Developmental Disorders of Oral Cavity

Prof. Shaleen Chandra

CONTENTS

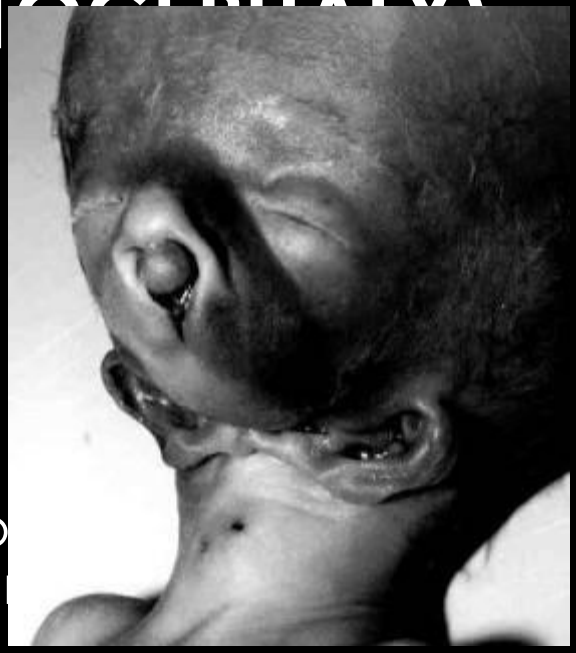
- Developmental disturbances
  - Jaws
  - Lips and palate
  - Gingiva
  - Oral mucosa
  - Tongue
  - Salivary gland
  - Tooth size
  - Tooth shape
  - Tooth structure

DEVELOPMENTAL DISTURBANCES OF JAWS

1. Agnathia
2. Micrognathia
3. Macrognathia
4. Facial Hemihypertrophy
5. Facial Hemiatrophy

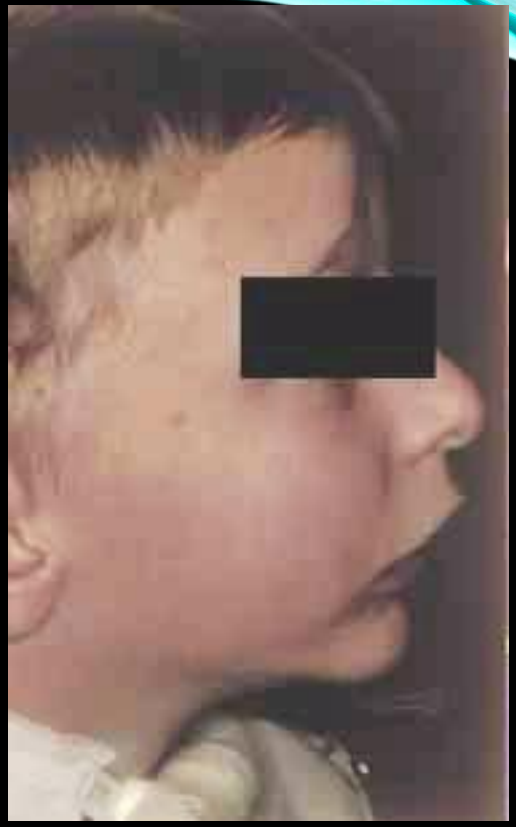
AGNATHIA (OTOCEPHALY)

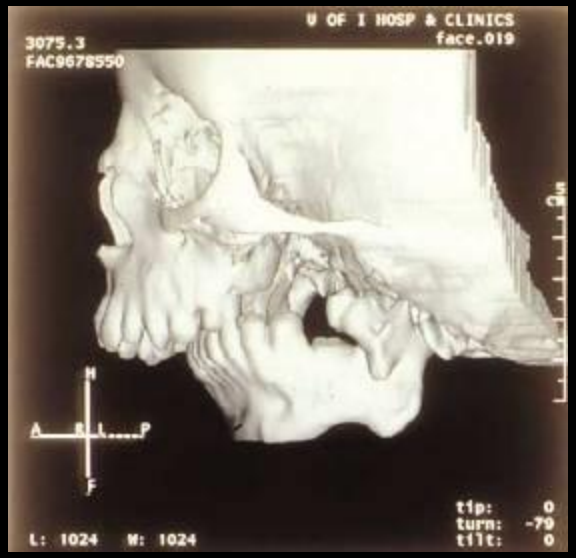
- Hypoplasia / absent mandible
- Autosomal recessive
- Unilateral missing jaw
- Ramus → ear deformities
- Etiology
  - Failure of migration of Neural crest cells into
    maxilary prominance in 4-5th week gestatio



MICROGNATHIA

- Small jaw
- DD → Abnormal positioning
- Classification
  - Congenital
    - Congenital heart disease
    - Pierre Robin syndrome
    - Maxillary micrognathia → mouth breathing
  - Acquired
    - TMJ → trauma, infection, ankylosis





- Congenital conditions
  - Catel-Manzke syndrome
  - Cerebrocostomandibular syndrome
  - Cornelia de Lange syndrome
  - Femoral hypoplasia - unusual facies syndrome
  - Fetal aminopterin-like syndrome
  - Miller-Dieker syndrome
  - Nager acrofacial dysostosis
  - Pierre Robin syndrome
  - Schwartz-Jampel-Aberfeld syndrome
  - van Bogaert-Hozay syndrome
- Intrauterine acquired conditions
  - Syphilis, congenital
- Chromosomal abnormalities
  - 49,XXXXX syndrome
  - Chromosome 18 trisomy syndrome
  - Chromosome 8 recombinant syndrome
  - Chromosome 8 trisomy syndrome
  - Cri du chat syndrome 5p-
  - Turner's syndrome
  - Wolf-Hirschhorn syndrome
- Mendelian inherited conditions
  - CODAS (cerebral, ocular, dental, auricular, skeletal) syndrome
  - Diamond-Blackfan anemia
  - Noonan's syndrome
  - Opitz-Frias syndrome

- Autosomal dominant conditions
  - Camptomelic dysplasia
  - Cardiofaciocutaneous syndrome
  - CHARGE syndrome
  - DiGeorge's syndrome
  - Loeys-Dietz syndrome
  - Marfan syndrome
  - Micrognathia with peromelia
  - Pallister-Hall syndrome
  - Treacher Collins-Franceschetti syndrome
  - Trichorhinophalangeal syndrome type 1
  - Trichorhinophalangeal syndrome type 3
  - Wagner vitreoretinal degeneration syndrome
  - Weissenbacher-Zweymuller syndrome
- Autosomal recessive conditions
  - Bowen-Conradi syndrome
  - Carey-Fineman-Ziter syndrome
  - Cerebrohepatorenal syndrome
  - Cohen syndrome
  - Craniomandibular dermatodysostosis
  - De la Chapelle dysplasia
  - Dubowitz syndrome
  - Fetal akinesia-hypokinesia sequence
  - Hurst's microtia-absent patellae-micrognath syndrome
  - Kyphomelic dysplasia
  - Lathosterolosis
  - Lethal congenital contracture syndrome
  - Lethal restrictive dermopathy
  - Marden-Walker syndrome
  - Orofaciodigital syndrome type 4
  - Postaxial acrofacial dysostosis syndrome
  - Rothmund-Thomson syndrome
  - Smith-Lemli-Opitz syndrome
  - ter Haar syndrome
  - Toriello-Carey syndrome
  - Yunis-Varon syndrome
- X-linked inherited conditions
  - Atkin-Flaitz-Patil syndrome
  - Coffin-Lowry syndrome
  - Lujan-Fryns syndrome
  - Otopalatodigital syndrome type 2
  - Scott craniodigital syndrome
- Autoimmune conditions
  - Juvenile chronic arthritis

MACROGNATHIA

- Large jaws
  - Gigantism
  - Pagets disease
  - Acromegaly
  - Leontiasis ossea
- DD ➔ prognathism
  - Increased ramus height
  - Increased mandibular body length
  - Decreased maxillary length

HEMIFACIAL HYPERTROPHY

- Hyperplasia rather than hypertropy
- Syndromes associated
  - Beckwith Wiedmann syndrome
  - Neurofibromatosis
  - McCune albright syndrome
  - Mafucci's syndrome
- Classification *(hoyme et al 1998)*
  - Complex hemihyperplasia → half of body
  - Simple Hemihyperplasia → single limb
  - Hemifacial hyperplasia → One half of face

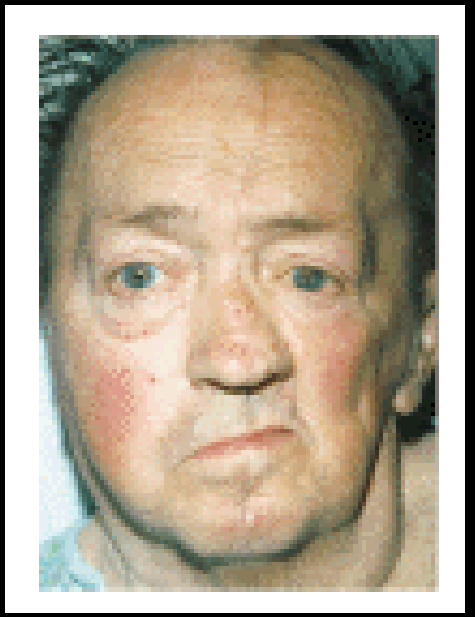
CLINICAL FEATURES

- F> M
- Macroglossia
- Premature development and eruption of teeth
  - Rowe et al
    - Crown size
    - Root size and shape
    - Rate of development

Histologically → NO MUSCULAR HYPERTROPY

FACIAL HEMIATROPY

- Parry Romberg syndrome
  - Progressive atropy of soft tissues
  - Confined to one half of face
- Etiology
  - Cerebral disturbance
    - Unregulated activity of sympathetic NS
  - Local trauma
    - Extraction of teeth
    - Infection
  - Genetic factors



CLINICAL FEATURES

- Painless cleft
- *Coup de sabre (*mid line of face)
- Bluish hue
  - *Atropic fat*
- Dental malformations
  - Incomplete root formation
  - Delayed eruption
  - Severe facial asymmetry

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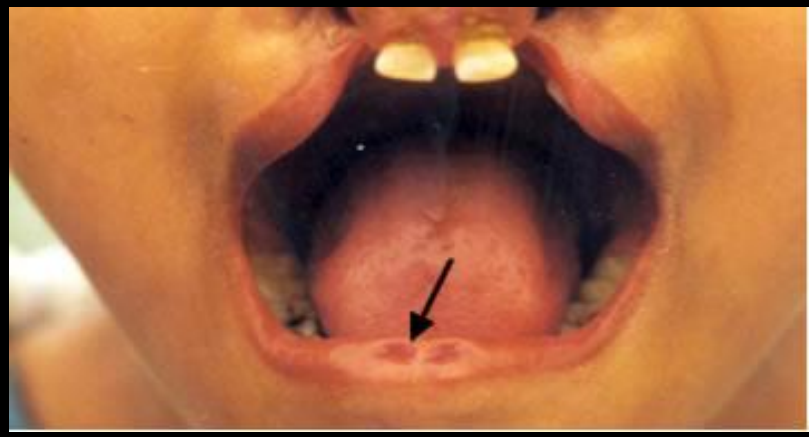
DEVELOPMENTAL DISTURBANCES OF LIPS AND PALATE

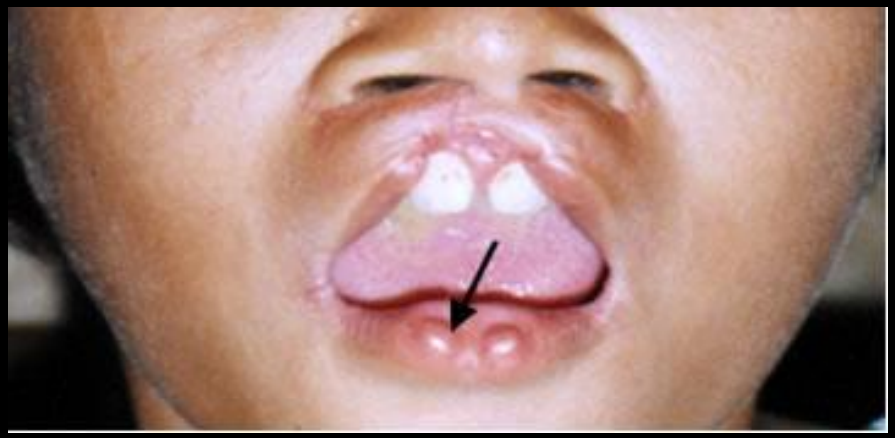
CONGENITAL LIP PITS AND COMMISURAL PITS

- Etiology
  - Notching of lip (early stage) → fixation of tissue at the base of the notch
  - Failure of complete union of embryonic lateral sulci of lip
  - Commisural pits
    - Defective development of embryonic fissure
- Clinical features
  - Unilateral / bilatera
  - LL > UL



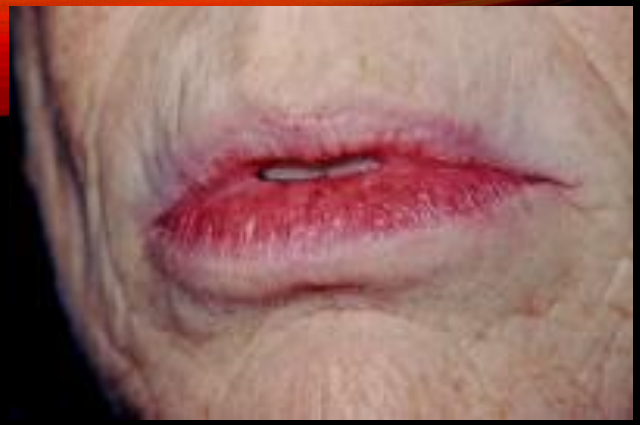
VAN DER WOUDE'S SYNDROME

- Autosomal dominant
- Deletion of chr 1q32 and alteration in chr 17p11
- Features
  - Cleft lip +- palate
  - Pits of lower lip
  - Maxillary hypodontia
  - Syngnathia
  - Ankyloglosia

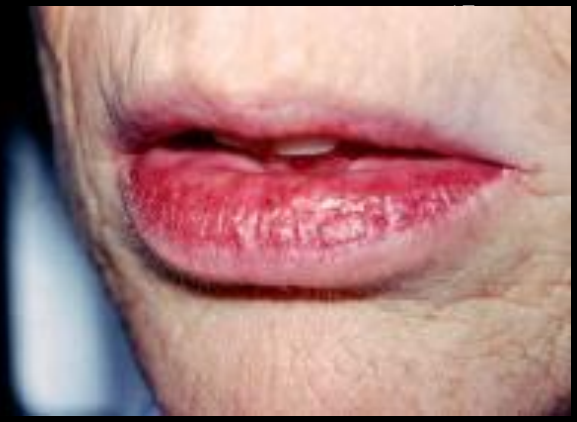


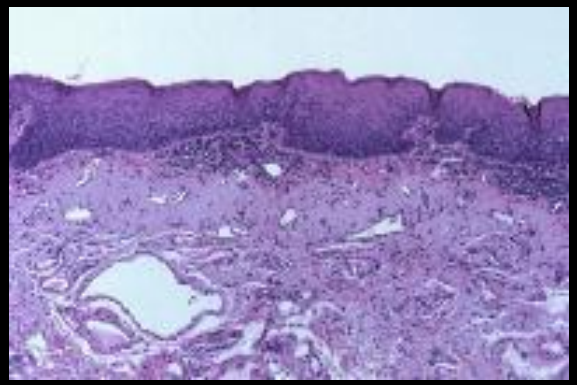
CHELITIS GLANDULARIS (ACTINIC CHELITIS)

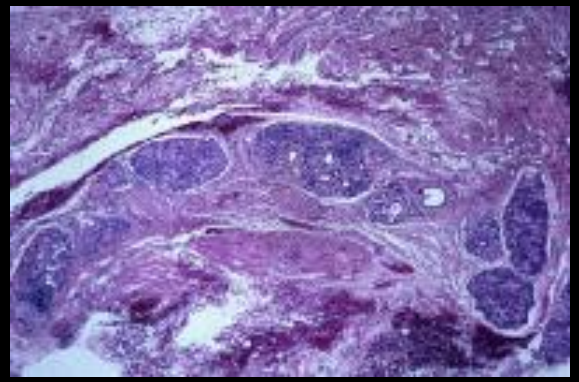
- Progressive enlargement and eversion of lower labial mucosa
- Exposure
- Erosion + ulceration + crusting

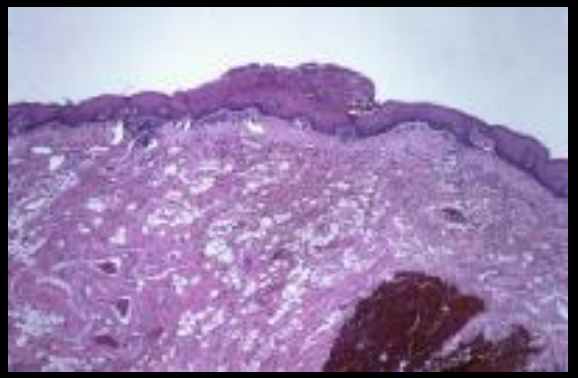


- Burning + Pain
- Desiccation
- Suppuration
- Fair skin more common









- Basophillic collagen degeneration
- Ductal ectasia, atrophy
- Hyperkeratosis and fibrosis

CLASSIFICATION

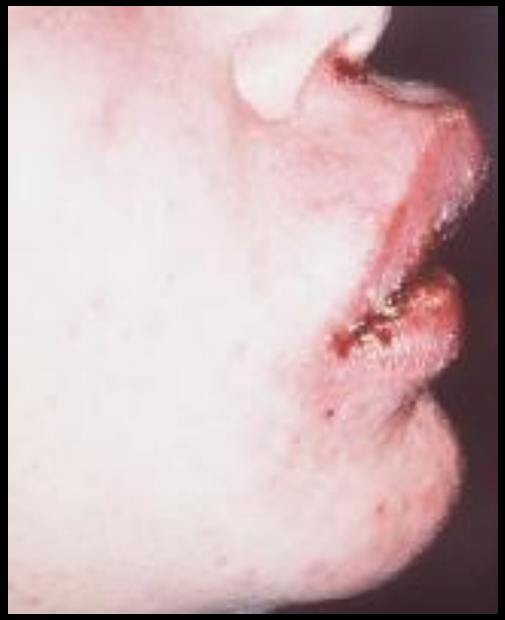
- Simple type
  - Multiple painless, papules with central depression
- Superficial (suppurative) type
  - Baelz disease
  - Painless indurated swelling of lip with shallow ulceration
- Deep suppurative type
  - Deep seated abscess + sinus

