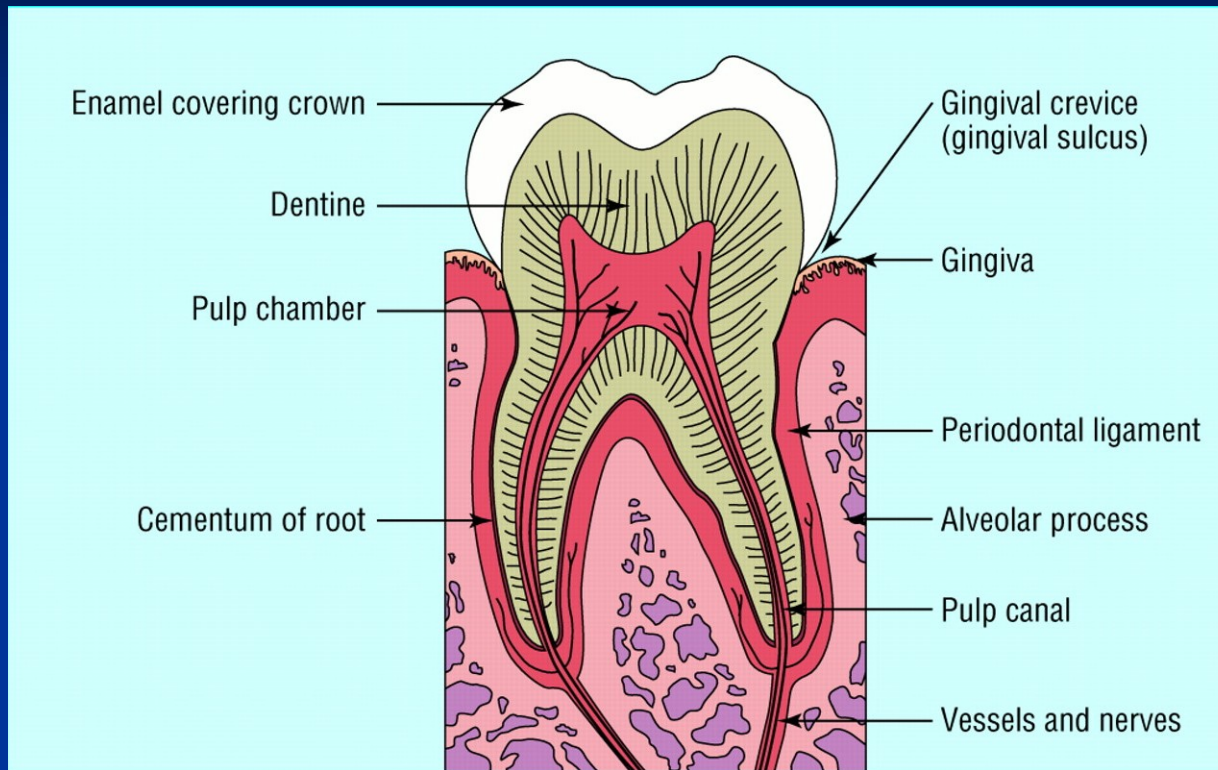


# Disorders of development of teeth and craniofacial anomalies.

Markéta Hermanová

# Disorders of development of teeth.



- **Disturbances in number of teeth**
- **Disturbances in size of teeth**
- **Disturbances in form of teeth**
- **Disturbances in structure of teeth**
- **Craniofacial anomalies**

# Disorders of development of teeth.

- Prenatal
- Postnatal
  
- Inherited
- Aquired

# Disturbances in number of teeth.

- Hypodontia, anodontia, and associated syndromes
- Hypohidrotic ectodermal dysplasia
- Hyperdontia (supernumerary teeth)

■ **Hypodontia:** congenital absence of teeth

- More common in the permanent dentition (2-10 % in populations)
- In primary dentition <1 %; assoc. with the absence of permanent succesional tooth
- Racial and geographic differences
- Symetric/asymmetric
- 3rd molar, permanent maxillary lateral incisors, mandibullary 2nd premolars
- A role of control and regulating genes in the development of teeth
- Assoc. wih other craniofacial anomalies and syndromes

