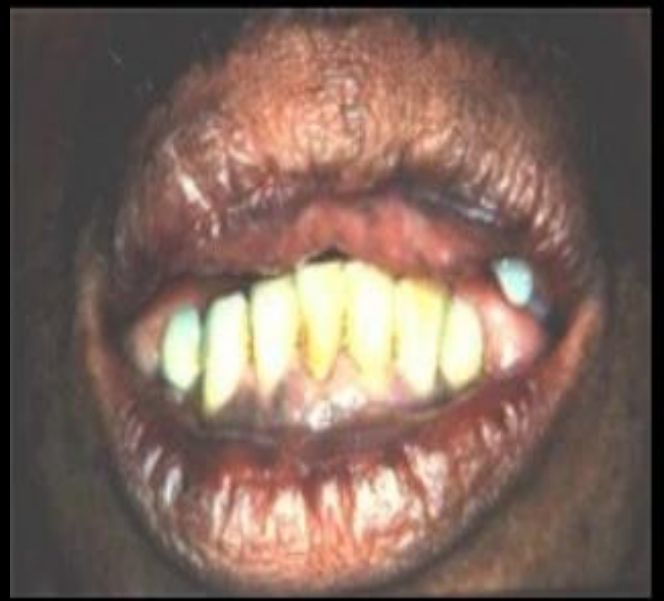
- Seen in 12-20 % of adult population.
- Males > Females
- Unilateral / Bilateral
- May be associated with preauricular pits
- No treatment required

Double Lip



- Rare
- Redundant fold of tissue on the mucosal side of the lip.
- May be Congenital or acquired(from trauma or oral habits such as lip sucking)

Double lip

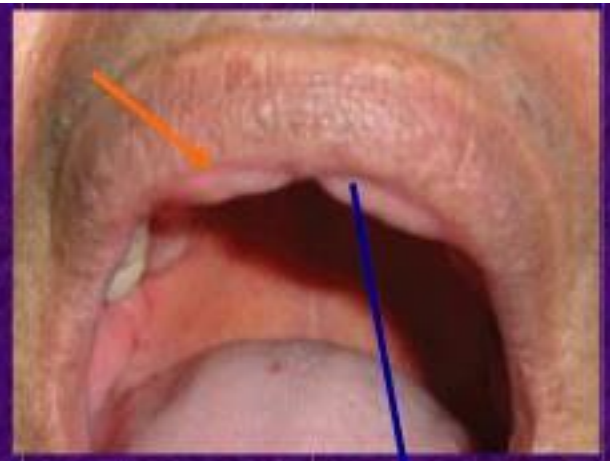


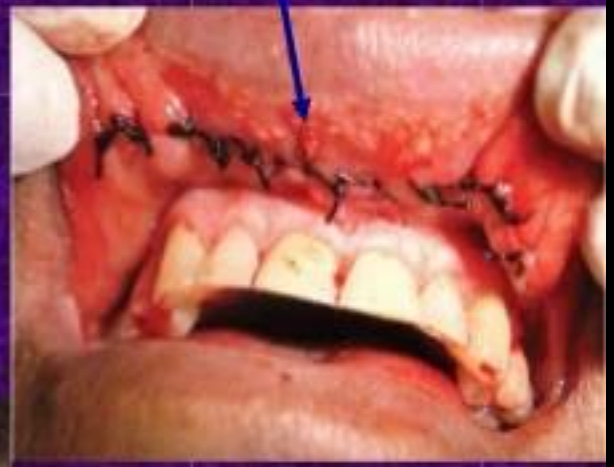
- More common in upper lip
- Sometimes both lips affected
- Seen on smiling.
- Feature of Ascher's syndrome