CHEILITIS GRANULOMATOSA

- Melkersson Rosenthal syndrome
  - Granulomatous inflammation
  - Cheilitis
  - Facial nerve palsy
  - Plicated tongue
- Etiology
  - Genetic → siblings affected

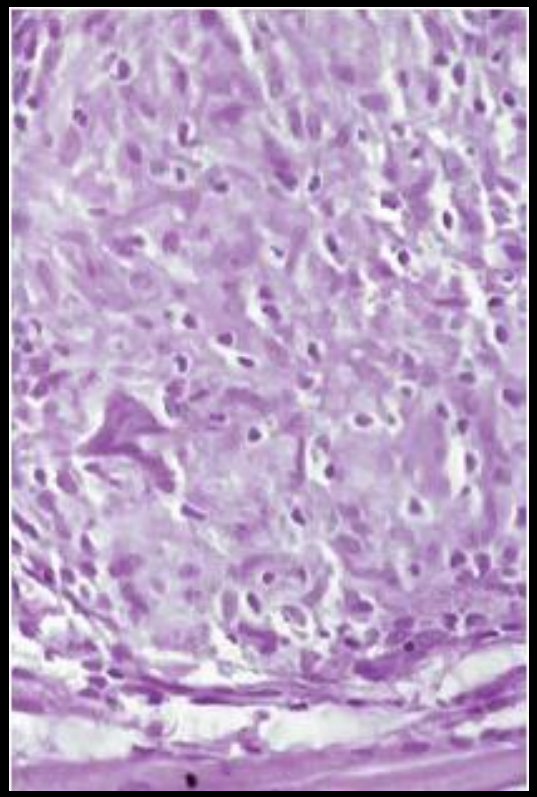
CLINICAL FEATURES

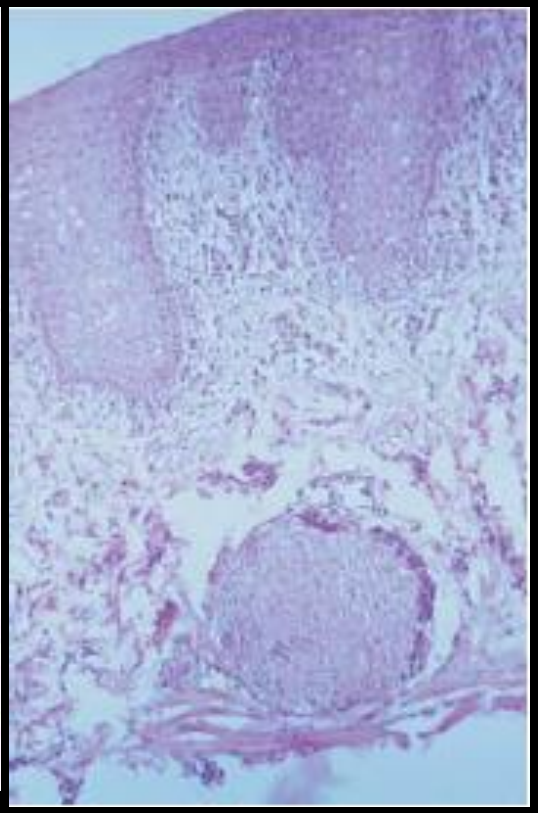
- Chelitis and ulceration
  - Episodic
  - Nontender swelling
  - Cracked fissured lips
  - Red to brown discolouration
  - Fissured tongue → 20- 40% cases
  - Facial nerve palsy → 30 %



HISTOPATHOLOGY

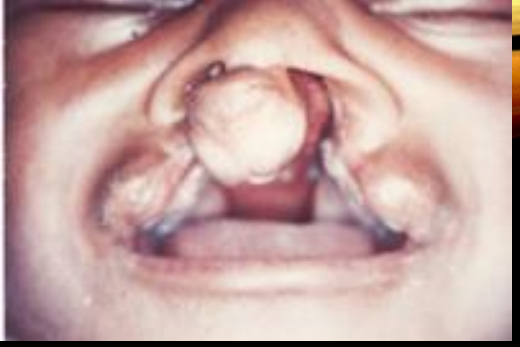
- Tuberculoid granuloma
- Chronic inflammatory cell infiltrate
- Focal noncaseating granuloma
- Epitheloid cells
- Langhans cells





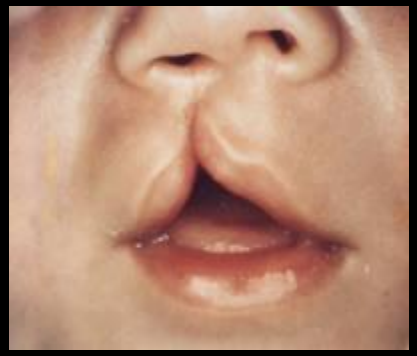
- Diagnosis
  - Serum ACE test
  - Chest radiograph
  - Gallium or positron emission tomography

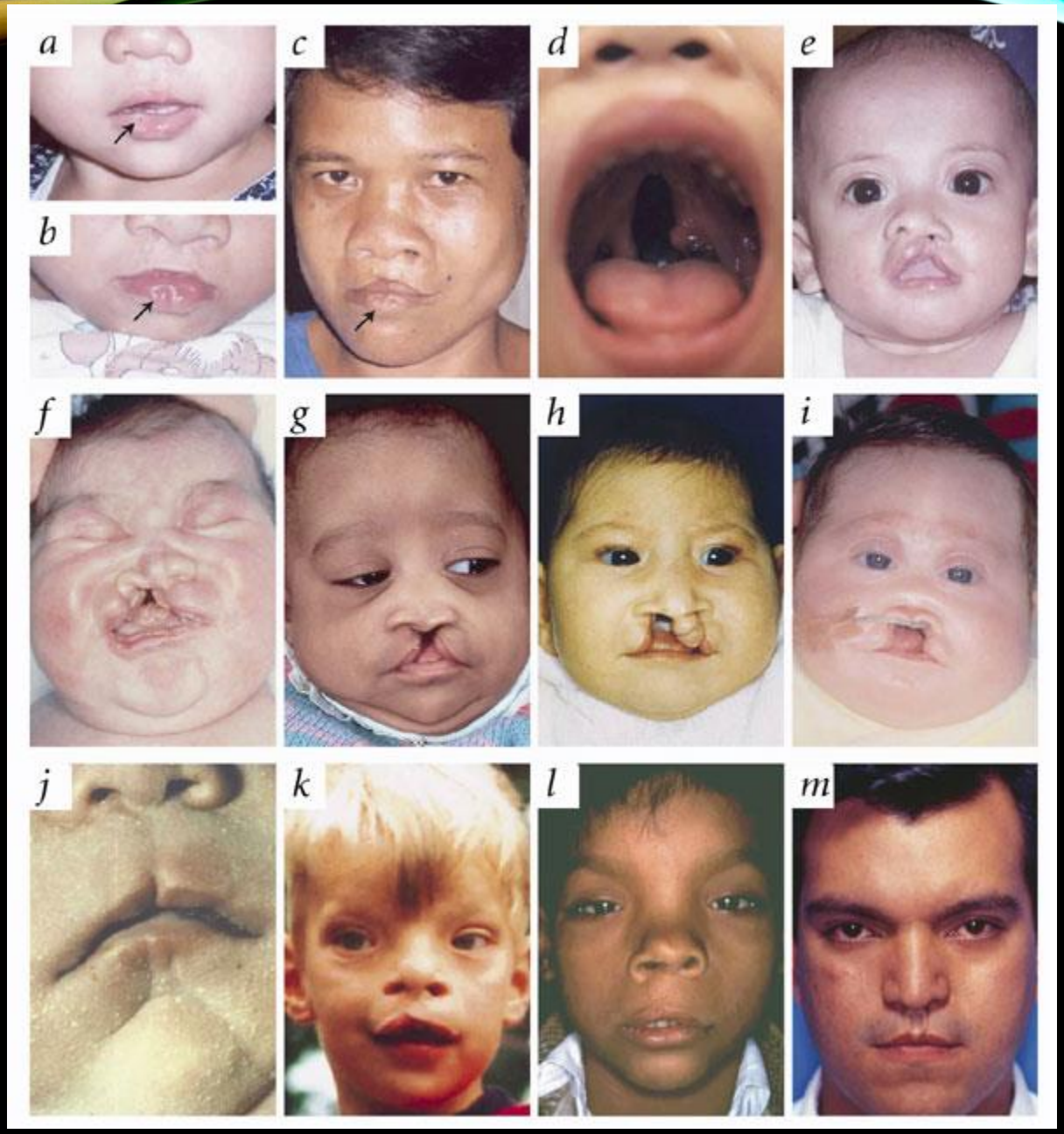
- Rule out sarcoidosis



Orofacial clefts

- A developmental defect characterized by the failure of fusion of facial processes.
- 6th and 7th week → upper lip
- 8th week → palate
  - Anterior to posterior
- Median nasal process vs maxillary process ➔ cleft lip
- Maxillary process ➔ cleft palate





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ETIOLOGY

- Heredity
  - Single mutant gene
    - Syndromic (high risk)
  - Polygenic → low risk

    The total genetic liability of an individual reaches a certain minimum level.
- Nutritional disturbances
- Physiological, emotional and traumatic stress
- Defective vascular supply
- Mechanical disturbances
- Infections
- Lack of inherent developmental force

CATEGORIES

- CL +- CP ➔ same etiology
- CP ➔ separate etiology

MEDIAN CLEFT FACE SYNDROME

- Hyper telorism
- Median cleft of premaxilla and palate
- Etiology
  - Precocious limitation of growth of primary ossification centers on either side of mid line
  - Failure to fuse

TREATMENT

- Multispeciality
  - Rule of 10
    - 10 weeks
    - 10 lbs
    - 10 mg / dl Hb
    - 10000 WBC count
  - Surgery, orthodontics, speech therapy

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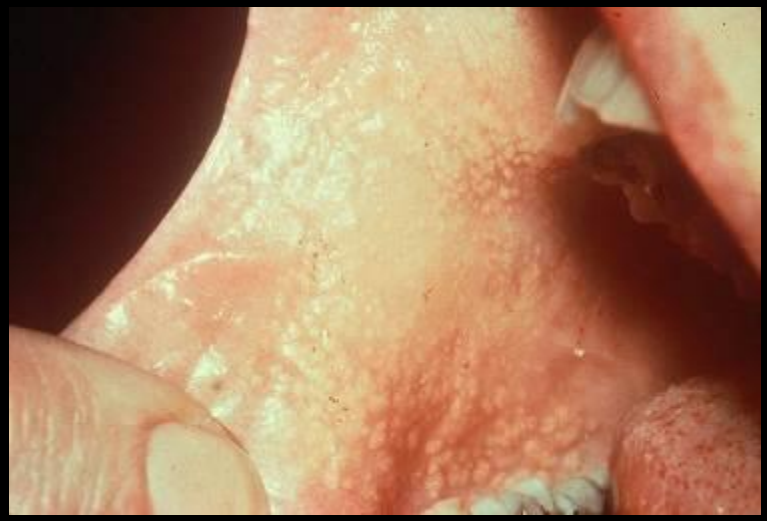
DEVELOPMENTAL DISTURBANCES OF THE ORAL MUCOSA

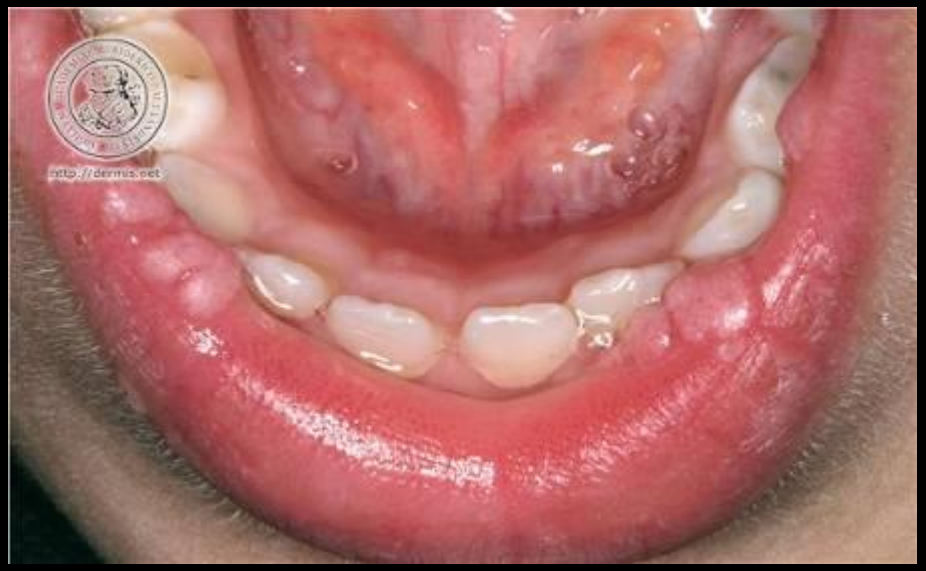
FORDYCE'S GRANULES

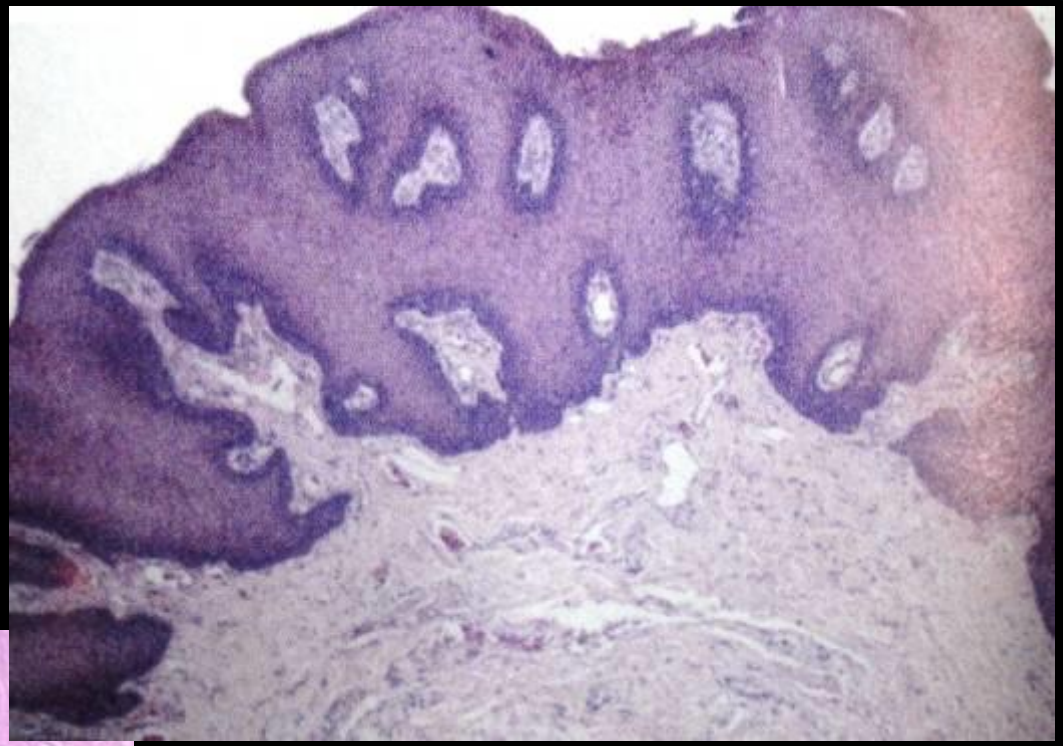
- Heterotrophic collections of sebaceous glands
- Usually symmetrical
- Appear at puberty
  - Not all cases (*mile)*
    - Sebaceous nevi