■ **Anodontia:** complete absence one or both dentitions

## ■ Hypohidrotic ectodermal dysplasia

- Congenital absence of ectodermal structures
- X-linked (GR), mutation in EDA gene (signalling molecule), failure of interactions between epithelial and mesenchymal tissues; rarely AR
- Smooth dry skin, scanty hairs, partial or total absence of sweat glands (hyperthermia)
- Severe hypodontia (teeth retarded in eruption, deformed teeth, conical crowns of teeth)
- Female carriers - minimal hypodontia

## ■ Hyperdontia (supernumerary teeth)

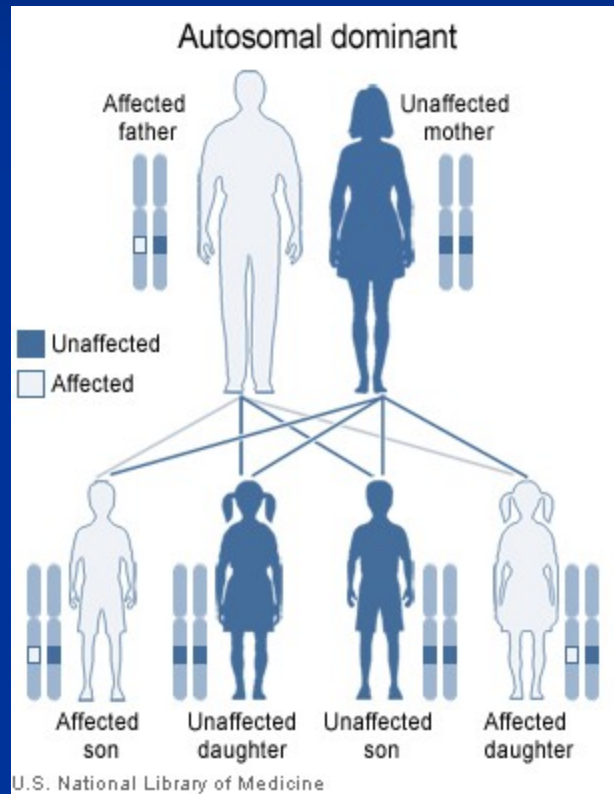
- maxilla (anterior and molar regions)
- assoc. with cleft palate and cleidocranial dysplasia
- F>M
- Unusual in deciduous dentition
- Prevent the eruption, causes malposition, resorption of adjacent teeth, development of dentigerous cysts if unerupted
- *Mesiodens*: between maxillary central incisors
- *Paramolar*: alongside the maxillary molars, usually buccaly placed
- *Distomolar*: distally to a 3rd molar



<b>Syndrom/anomaly</b>	<b>Associated features</b>
<b>Hypodontia</b>	
<b>Cleft lip/palate</b>	Deafness, cranial and skeletal abnormalities
<b>Crouzon syndrome (FGFR gene)</b>	Craniosynostosis, maxillary hypoplasia, hypertelorism
<b>Down syndrome (trisomy 21)</b>	Multiple, e.g. mental retardation, macroglossy, maxillary hypoplasia, anomalies of heart
<b>Hypohidrotic ectoderma dysplasia</b>	Hypotrichosis, hypohidrosis, saddle-nose
<b>Ellis-van Creveld syndrome</b>	Dwarfism, polydactyly, cardiac malformations
<b>Oro-facial digital syndrome</b>	Cleft palate, hypoplasia of nose, digital malformations
<b>Hyperdoncie</b>	
<b>Cleft lip/palate</b>	Deafness, cranial and skeletal abnormalities
<b>Cleidocranial dysplasia (RUNX2 gene)</b>	Aplasia of clavicles, delayed ossification of fontanelles, enlargement of cranium
<b>Gardner syndrome (APC gene)</b>	Osteomas of jaws, skin cysts and fibromas, intestinal polyposis-carcinomas
<b>Sturge-Weber angiomatosis</b>	Venous angiomatosis (also facial and oral), cerebral angiomatosis
<b>Oro-facial digital syndrome</b>	Cleft palate, hypoplasia of nose, digital malformations