Double lip: usually **congenital**

- Horizontal folds of mucosal tissue

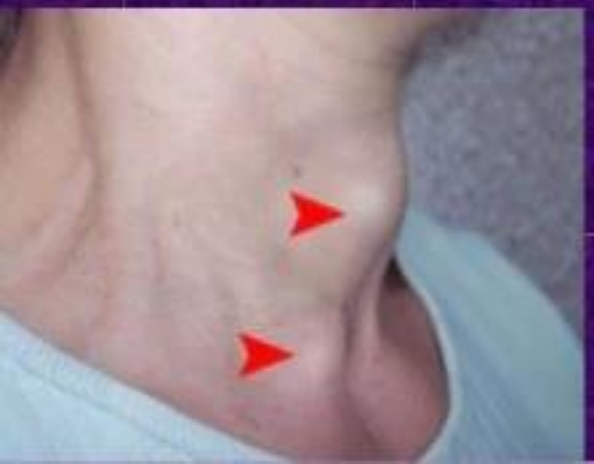
Inner aspect of U > L lip

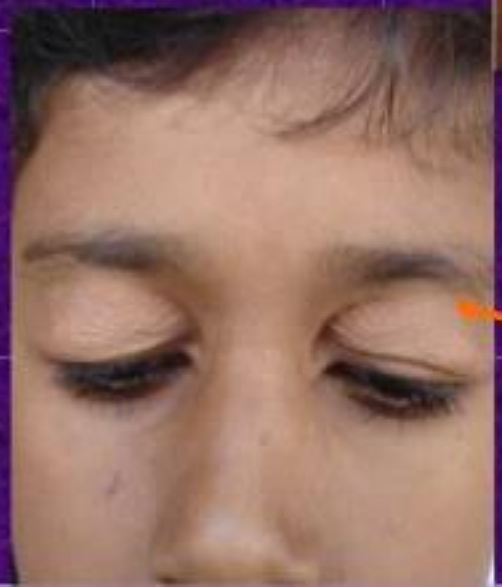




- Ascher syndrome:

+ Goitre and edema and dropping of upper U eyelids





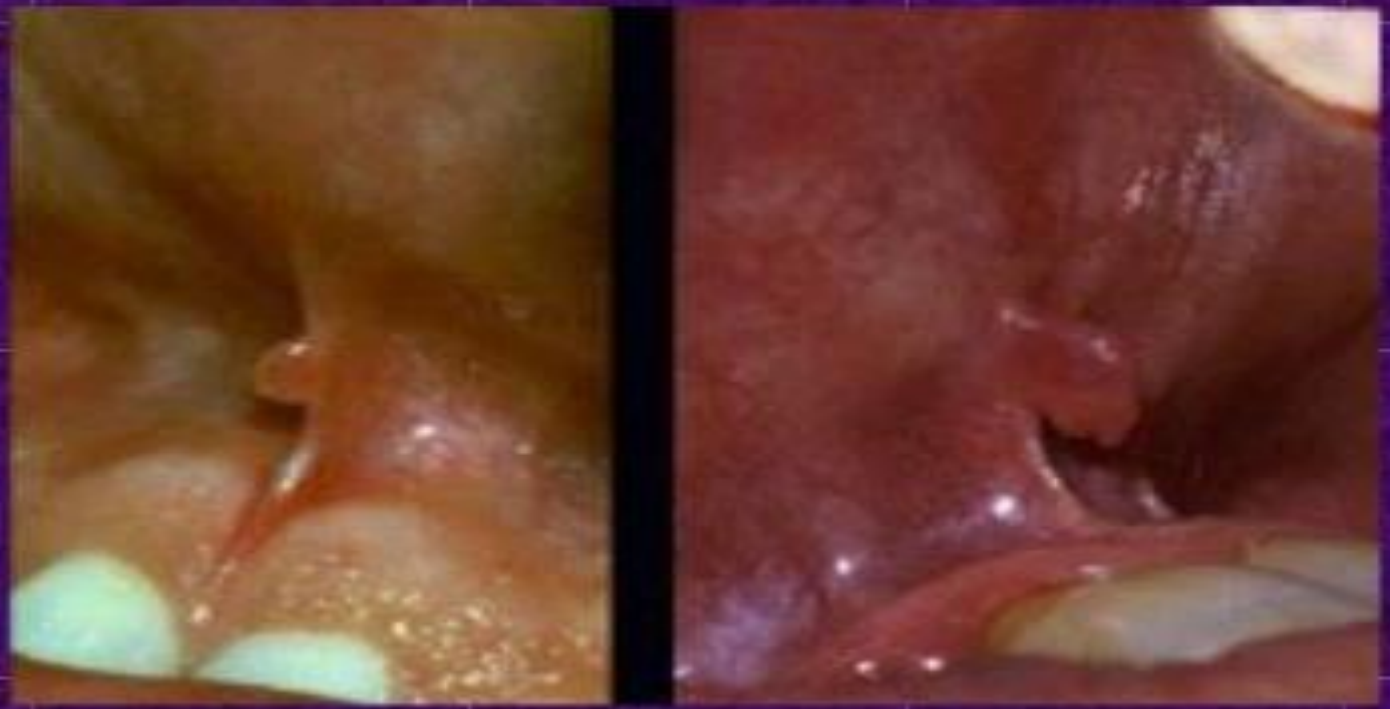
Blepharochalasis

Ascher's syndrome

- Triad of :
- Double lip
- Blepharochalasis
- Nontoxic enlargement of thyroid gland

Frenal Tag:

- Autosomal D
- U labial frenum
- Significance

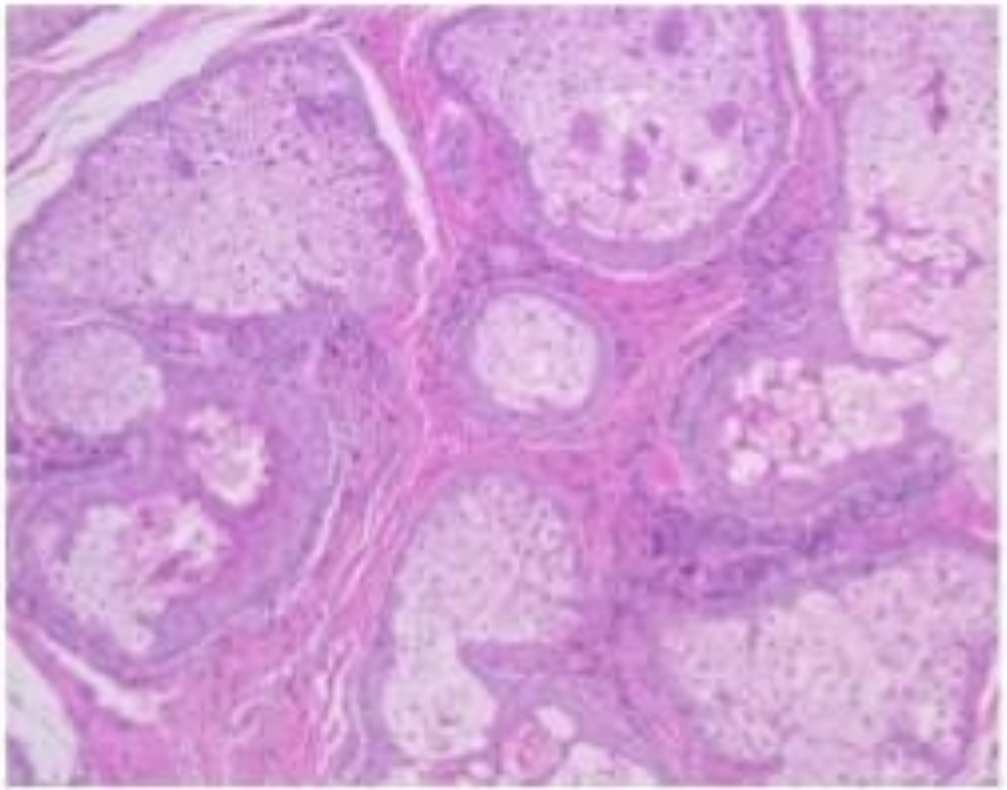


Fordyce Granules

- ✓ Multiple yellow or white papular lesions, common on buccal mucosa & lateral part of the vermilion of the upper lip.
- ✓ Mostly present in adults as a result of hormonal factors, puberty is a major factor in it
- ✓ Asymptomatic

Histopathological Features

- These are similar to the normal sebaceous glands except the absence of hair follicles.
- Acinar cells are present beneath the surface
- The sebaceous glands in these lobules are polygonal in shape, containing centrally located nuclei and foamy cytoplasm.