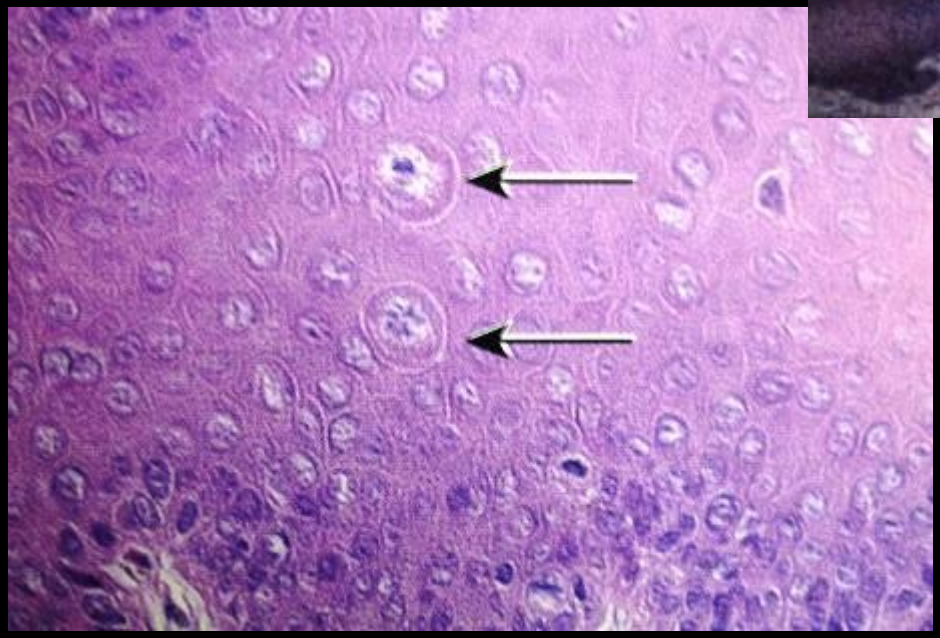


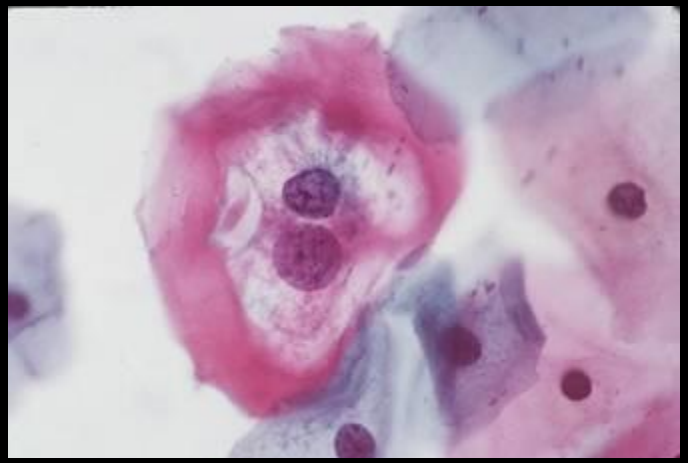
FOCAL EPITHELIAL HYPERPLASIA

- HPV 13, 32
- Epithelium 8- 10 times thicker









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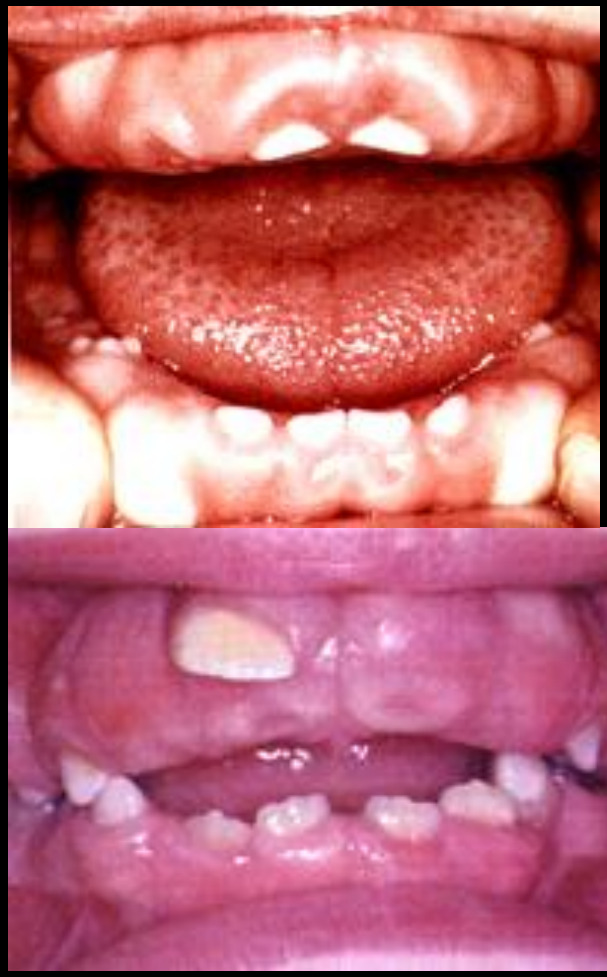
DEVELOPMENTAL DISTURBANCES OF GINGIVA

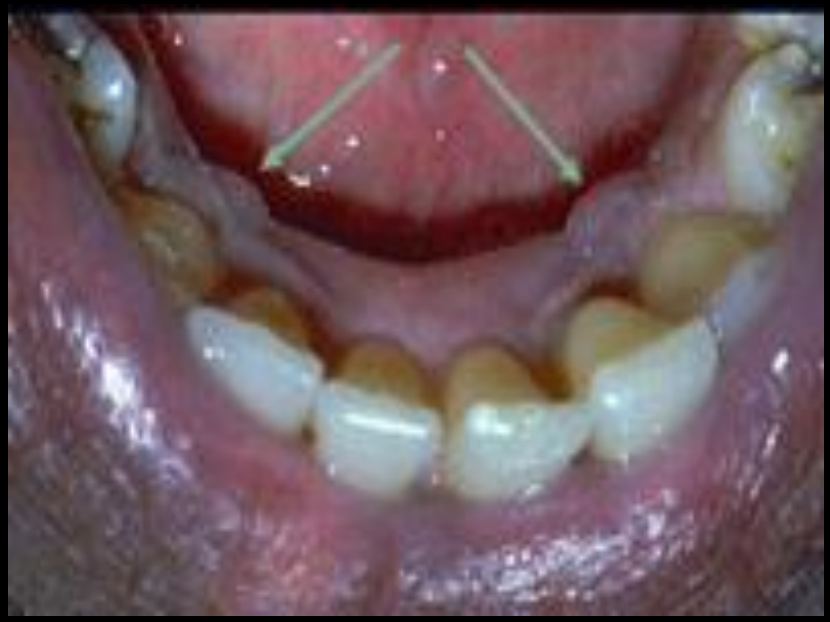
HEREDITARY GINGIVAL FIBROMATOSIS

- Benign → idiopathic
- Autosomal dominant
- Nodular form
- Clinical features
  - Dense , diffuse , growth
  - Crown may be hidden
  - No inflammation
  - Normal / pale colour



RETROCUSPID PAPILLA

- Hirshfield 1933
- Soft well circumscribed
- Between
  - Free gingival margin and
  - Mucogingival junction
- Elevated mucosal tag
  - Hyper orthokeratosis
  - Highly vascular CT
  - Large stellate fibroblasts



DEVELOPMENTAL ANOMALIES OF SALIVARY GLANDS

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ABERRANT SALIVARY GLANDS

- Location
  - Cervical region near parotid
  - Body of mandible
  - Region of brachial clefts and bronchial cleft cysts
  - Tongue
- Histology similar to the normal salivary gland

APLASIA AND HYPOPLASIA

- Along with congenital anomalies
  - Cleft palate
  - Mandibulofacial dysostosis
- Symptoms
  - Xerostomia
  - Dentinal caries
  - Melkerson Rosenthal syndrome

ACCESSORY DUCTS

- Common > 50% cases
- Superior and anterior to the normal stensons duct
- Rauch and Gorlin → 450 cases

DIVERTICULI

- Small pouches or out pocketings of the ductal system
- Recurrent acute parotitis
- Sialogram

POLYCYSTIC (DYSGENETIC) DISEASE OF PAROTID GLANDS

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Least common
Developmental malformation of the duct

CLINICAL FEATURES

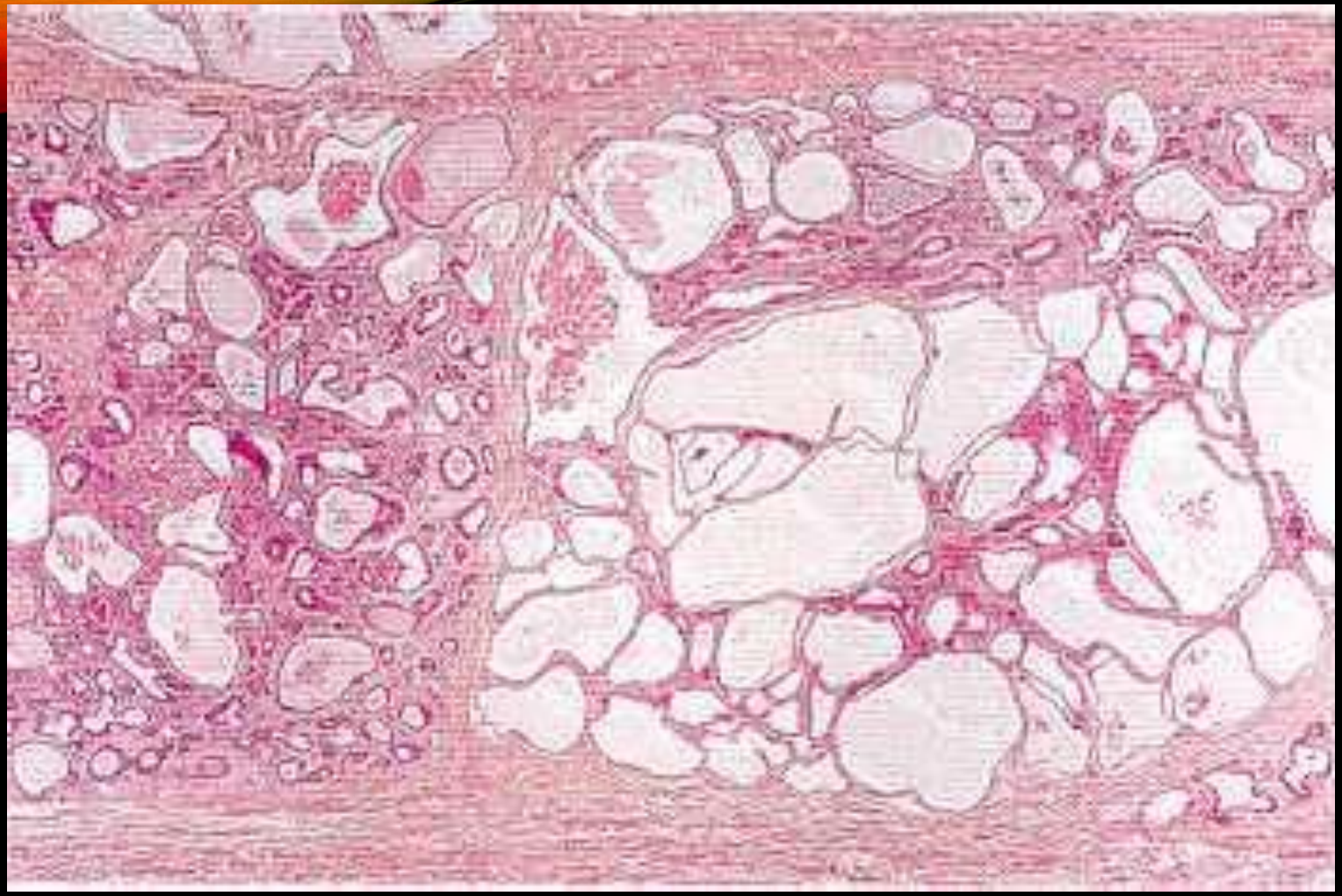
- Female (7/8)
- Recurrent painless swelling of the involved gland
- Swelling is due to the anomaly of the gland

GROSS

- Exaggerated lobularity of the subcapsular surface
- Cut surface
  - Mottled yellow ivory nodules
  - With fine spongy consistancy

HISTOLOGY

- Lobules markedly distended
- Cysts → honey combed or lattice like appearance
- Squamous cuboidal or Columnar cells have abundant eosinophilic cytoplasm
- Lumen contain eosinophillic material
- Spheroliths and microliths



- The lobular architecture is preserved, but variably sized cysts have replaced the normal lobular-ductal units.
- The cysts are formed by dilatation of the ducts and are lined by attenuated epithelial cells.

DIFFERENTIAL DIAGNOSIS

- Mucoepidermoid carcinoma
- Acinic cell adenocarcinoma
- Cystadenocarcinoma
- Differentiation
  - Wide spread involvement
  - Variable epithelial lining
  - Presence of spheroliths and microliths
  - Lack of inflammation

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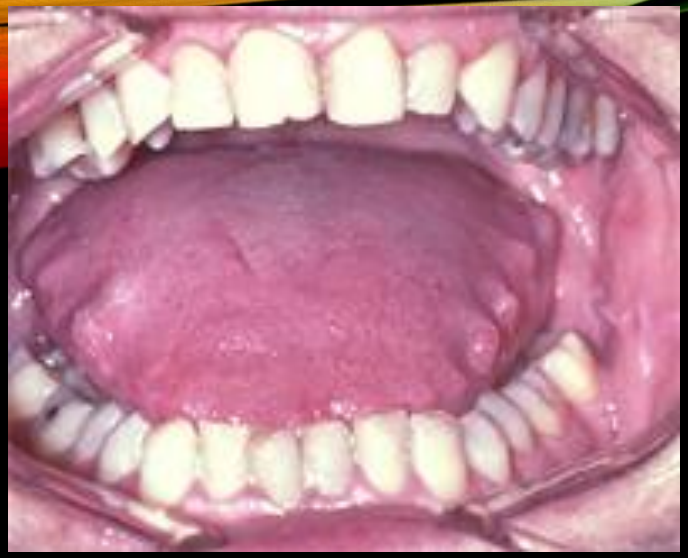
DEVELOPMENTAL DISTURBANCES OF TONGUE

AGLOSSIA / MICROGLOSSIA SYNDROME

- Extremely rare
- Associated with
  - Anomalis of hand and feet
  - Cleft palate
  - Dental agenesis
- Microglossia
  - Lack of muscle stimulus
    - Mandible fails to grow forward

MACROGLOSSIA

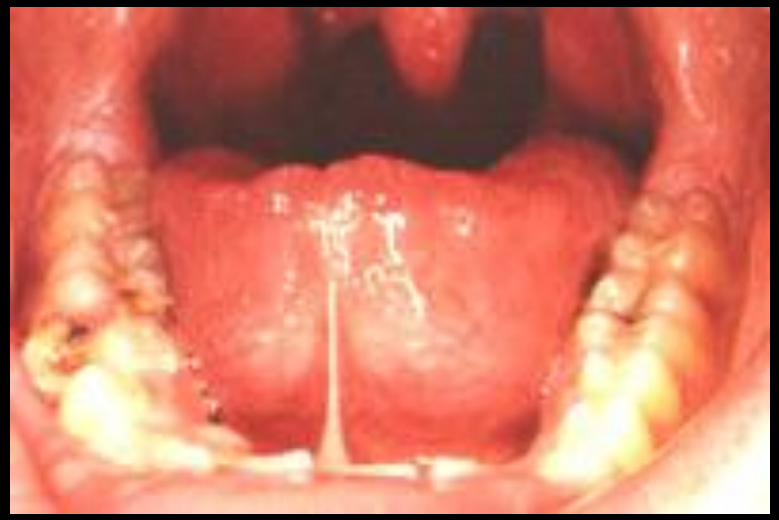
- Papyrus Ebers 1550 BC
- True macroglossia
  - Congenital
  - Acquired
- pseudo macroglossia
  - Relative small jaw
  - Atonia
  - Vitamin deficiencies
  - Neoplasms displacing tongue

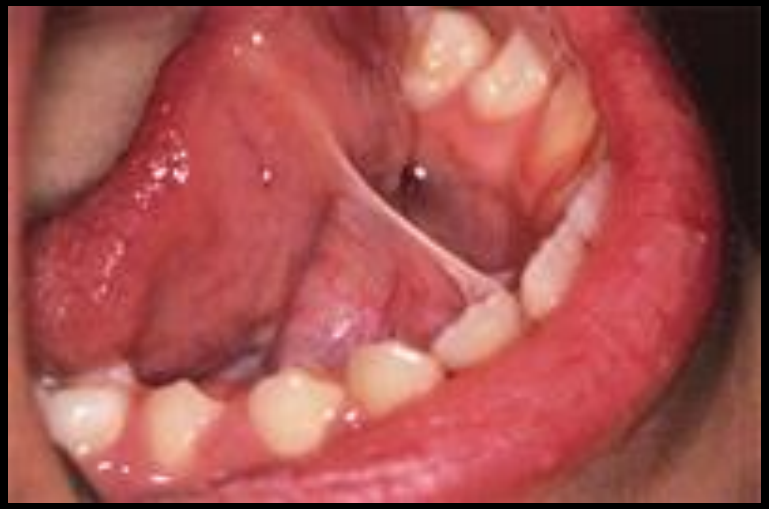


- Congenital
  - Muscle hypertrophy
  - Gland hyperplasia
  - Downs syndrome
  - Beckwith's weidmann's
  - Lymphangioma
  - Gargoylism
- Acquired
  - Hypothyroid
  - Syphillis
  - Candidiasis
  - Acromegaly
  - Amyloidosis
  - Sarcoidosis

ANKYLOGLOSSIA

- Short lingual frenum
- Speech problem

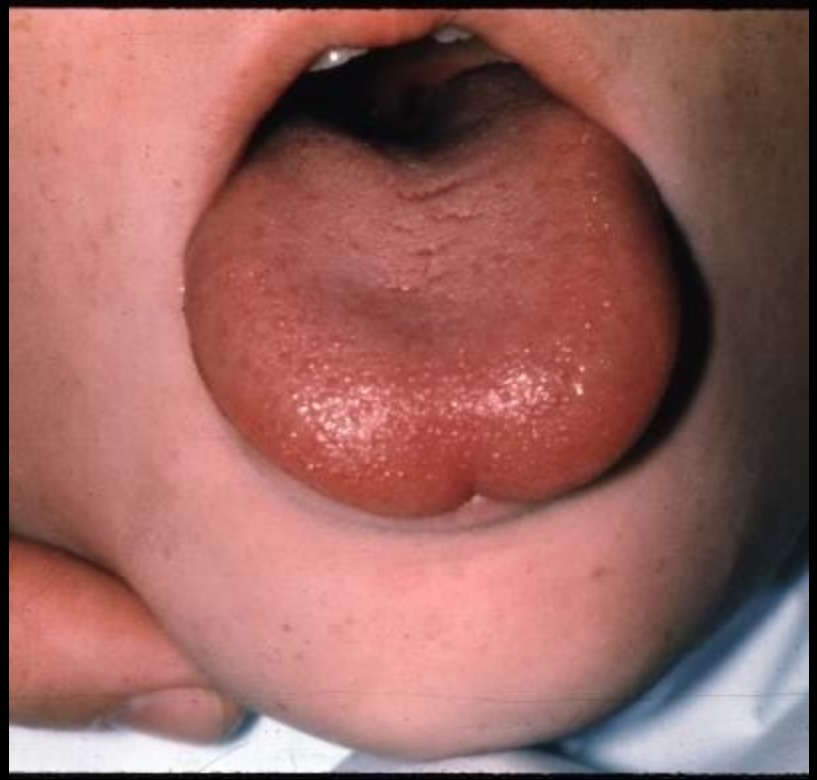




- Frenectomy

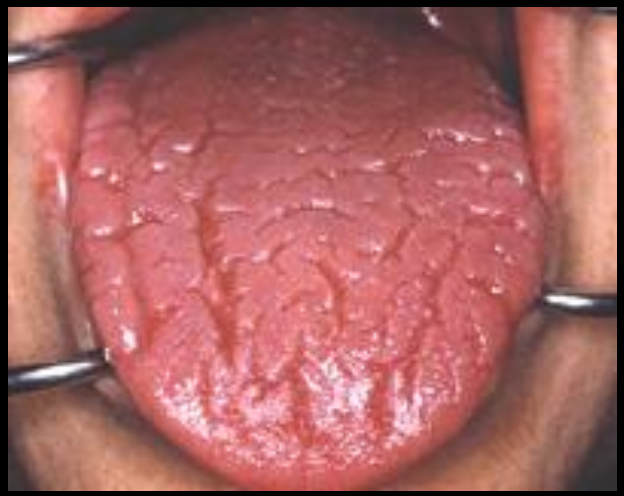
CLEFT TONGUE

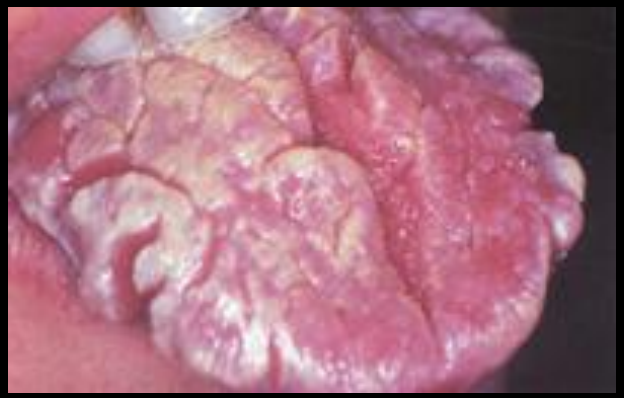
- Deep groove in midline of the dorsal tongue
- Associated with
  - Orofacial digital syndrome



FISSURED TONGUE (SCROTAL TONGUE)

- Grooves of varying depth
- Melkersson Rosenthal syndrome
  - Facial palsy
  - Chelitis granulomatosa
  - Fissured tongue
- Downs syndrome

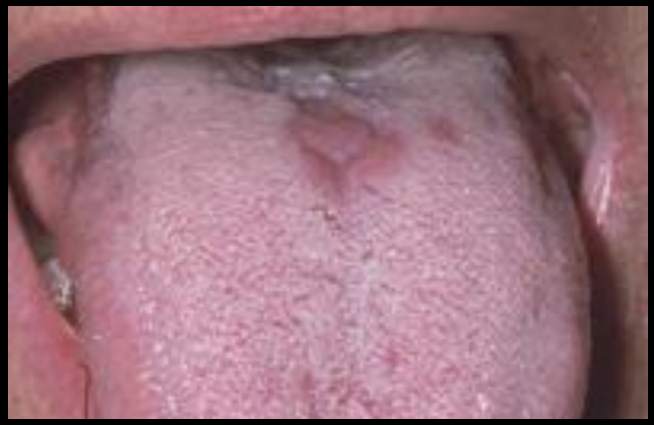


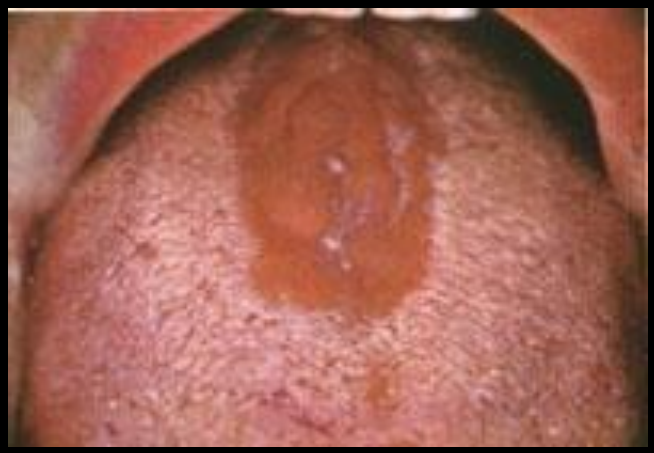


Histology : loss of filiform papillae and neutrophillic microabscesses

MEDIAN RHOMBOID GLOSSITIS

- Dorsal surface of the tongue along the midline, just anterior to the foramen cecum
- rhomboid or oval, well-demarcated shape
- red, flat or slightly multilobulated smooth,
- depapillated surface
- 1 to 3 cm
- usually asymptomatic

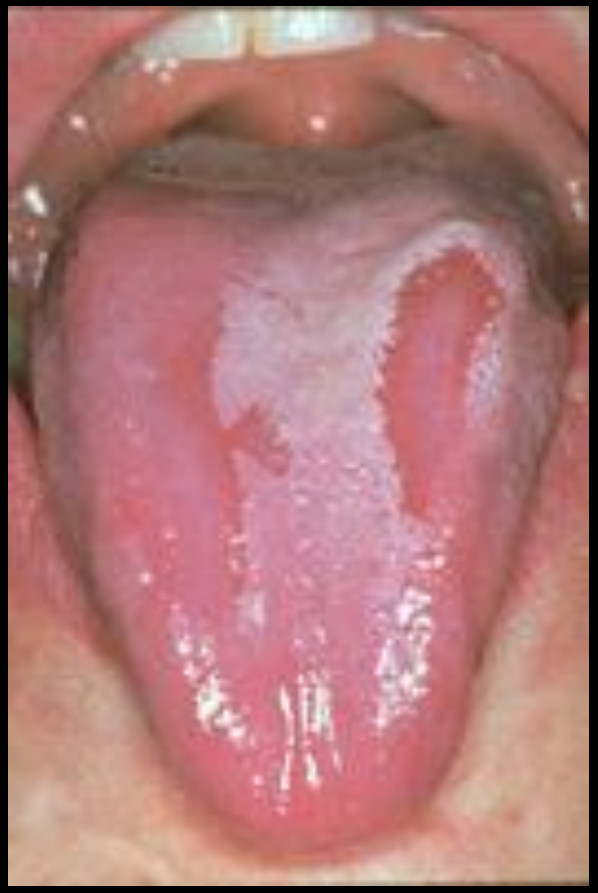


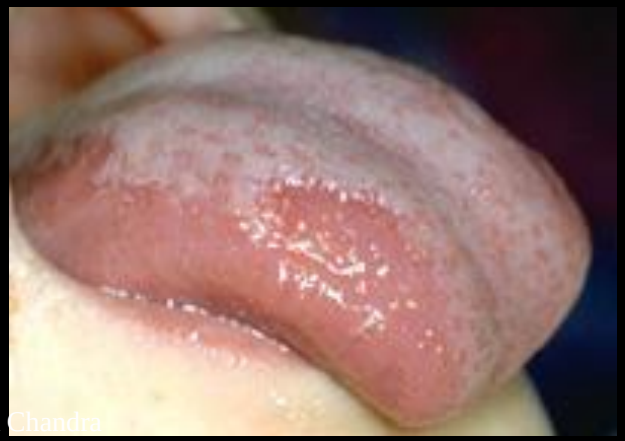


- Re-termed as POSTERIOR MIDLINE ATROPHIC CANDIDIASIS
- Atrophic stratified squamous epithelium
- Moderately fibrous CT
- Chronic candidal infection
- Always antifungal therapy prior to biopsy

BENIGN MIGRATORY GLOSSITIS

- Psorasiform mucositis
- Multiple sensitive irregularly shaped erythematous patches on the tongue
- Arcuate white rims that enlarge and change





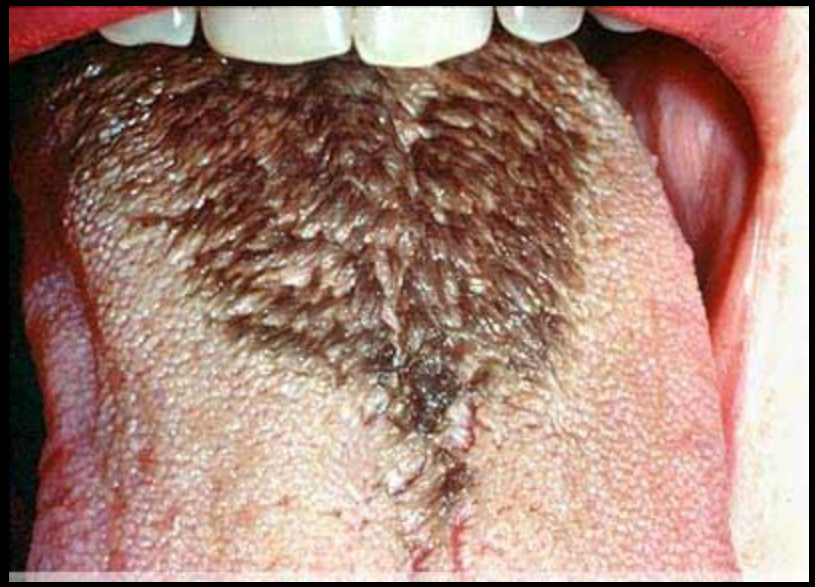
- Associations with human leukocyte antigen DR5 (HLA-DR5), DRW6 (HLA-DRW6), and Cw6 (HLA-Cw6)
- Similar to psoriasis
- Histopathology
  - Neutrophillic exocytosis
  - Monro's abscess
  - Thin long rete ridges
  - Small epithelium over the papillae

HAIRY TONGUE

- Defective desquamation of filiform papillae
- Black – brown to white
- Etiology
  - Hypertrophy of filiform
  - Lack of mechanical stimulation
  - Tobacco
  - Coffee

CLINICAL FEATURES

- M > F
- 1 – 15 mm papillae
- Tickling soft palate
- Asymptomatic
  - Candida → glossopyrosis
- Halitosis

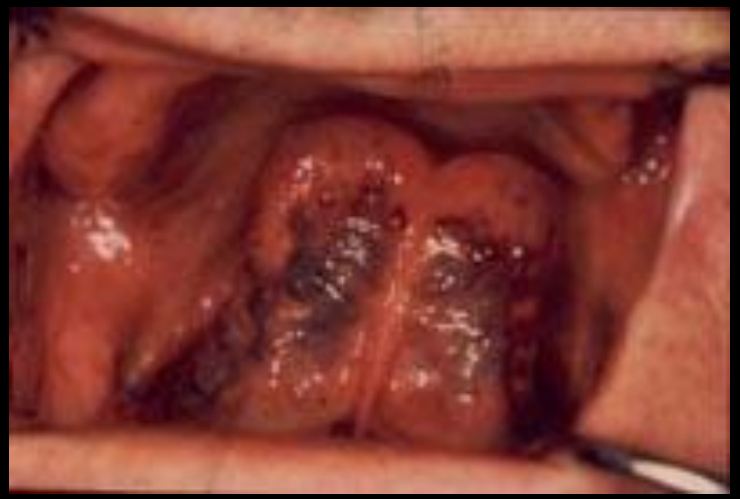


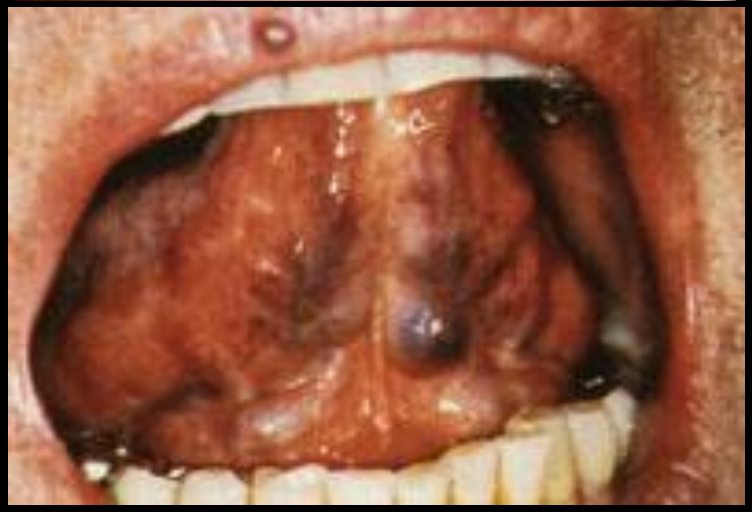
HISTOLOGY

- Mild elongated papillae
- Mild hyper keratosis
- Occational inflammatory cells
- Accumulated debris

LINGUAL VARICES

- Varix → Dialated, tortuous Vein
  - Increased hydrostatic pressure
  - Poorly supported by surrounding tissue
- Lingual Ranine veins
  - Red to purple shot like cluster of vessels
  - Ventral and lateral surfaces





- No direct association between varicosities and organic diseases
- Kleinman
  - Aging process
  - < 50 years if present
    - Premature aging

DEVELOPMENTAL DISTURBANCES INVOLVING THE TOOTH SIZE

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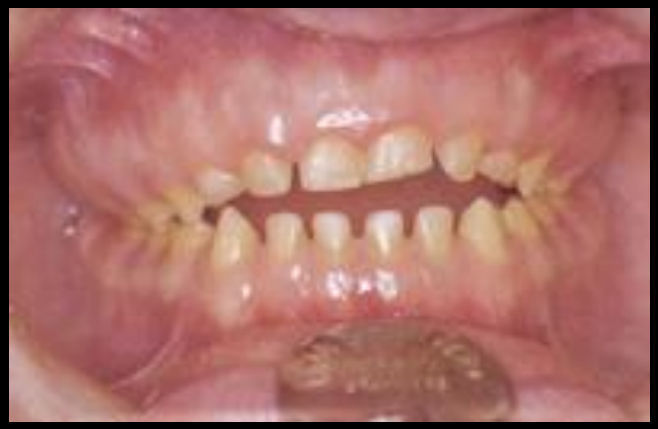
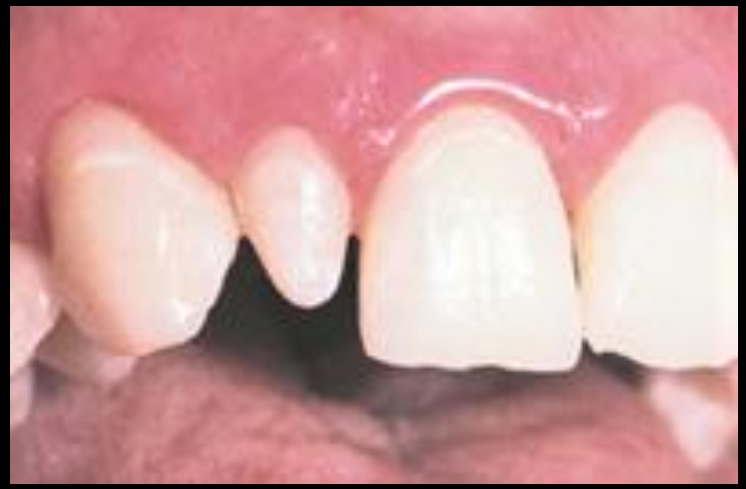
MICRODONTIA

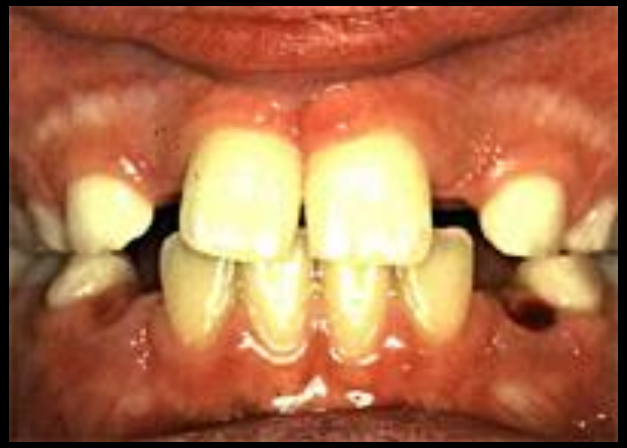
- Teeth smaller than normal
- Types
  - True generalised
    - All teeth smaller than normal
    - Pitutary dwarfism
  - Relative generalized
    - Normal or slightly smaller
    - Jaws larger
  - Microdontia of single tooth
    - Maxillary lateral (peg lateral)
    - Third molar

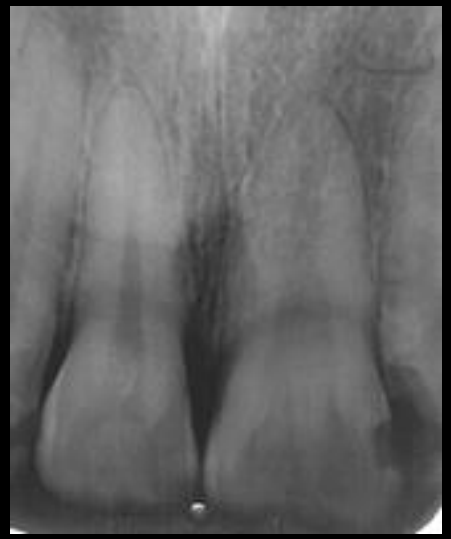




MACRODONTIA

- True generalised macrodontia
  - Pitutary gigantism
- Relative generalised
  - Hereditary
  - Relative larger size
- Macrodontia of single teeth
  - Hemihypertrophy
    - One side larger





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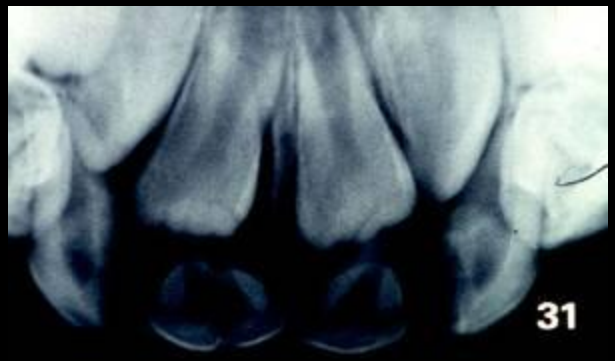
DEVELOPMENTAL DISTURBANCES OF SHAPE OF TEETH

GEMINATION

- Attempted division of single tooth germ
- Complete or incompletely seperated crowns
  - Single root and root canal
- DD
  - Fusion b/n normal teeth and supernumerary tooth