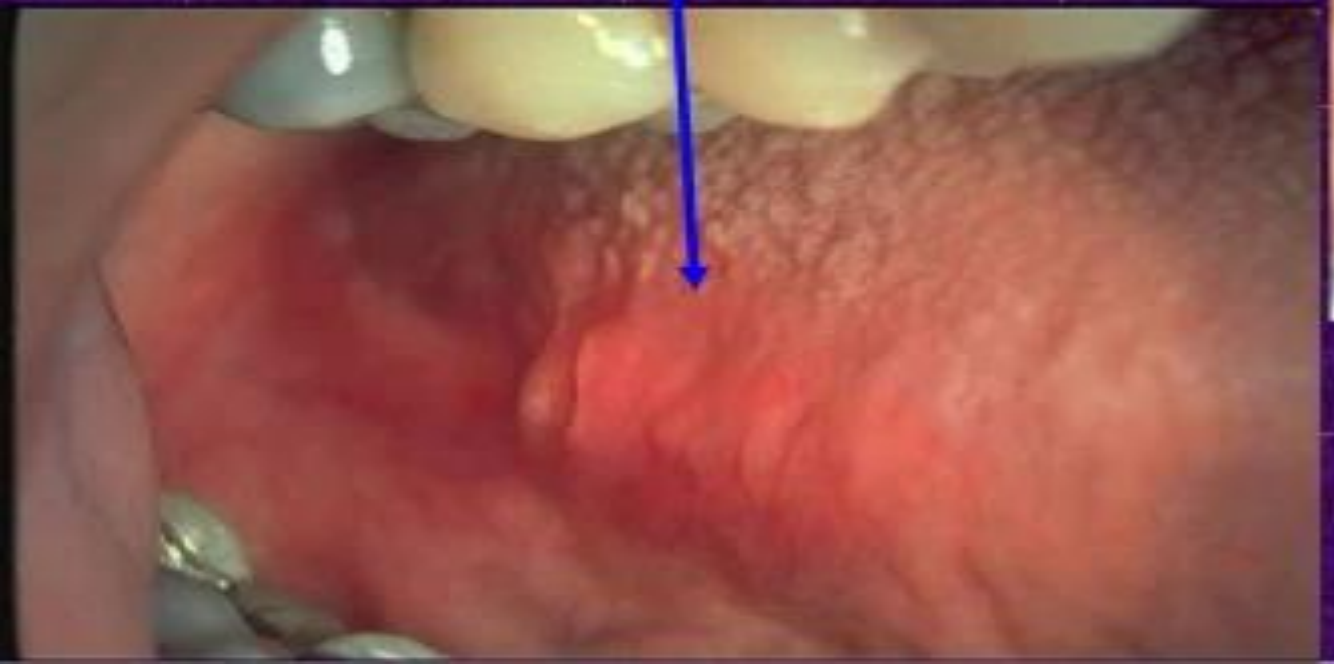
Treatment

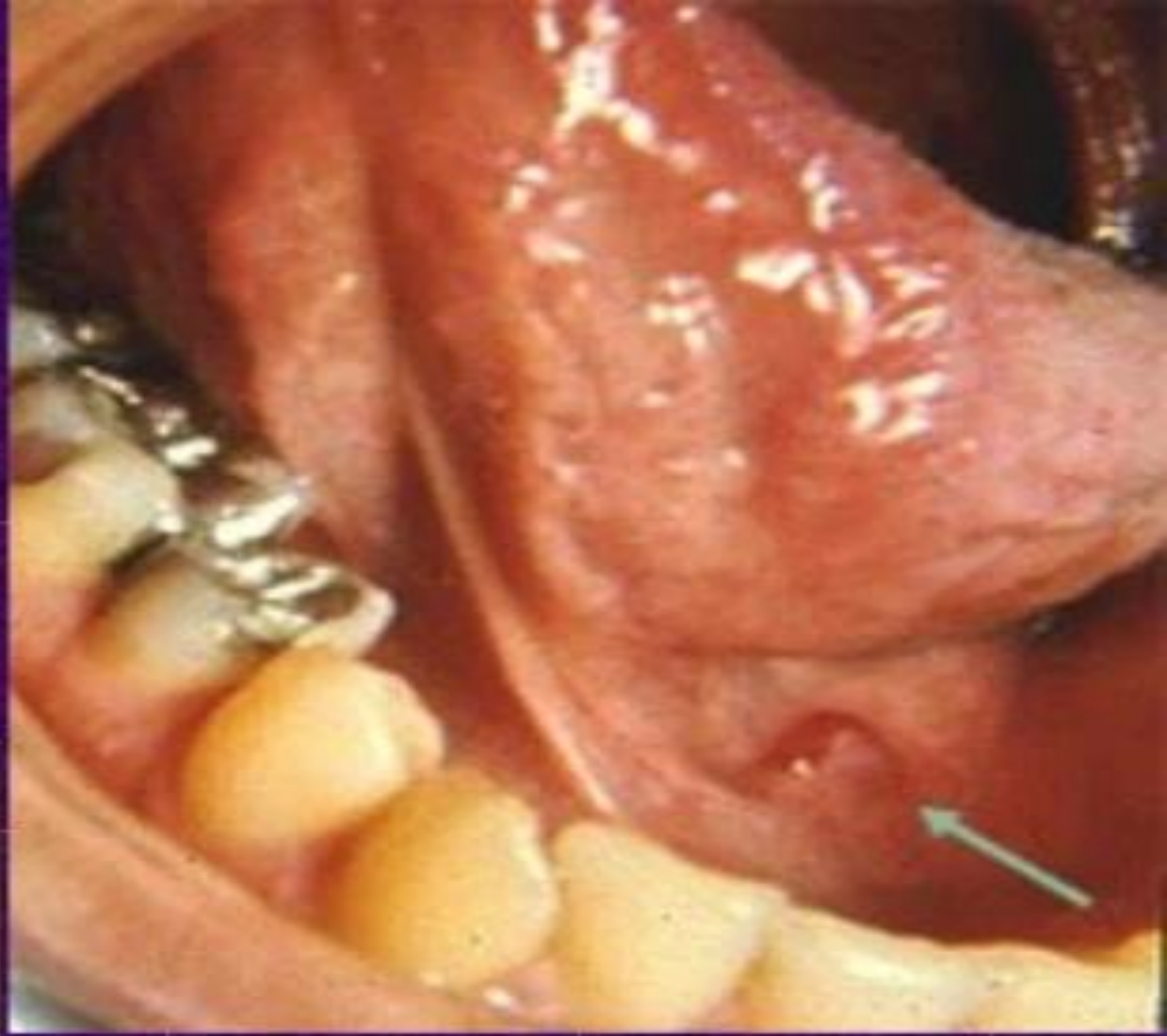
- As these are asymptomatic and a variation of normal so they don't need any treatment
- Sometimes they become hyperplastic and filled with keratin

Oral tonsils:

- Slightly elevated reddish plaques/FOM

- **Foliate Papillitis:**

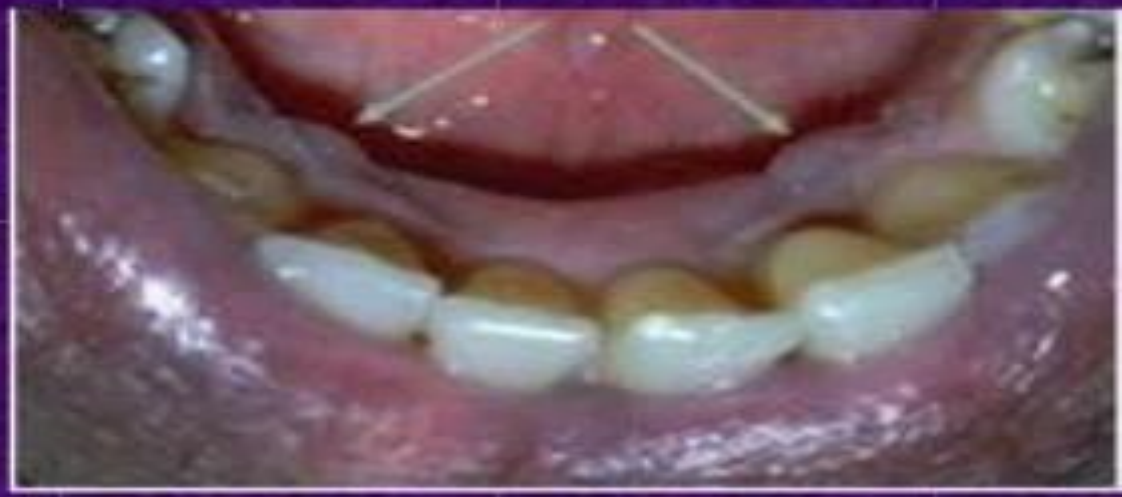


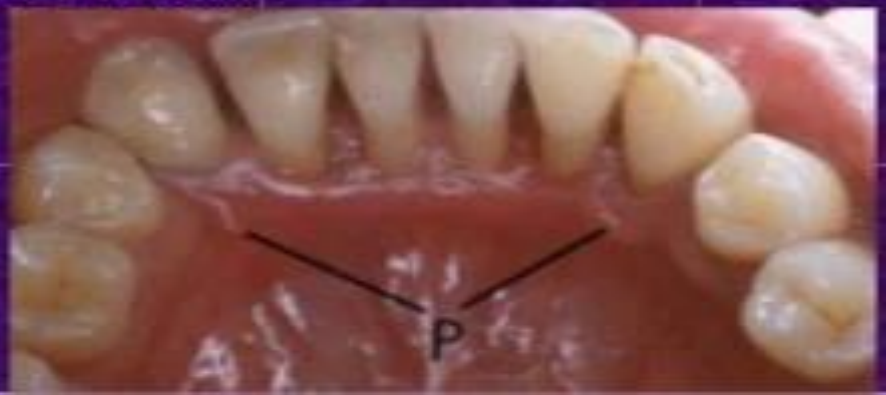


Cancer

Retrocuspid Papilla_

- Slightly raised area, about 2-4 mm, often bilaterally
- Commonly located lingual to the cuspids
- Attached gingiva
- ≈ incisive papilla
- Histologically:
  - A focus of fibrovascular tissue
  - With an orthokeratinized /parakeratinized surface
  - Covers the osseous foramen of a nutrient blood vessel





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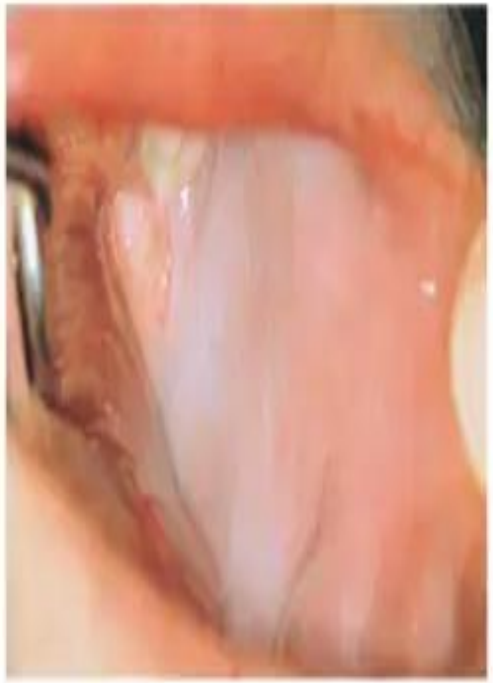
Leukoedema