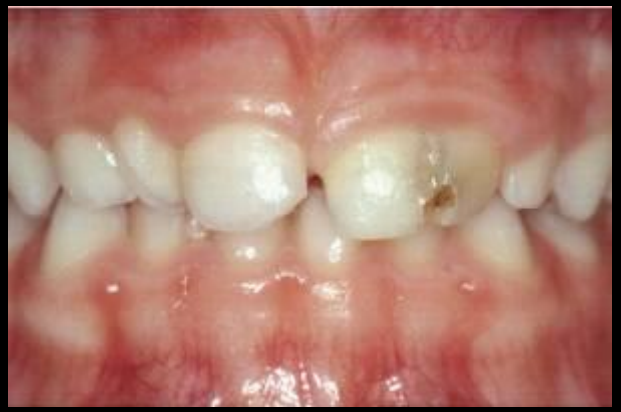
TWINNING

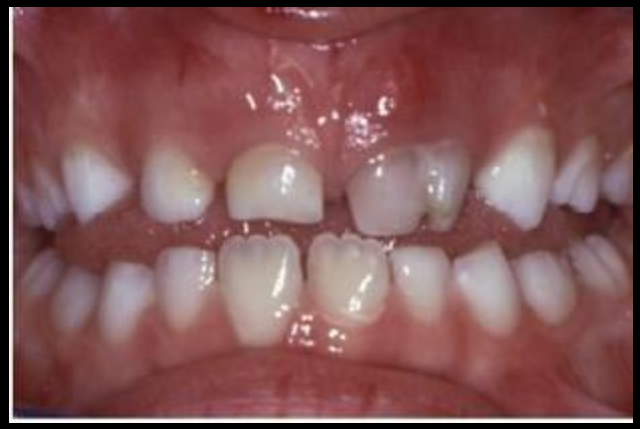
- *Schizodontia*

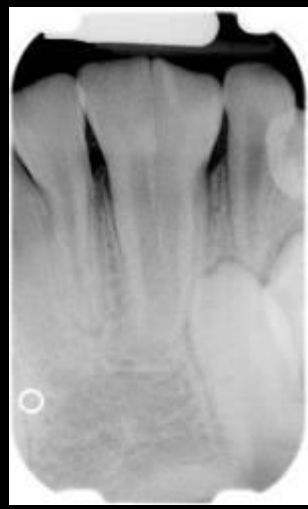
- Complete cleavage of tooth bud
- Extra tooth formation
  - One normal and one supernumerary

FUSION (*SYNODONTIA*)

- Union of two normally seperated tooth germs
  - Complete / Incomplete
  - Before calcification
    - Complete fusion
  - Only roots
- Pathogenesis
  - Physical force / pressure
- Deciduous>permanent

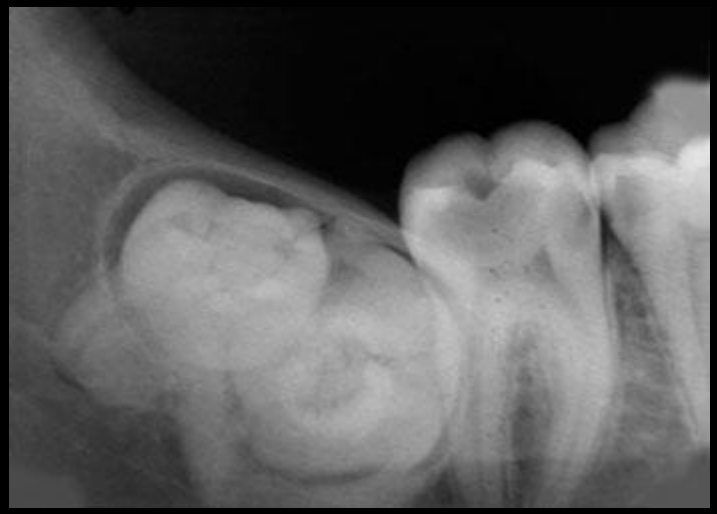


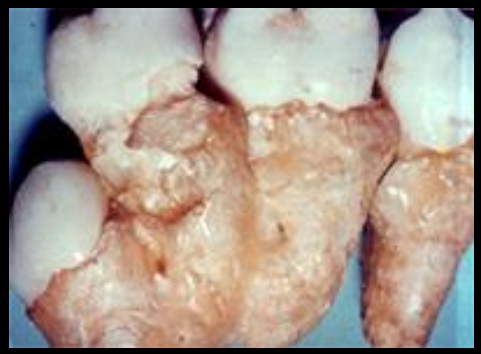


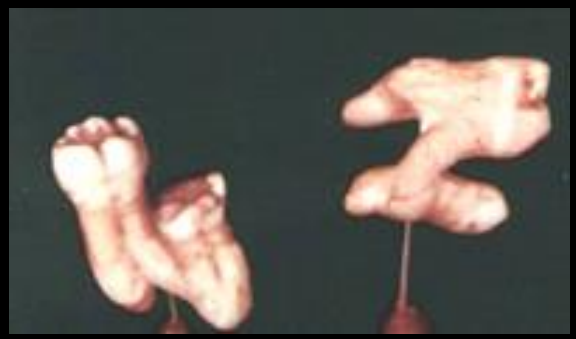


CONCRESCENCE

- Form of fusion
  - After root completion
  - United by cementum
- Cause
  - Trauma
  - Crowding
- Types
  - True → union during dev
  - Acquired → after root completion → hypercementosis

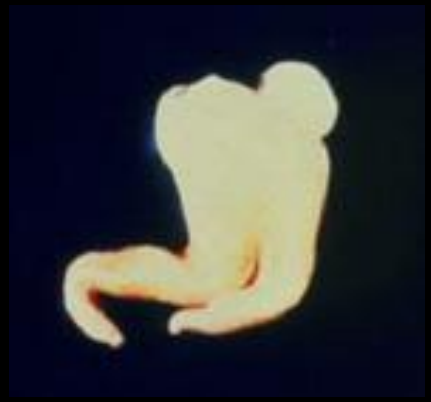


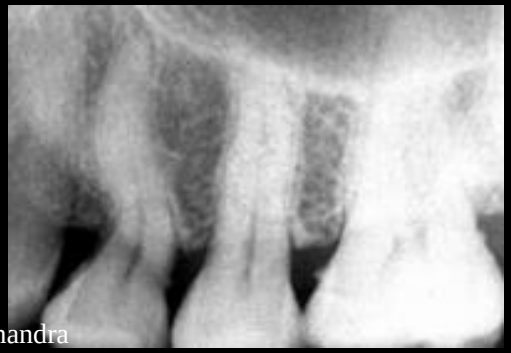


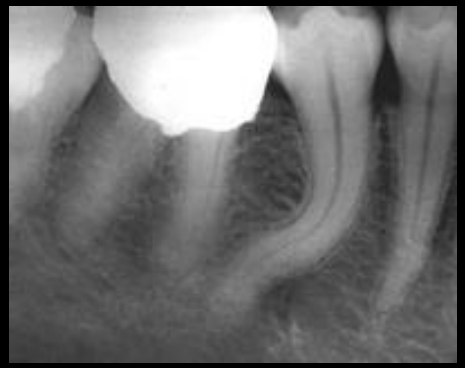


DILACERATION

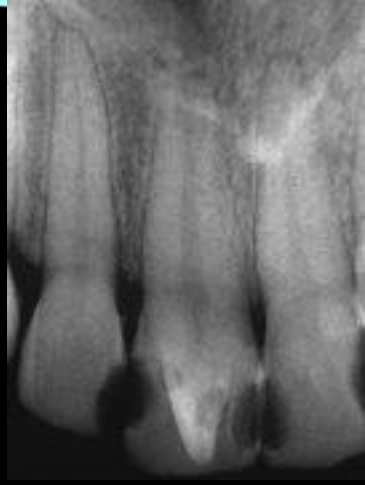
- Angulation
  - Sharp bend or curve
  - Root / crown of tooth
- Etiology
  - Trauma
  - Deciduous injures the permanent bud
- Radiograph always needed prior to extraction



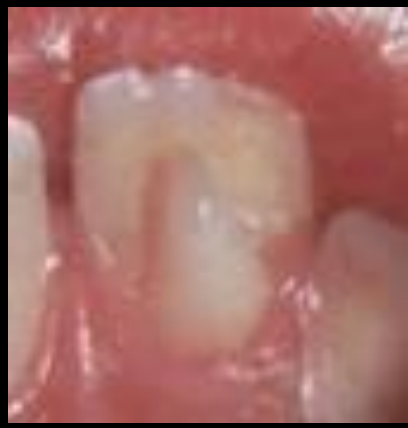


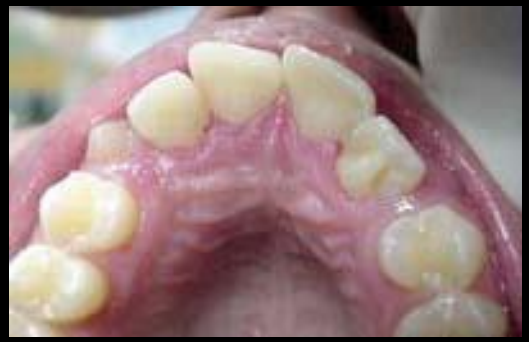


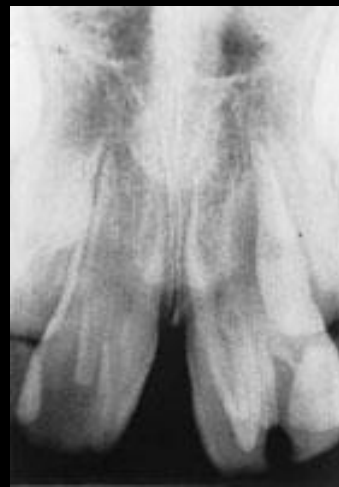
TALON'S CUSP



- Cingulum areas
  - Maxillary or mandibular incisors
  - Deep developmental grooves
  - Normal enamel and dentine
  - Normal pulp horn
- *Rubinstein Taybi syndrome*
  - Developmental retardation
  - Broad thumb's and great toes
  - Incomplete decent of testes

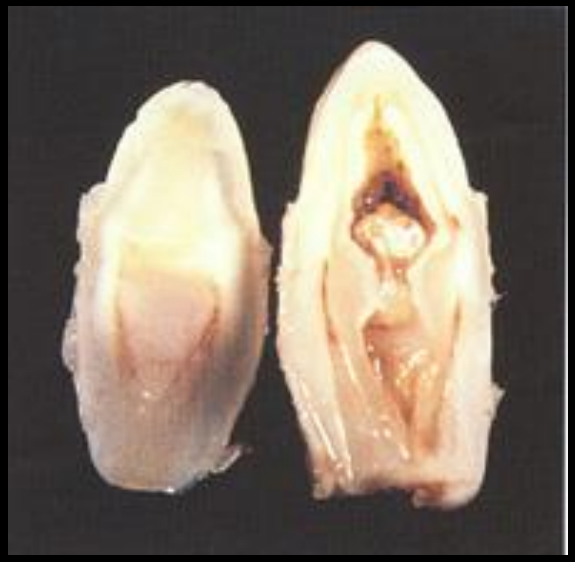






DENS IN DENTE (DENS INVAGINATUS)

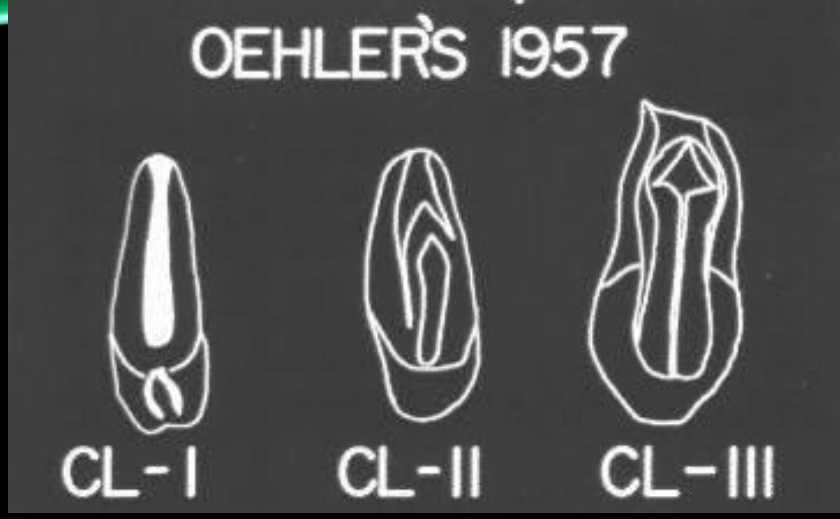
- Etiology
  - Invagination in the surface of tooth crown before calcification
    - Growth retardation
    - Trauma → localised external pressure
    - Focal growth stimulation
- Maxillary lateral incisors
  - Accentuation of lingual pit

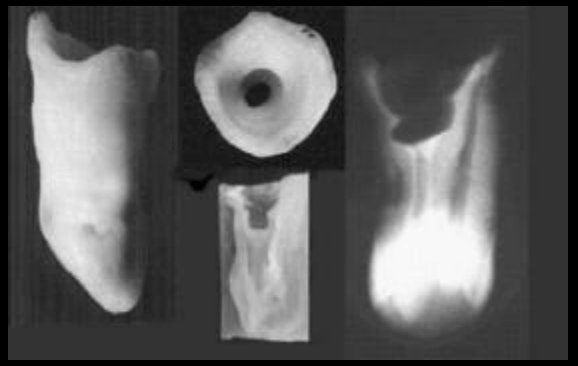


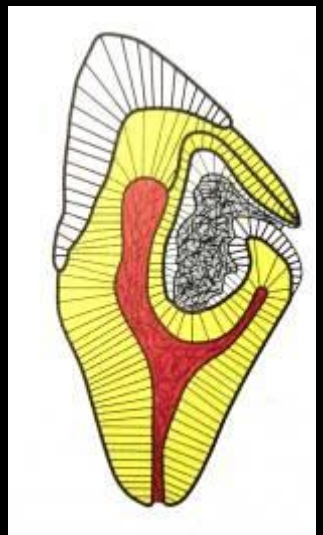
CLASSIFICATION

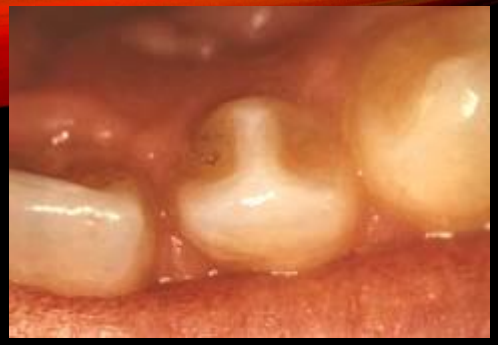
- Oehler's

- Hallet's 1953
  - Type 1:
    - Definite cleft parallel
    - No expansion
  - Type 2
    - Extends towards pulp chamber
  - Type 3
    - Deep into pulp chamber + dialated
  - Type 4
    - Occludes coronal pulp chamber
    - Beyond CEJ

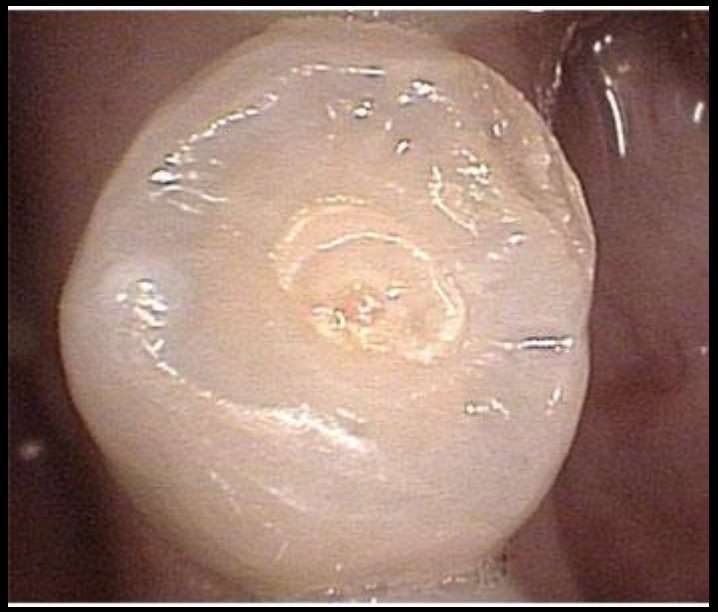








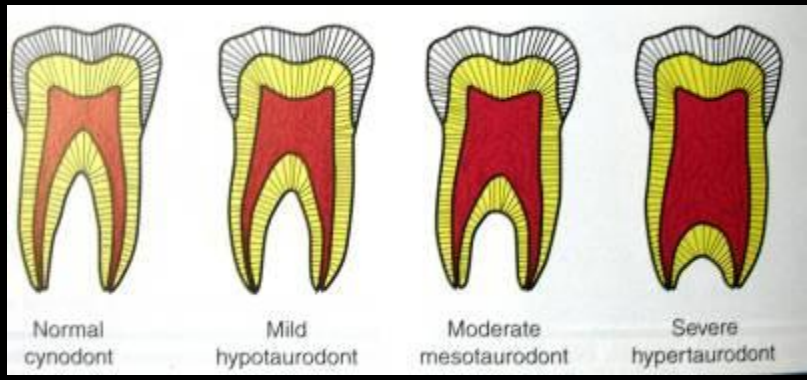
DENS EVAGINATUS



- Leong's premolar
- Pathogenesis
  - Proliferation and evagination
  - Odontogenic mesenchyme
- Clinical features
  - Mongoloid ancestry
  - Accessary cusp
  - Globule of enamel between cusps
  - Extra cusp → displacement of teeth, pulp exposure

TAURODONTISM

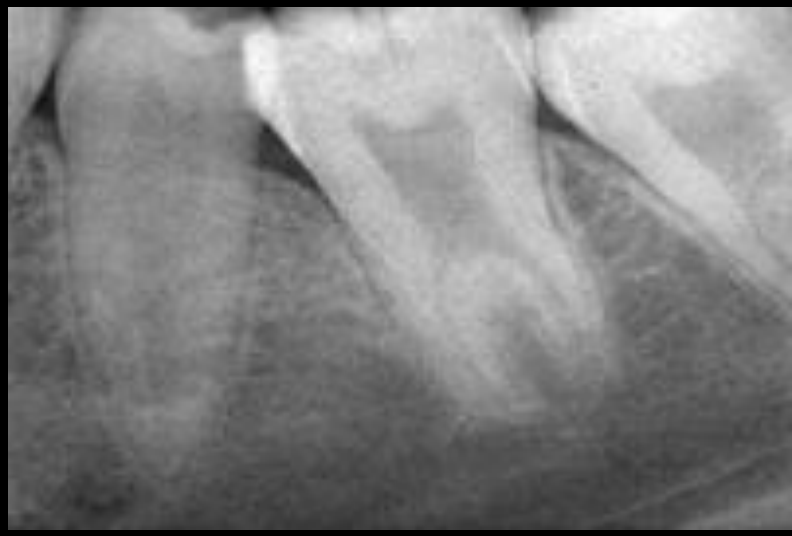
- Sir Arthur Keith 1913
- Bull like teeth
- Body of the teeth expanded at the expense of root.
- *Shaw* classification
  - Hypotaurodont (mildest)
  - Mesotaurodont
  - Hypertaurodont (at apex)



CAUSES OF TAURODONTISM

- Mendelian recessive trait
- Atavistic feature
- Mutation resulting from odontoblastic deficiency
- Failure of hertwigs root sheath to invaginate at proper horizontal level

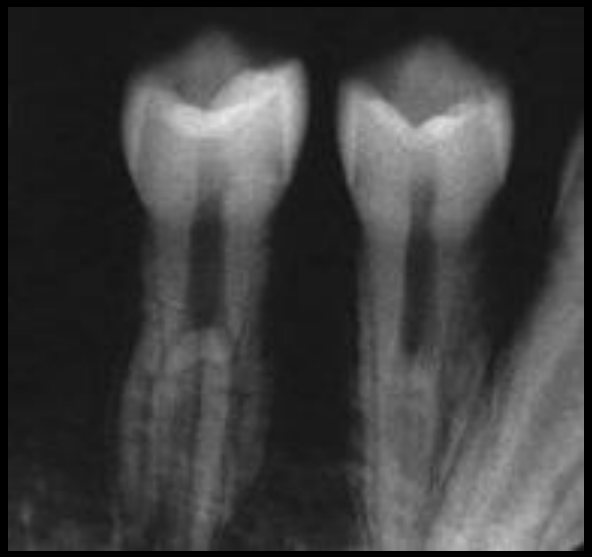
TAURODONTISM



- may occur in patients with
  - amelogenesis imperfecta,
  - Down syndrome, and
  - Klinefelter syndrome
    - Due to extra X
    - Male patients with taurodontism must have chromosome analysis performed

SUPERNUMERARY ROOTS

- Common
- Single root →
  - mandibular bicuspid & cuspids
- Molars most commonly affected
- Significant in exodontia



DEVELOPMENTAL DISTURBANCES IN NUMBER OF TEETH

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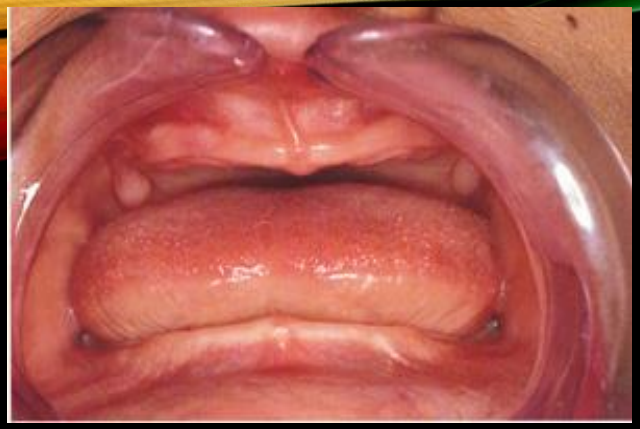
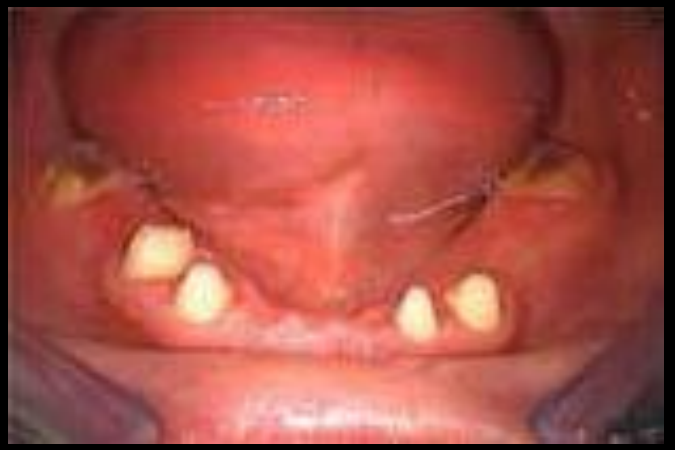


ANODONTIA

- True anodontia
  - Total
    - All the teeth are missing
    - May involve deciduous and permanent dentition
    - Hereditary ectodermal dysplasia
  - Partial
    - Hypo/oligodontia
    - 3rd molar > max lateral > second molar
- Pseudo anodontia
  - Total extraction



ANODONTIA : ETIOLOGY

- Familial tendency
- Point mutations
- Autosomal dominant

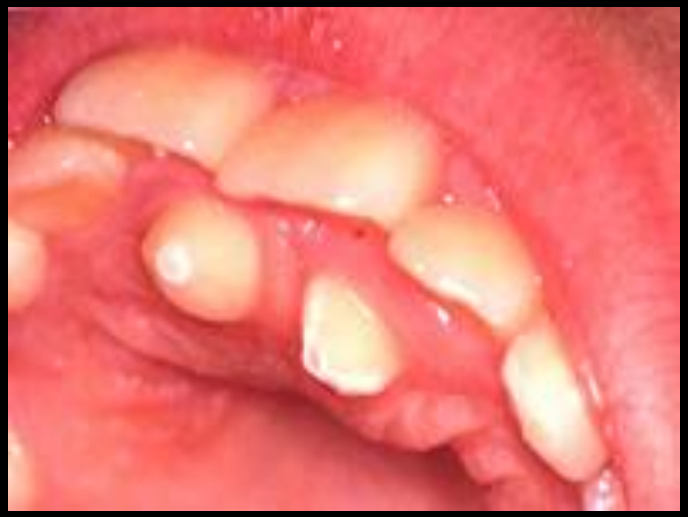
- X-ray irradiation
  - Single quadrant teeth missing

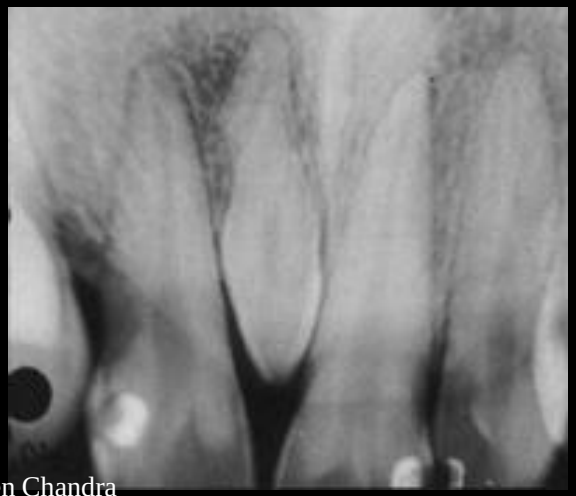
SUPERNUMERARY TEETH

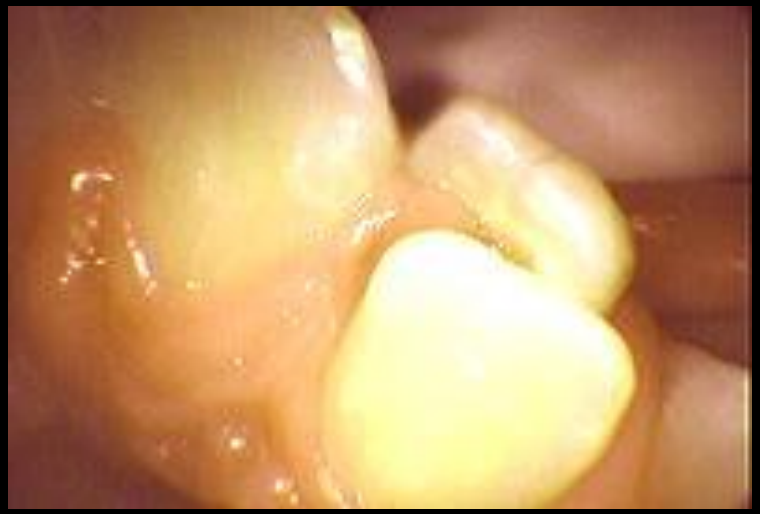
- Etiology
  - Extra tooth bud
  - Splitting of tooth bud
  - Hyperactivity theory
    - Local independent , conditioned hyper activity of dental lamina
- Associated with
  - Cleft lip and palate
  - Cleidocranial dysplasia
  - Gardner syndrome

CLASSIFICATION : SUPERNUMERARY TEETH

- Morphology and location
  - Conical
  - Tuberculate
  - Supplemental
  - Odontome



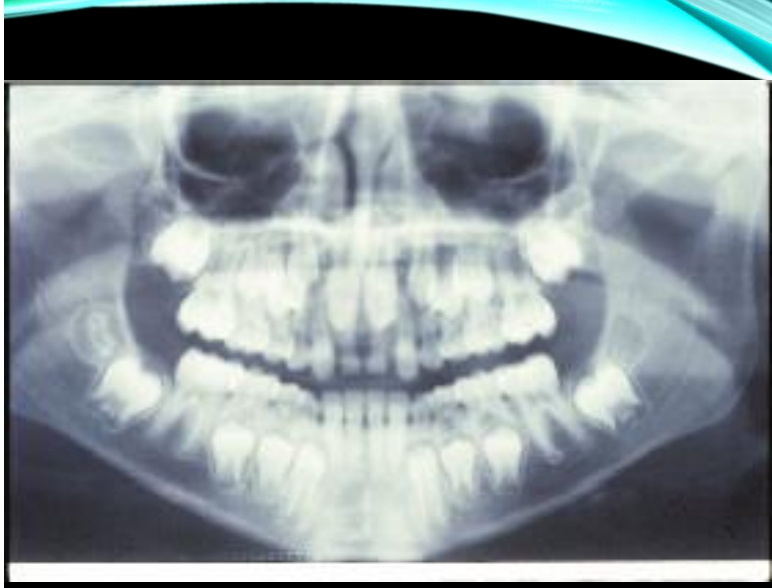


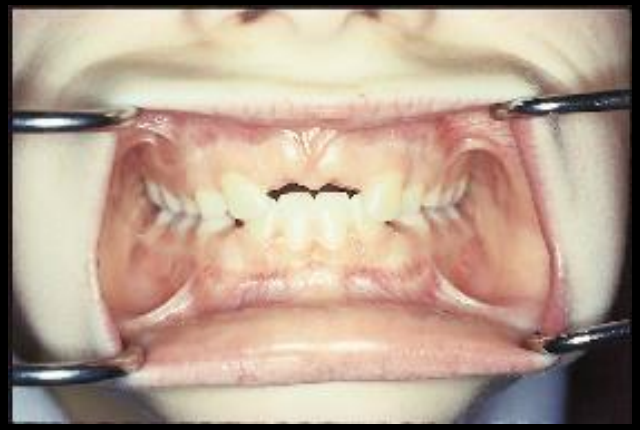


TYPES

- Conical
  - This small peg-shaped conical tooth
  - Commonly found in the permanent dentition
  - It develops with root formation ahead of or at an equivalent stage to that of permanent incisors and usually presents as a **mesiodens**.
    - inverted into the palate
    - horizontal position.
    - result in rotation or displacement of the permanent incisor, but rarely delays eruption.

- Tuberculate
  - More than one cusp or tubercle.
  - barrel-shaped and may be invaginated
  - Root formation is delayed compared to that of the permanent incisors.
  - Often paired
  - Commonly located on the palatal aspect of the central incisors.

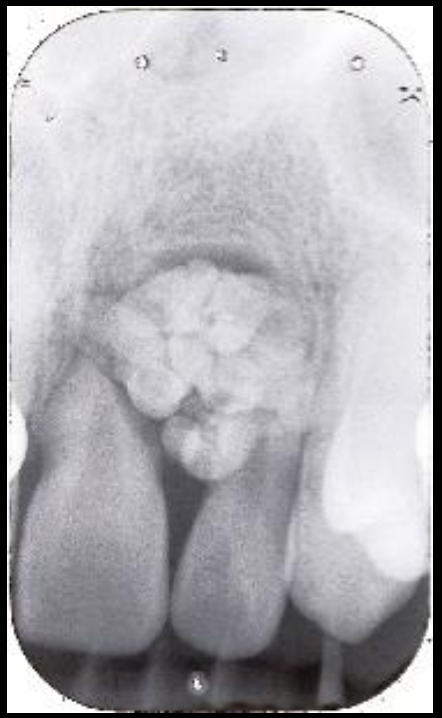




- Supplemental
  - Duplication of teeth in the normal series and is found at the end of a tooth series
  - The most common : permanent maxillary lateral incisor,
  - Majority supernumeraries found in the primary dentition are of the supplemental type

ODONTOMA (*HOWARD*)

- Category is not universally accepted
- Hamartomatous malformation rather than a neoplasm.
- Two types
  - complex composite odontoma
    - the diffuse mass of dental tissue which is totally disorganized
  - compound composite odontoma.
    - the malformation which bears some superficial anatomical similarity to a normal tooth



GARDNER'S SYNDROME

- Desmoid tumours
- Osteomas
- Polyposis of large intestine
- Sebaceous cysts
- Imacted supernumerary teeth

- ***Fader and Duncan***

- *Cause*
  - Pleiotropic gene
  - Autosomal dominant
  - Complete penetrance

PREDECIDUOUS DENTITION

- Hornified epithelial structures
- Over the crest of ridge on the gingiva

- At birth →natal teeth
- < 28 days eruption → neonatal teeth

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DEVELOPMENTAL DISTURBANCES IN STRUCTURE OF TEETH

ENAMEL HYPOPLASIA

- Incomplete or defective formation of organic enamel matrix
- Types
  - Hereditrary
    - Amelogenesis imperfecta
  - Environmental
    - Nutritional deficiency (Vit A, C , D)
    - Exanthematous diseases
    - Congenital syphillis
    - Birth injury
    - Ingesion of chemicals
    - Idiopathic causes

AMELOGENESIS IMPERFECTA

- Autosomal dominant
- Autosomal recessive
- X – linked
- Types
  - Hypoplastic (60-73%)
  - Hypocalcified (7%)
  - Hypomature (20-40%)

Table 2-1 *Classification of Amelogenesis Imperfecta*

TYPE	PATTERN	SPECIFIC FEATURES	INHERITANCE
IA	Hypoplastic	Generalized pitted	Autosomal dominant
IB	Hypoplastic	Localized pitted	Autosomal dominant
IC	Hypoplastic	Localized pitted	Autosomal recessive
ID	Hypoplastic	Diffuse smooth	Autosomal dominant
IE	Hypoplastic	Diffuse smooth	X-linked dominant
IF	Hypoplastic	Diffuse rough	Autosomal dominant
IG	Hypoplastic	Enamel agenesis	Autosomal recessive
IIA	Hypomaturation	Diffuse pigmented	Autosomal recessive
IIB	Hypomaturation	Diffuse	X-linked recessive
IIC	Hypomaturation	Snow capped	X-linked
IID	Hypomaturation	Snow capped	Autosomal dominant?
IIIA	Hypocalcified	Diffuse	Autosomal dominant
IIIB	Hypocalcified	Diffuse	Autosomal recessive
IVA	Hypomaturation-hypoplastic	Taurodontism present	Autosomal dominant
IVB	Hypoplastic-hypomaturation	Taurodontism present	Autosomal dominant

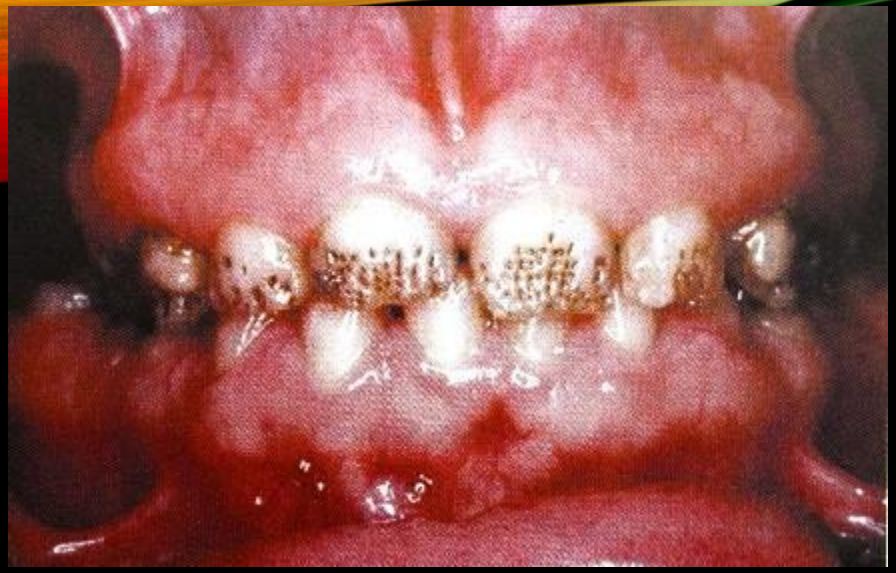
ETIOLOGY

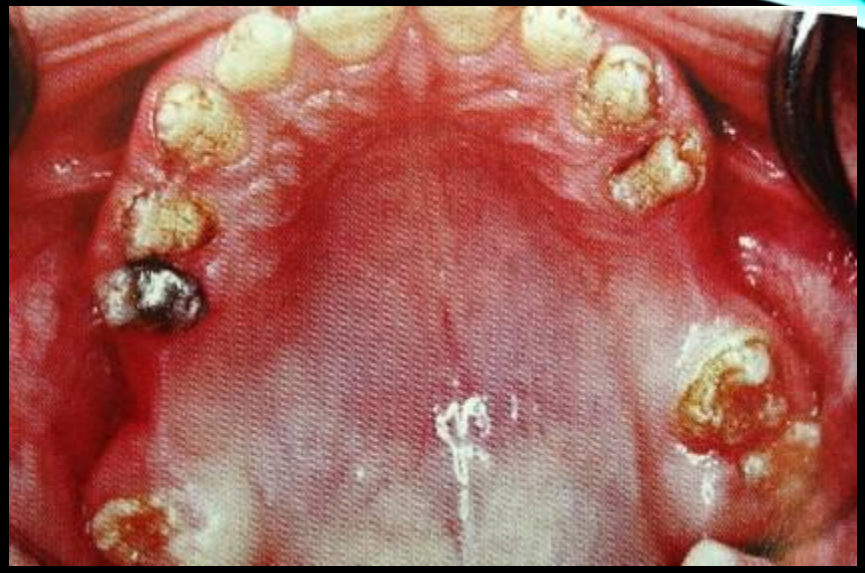
- Alterations in genes involved in formation and maturation of enamel
- DXS 85 at Xp22
  - Localization of amelogenin (*AMELX* and *AMELY*)
- Other genes involved
  - AMBN → ameloblastin
  - Enamelin → Multiple mutations *ENAM* gene mutations are associated with different autosomally inherited AI types