- A common oral mucosal condition of the unknown cause.
- Blacks > whites
- A variation of normal anatomical structure rather than a disease



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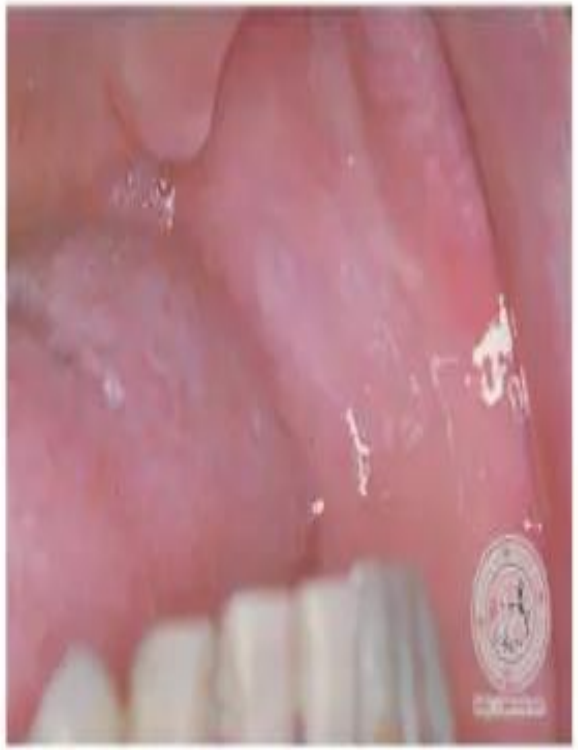
Clinical Features

- It is characterized by a diffuse, grey white, milky, opalescent appearance of the mucosa
- The surface usually appears folded showing wrinkles
- Lesions present bilaterally and do not rub off
- It can be easily diagnosed because wrinkles disappear when the cheek is everted



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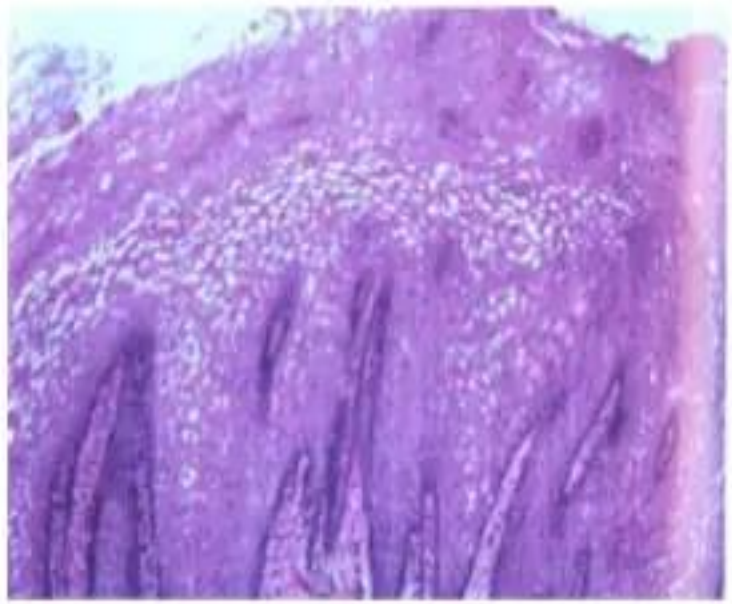
Histopathological Features

- Increase in epithelial thickness with intercellular edema of spinous layer
- Cells appear large & have pyknotic nuclei
- Surface is parakeratinized and rete ridges are broad and elongated



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Treatment

- It's a benign condition which requires no treatment


Thank You