- Enamelysin:
  - *MMP20* gene located on chromosome 11
  - proteinase that cleaves amelogenin for processing the enamel matrix proteins
  - Enamelysin knockout mouse has a reduced enamel thickness, poorly mineralized enamel and the enamel lacks a prismatic structure.
- Kalikryn 4:
  - *KLK4* gene located on chromosome 19
  - Proteinase that is secreted predominantly during the maturation stage
  - Mutation of *KLK4* is associated with autosomal recessive hypomaturation AI that is characterized by poorly mineralized enamel.
- Tuftelin

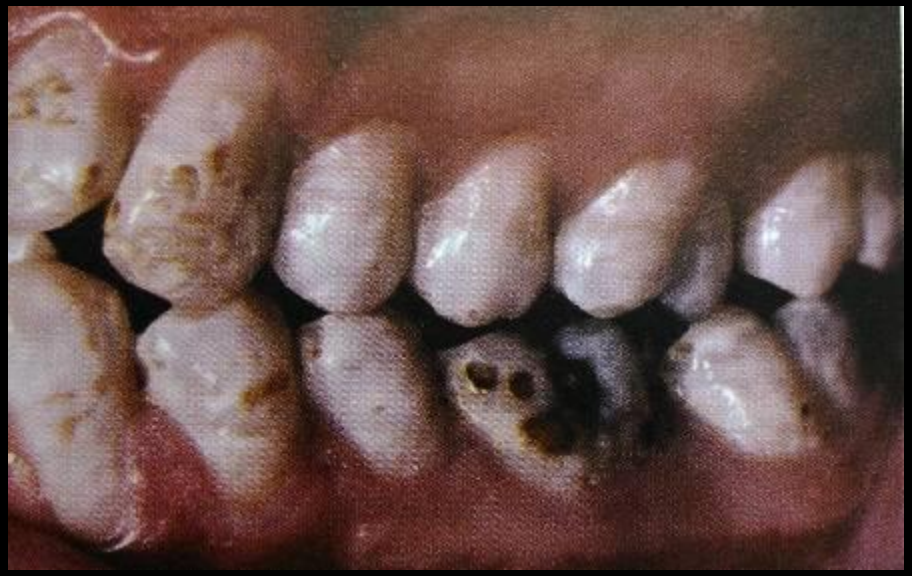
CLINICAL FEATURES

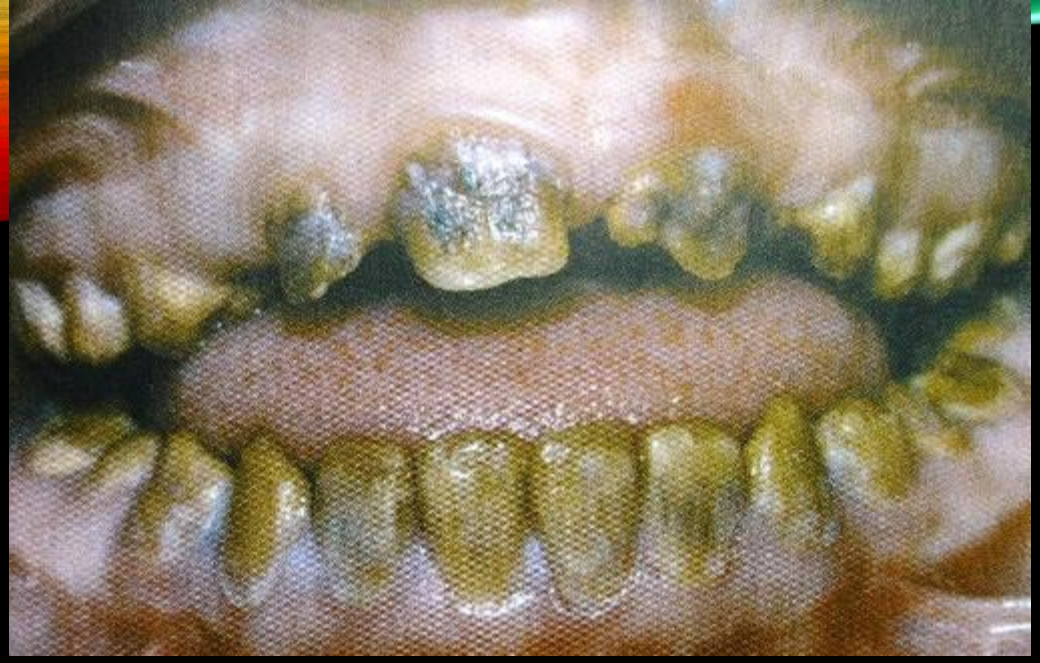
- teeth vary in color from white opaque to yellow to brown
- all teeth are affected, smaller and pitted
- normal pulps and dentin but reduced enamel





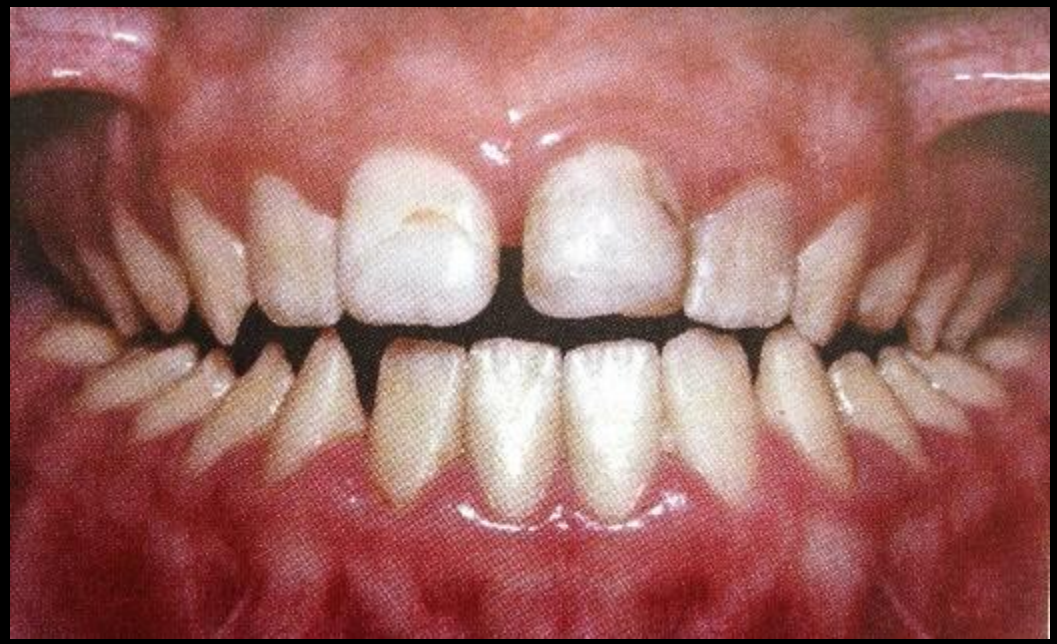
- Few small grooves
- Pits/ fissures
- Severe deep rows of pits
- Portion of enamel missing





Hypocalcified type

Smooth type



HISTOLOGY

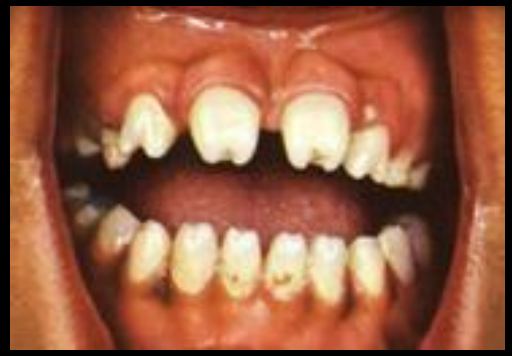
- Hypoplastic type
  - Disturbance of differentiation and viability of ameloblasts
- Hypo calcified type
  - Defects of matrix structure and mineral deposition
- Hypomaturation
  - Alterations in enamel rod and rod sheath structures

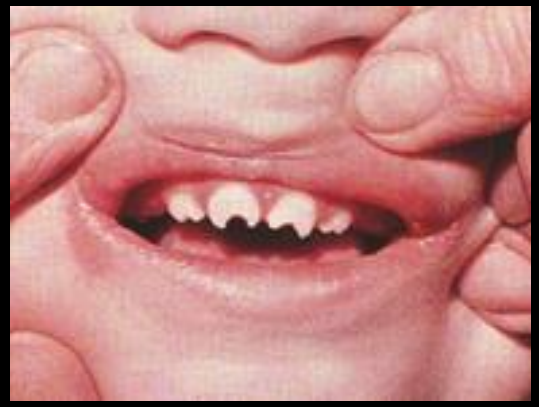
NUTRITIONAL DEFICIENCY AND EXANTHEMATOUS DISEASES

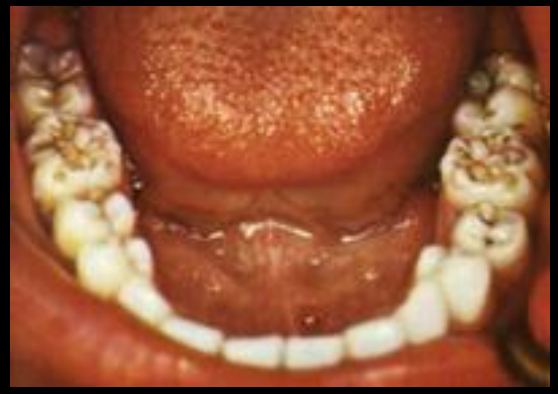
- Ameloblasts most sensitive
- Usually pitting variety
- 1 year after birth
  - Central
  - Lateral
  - Cuspid and
  - 1st molars affected

CONGENITAL SYPHILLIS

- Hutchinsons teeth
- Moons molars
- Hutchinsons triad







HYPOCALCEMIA

- Pitting variety
- Ca++ less than 6-8 mg / 100 ml
- Tetany
  - Vitamin D deficiency
  - Parathyroid deficiency

HYPOPLASIA DUE TO BIRTH INJURIES

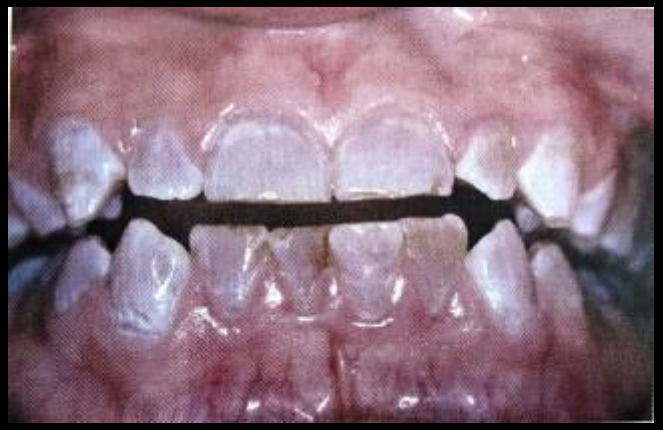
- Permanent maxillary incisors
- Maxillary / mandibular premolar
- Mild brownish discolouration → severe pitting

- TURNER'S TEETH / TURNER'S HYPOPLASIA

DENTINOGENESIS IMPERFECTA

- Autosomal dominant →
  - chromosome #4
  - Dentin sialophosphoprotein

- Affects both deciduous and permanent teeth
- Gray to yellowish brown
  - Tulip shape



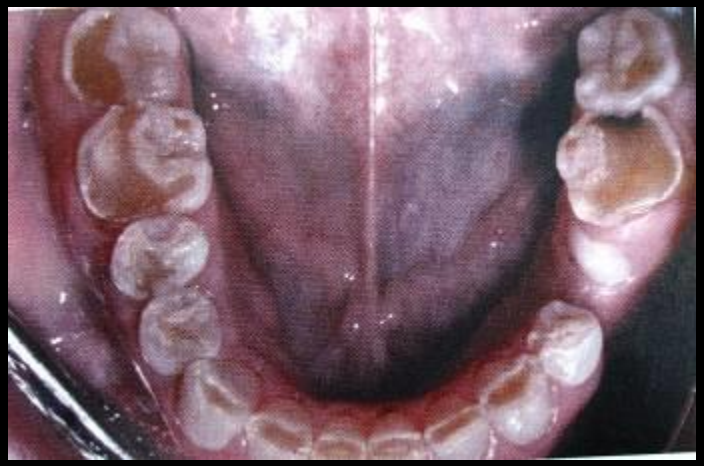
SHIELDS	CLINICAL PRESENTATION	WITKOP
Dentinogenesis imperfecta I	Osteogenesis imperfecta with opalescent teeth	Dentinogenesis imperfecta
Dentinogenesis imperfecta II	Isolated opalescent teeth	Hereditary opalescent teeth
Dentinogenesis imperfecta III	Isolated opalescent teeth	Brandywine isolate

PRESENT CLASSIFICATION

- Dentinogenesis imperfecta 1
  - Dentinogenesis without osteogenesis imperfecta
- Dentinogenesis imperfecta 2
  - Brandywine type

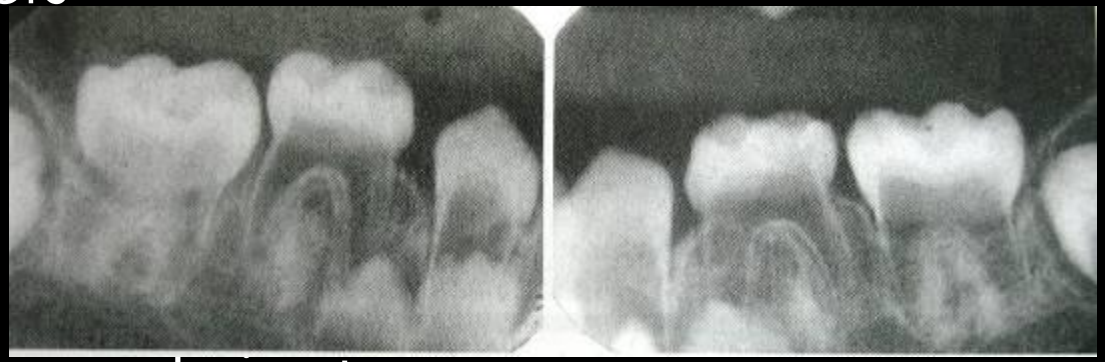
DENTINOGENESIS IMPERFECTA 1

- Mutation in DSPP gene chr 4q21.3
  - Encodes dentin phosphoprotein and sialoprotein
- Blue gray or amber brown opalescent
- Enamel may split readily



DENTINOGENESIS IMPERFECTA 2

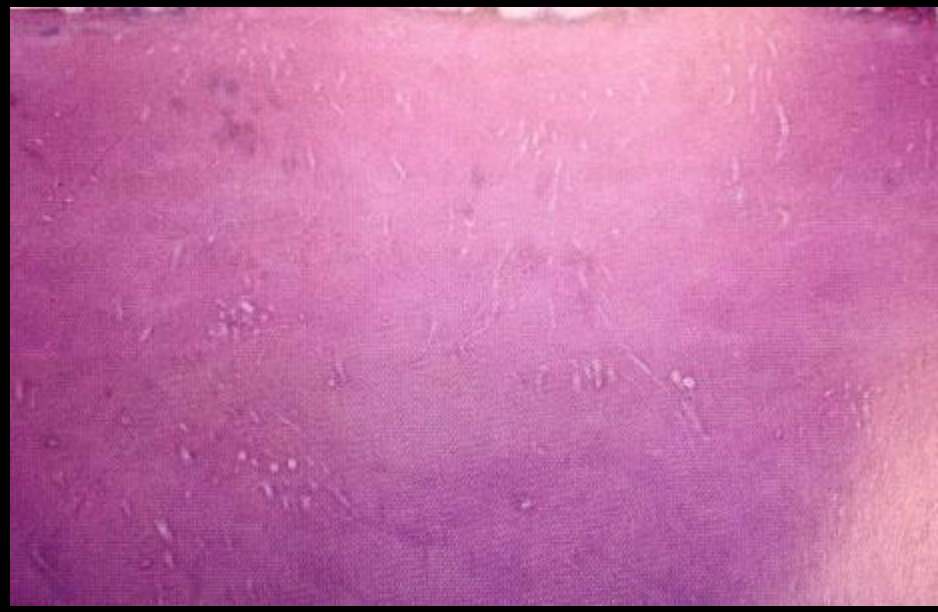
- Brandywine triracial isolate in maryland
- Clinical features
  - Rapid loss of enamel
  - Large pulp chambers
  - Shell teeth



- Dentin sialophosphoprotein + dentinmorphogenic protein + bone sialoprotein

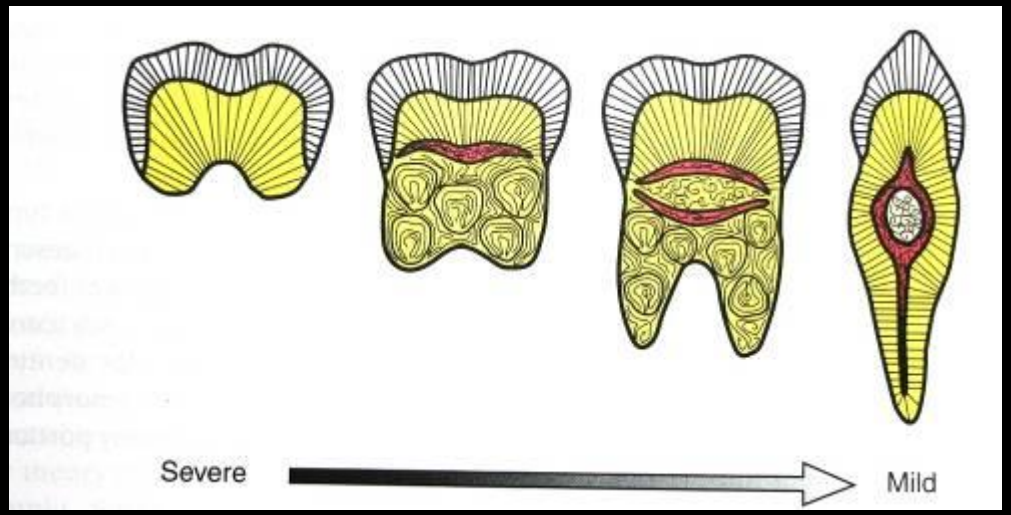
HISTOPATHOLOGY : DI

- Enamel normal
- Irregular tubules
- Areas of complete absence of tubules
- Physical characters
  - Reduced
    - water content
    - X- ray absorption
    - density



DENTIN DYSPLASIA

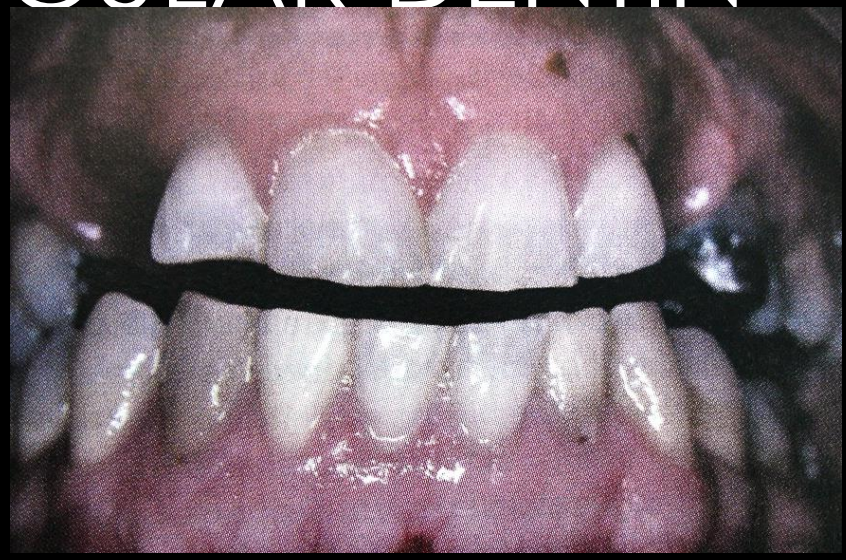
- Normal enamel
- Atypical dentin + abnormal pulp morphology
- Classification (WITKOP)
  - Type 1: Radicular dentin dysplasia (rootless teeth)

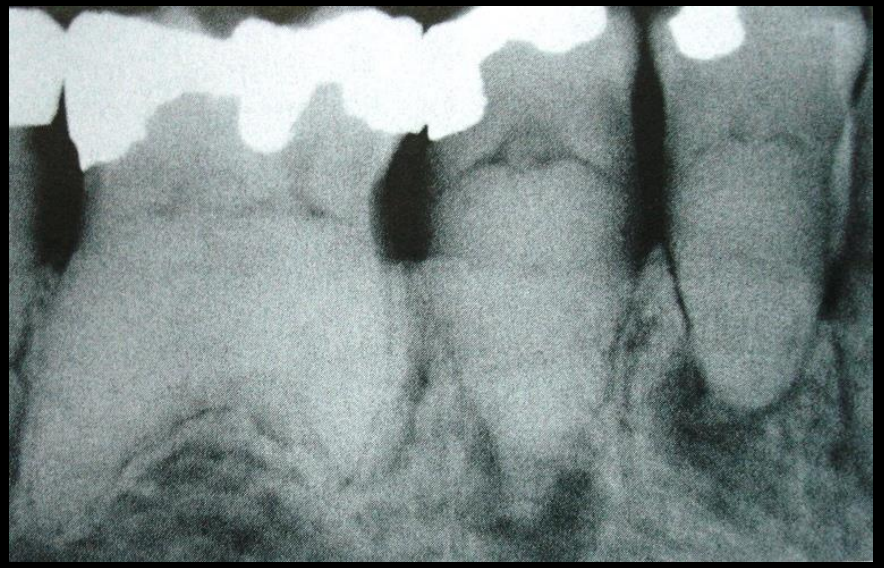


  - Type 2 : Coronal dentin dysplasia

RADICULAR DENTIN

- Autosomal dominant
- Both dentition affected
- Clinically → Appears normal
- Root is stunted
- Radiographically
  - Obliteration of pulp chamber
  - PA granuloma / cyst with out obvious reason

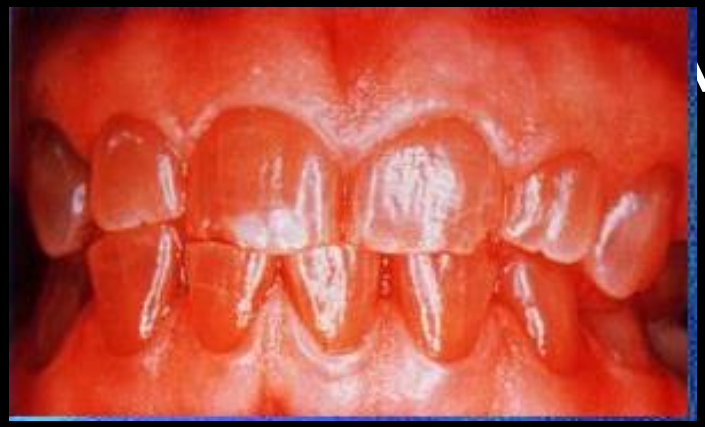


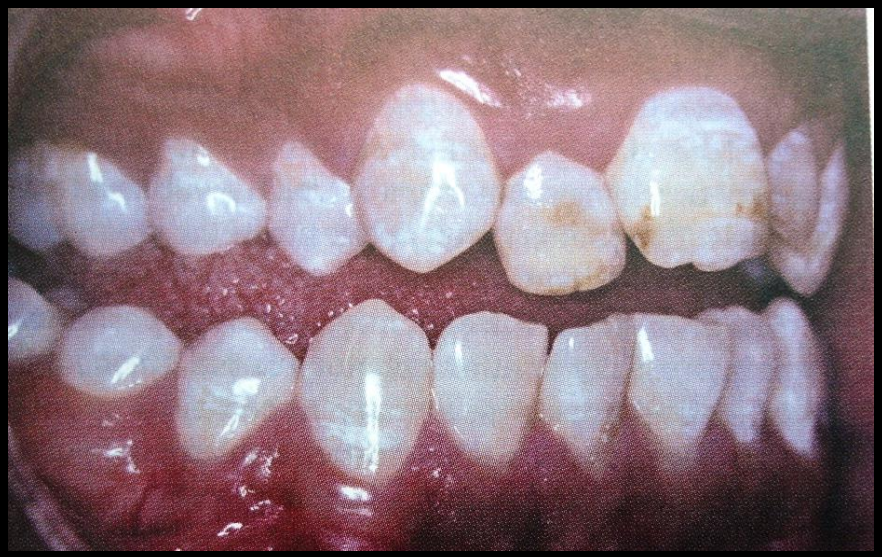


- Histology
  - Obliterated pulp chamber
  - Tubular dentin
  - Fused denticles
  - Osteodentin
  - Appearance of lava flowing around boulders

CORONAL DENTIN

- Autosomal dominant
- Both dentition affected
- Deciduous teeth
  - Appear yellow brown to blue
  - Complete obliteration
- Permanent normal
  - Thistle tube
  - Pulp stone most characeristic



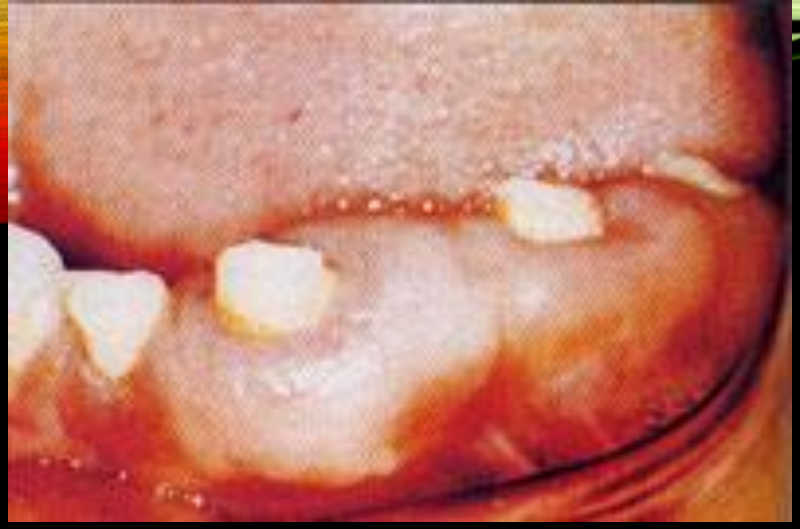


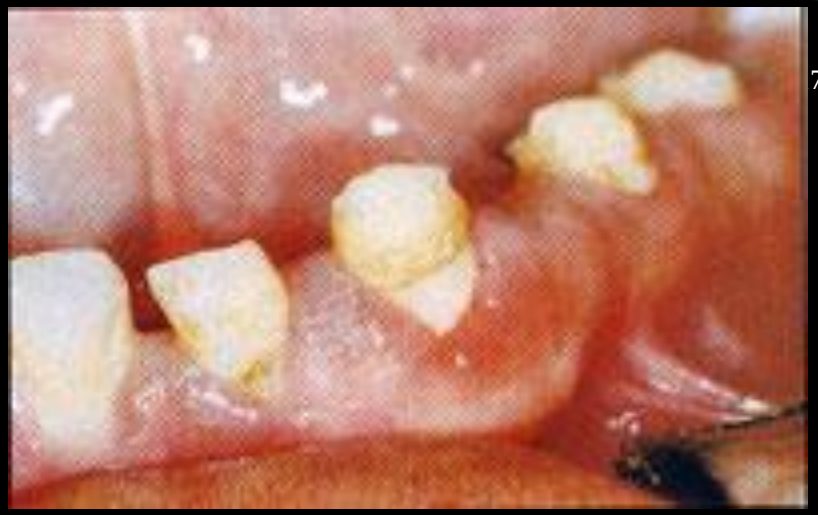
HISTOLOGY

- Deciduous tooth
  - Coronal dentin normal
  - Radicular dentin → atubular dentin
- Permanent
  - Normal
  - Pulp stones

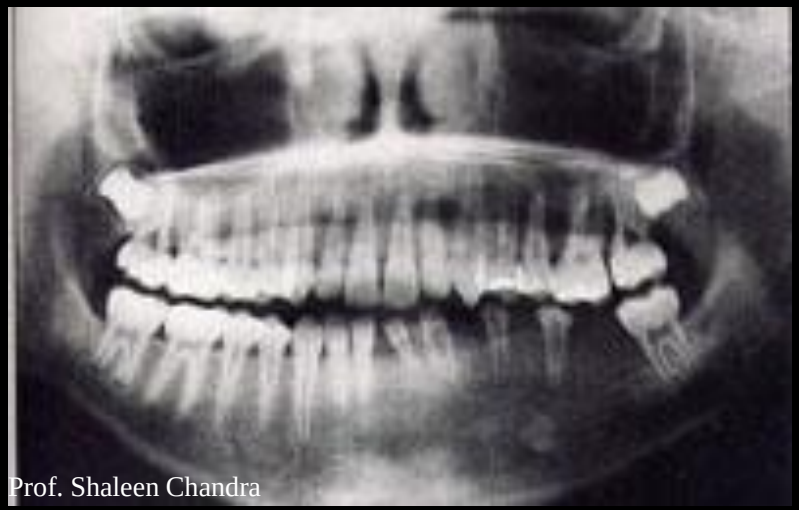
REGIONAL ODONTOGENIC DYSPLASIA

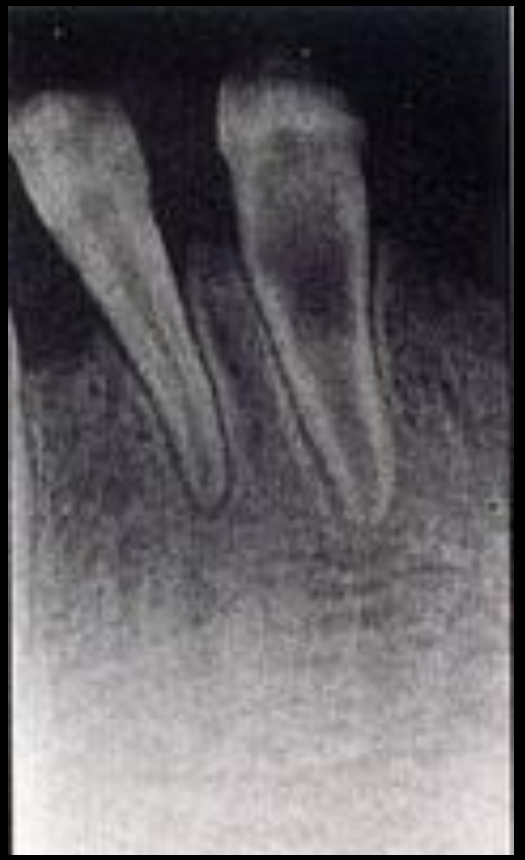
- Maxillary anterior region > mandible
- Etiology
  - Remnant viral infection
  - Vascular malformation (associated vascular nevi)





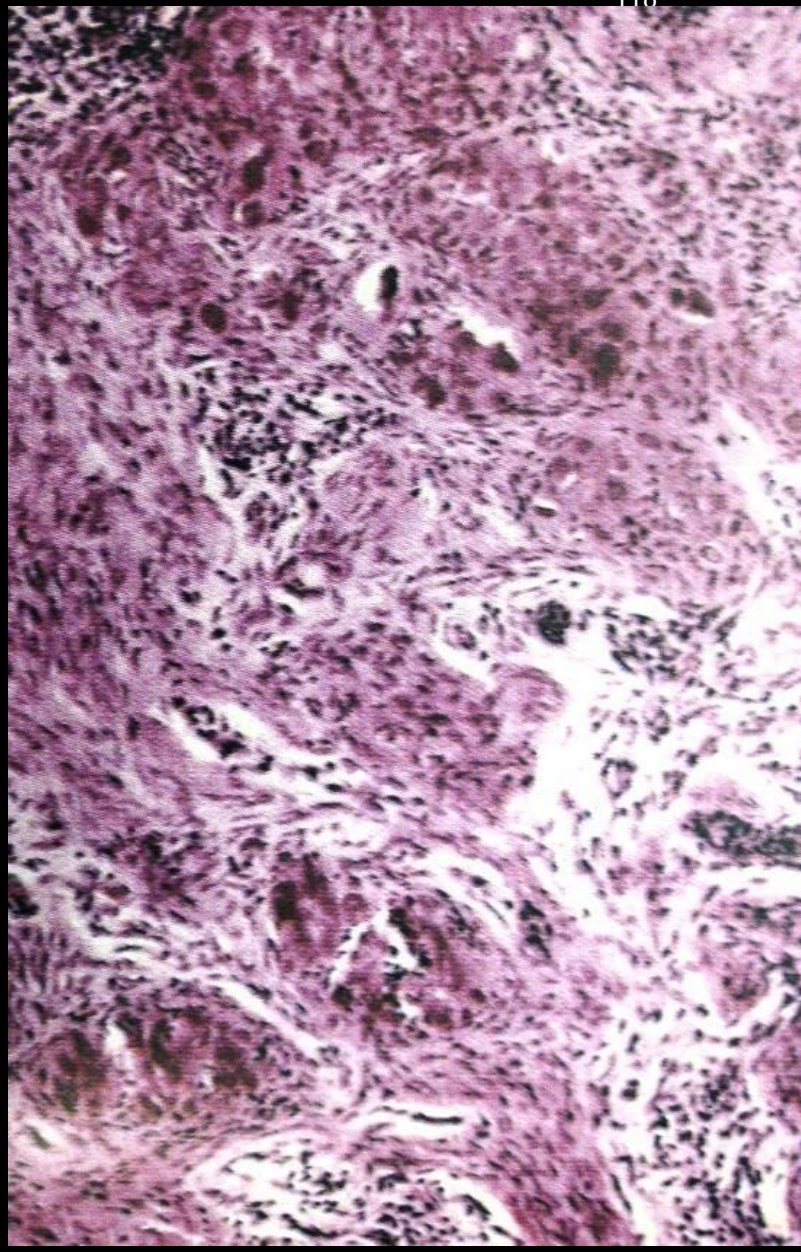
- DELAYED ERUPTION
  - Lack of calcification
    - Lack of density →GHOST LIKE TEETH





HISTOPATHOLOGY

- Pathology
  - Little amount of enamel and dentin
  - More predentin
  - MORE interglobular dentin
  - Follicular tissue around the crown is calcified
    - Enameloid conglomerates



CONCLUSION

- Developmental disorders
  - Variations in structure
- Oral manifestations may be a clue to many serious systemic unknown manifestations
- Less role of histopathology

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