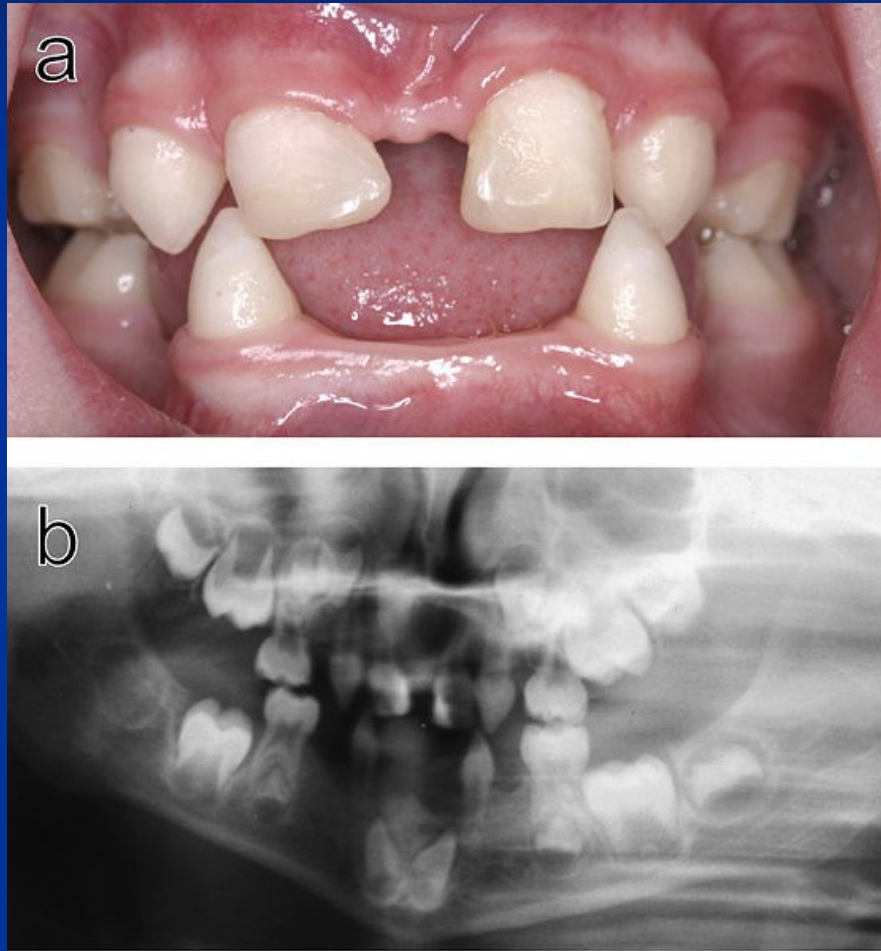
# Gardner syndrome



# Cleidocranial dysplasia.



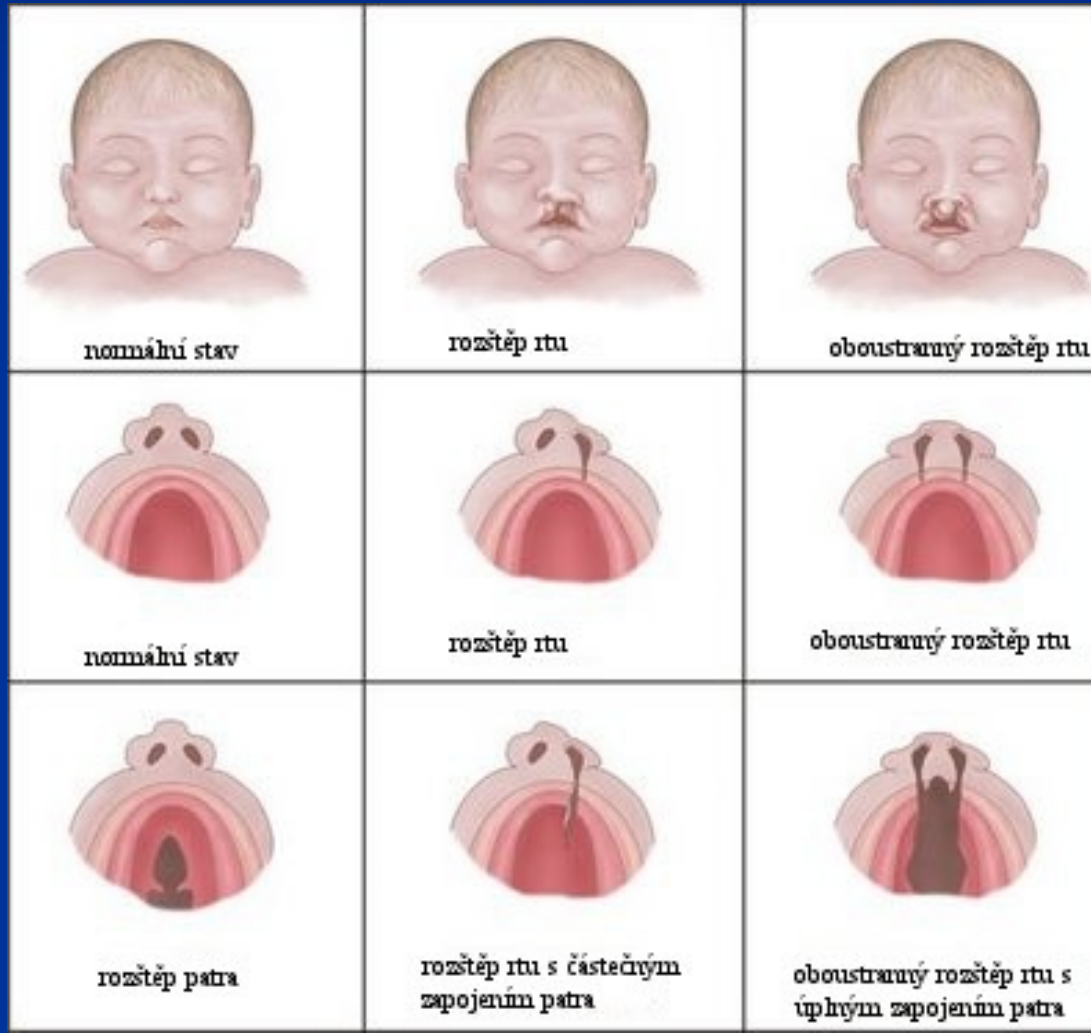
# Hypodontia.



# Orofacial clefts

- In combination with over 300 syndromes
- 70 % non-syndromic
- 1/500-1000 births
- Clefts of the lip and palate (45 %) > clefts of the palate (30 %) > clefts of the lip (25 %)
- Multifactorial causes

- **Cleft lip:** defective fusion of the medial nasal process with maxillary process
- **Cleft palate:** failure of palatal shelves to fuse



Cleft of the lip:

- Unilateral
- Bilateral

Cleft of the lip and palate

- Unilateral
- Bilateral

Cleft of the palate

- **Lateral facial cleft** (isolated or with mandibulofacial dysostosis): lack of fusion of the maxillary and mandibullary processes; uni- or bilateral
- **Oblique facial cleft**  
(from upper lip to the eye, +CP; failure of fusion of the lateral nasal process with the maxillary process or caused by amniotic bands)
- **Median cleft of the upper lip**  
(failure of fusion of the medial nasal processes; in several syndromes, in holoprosencephaly)
- **Median maxillary anterior alveolar clefts**  
(bony defect in the midline of the maxilla between incisors)

# Disturbances in size of teeth

- **Macrodontia**

- **Microdontia**

- **Genetic factors**

(microdontia in Down syndrome, in congenital heart diseases)

- **Environmental factors**

- **May involve the entire dentition**

# Disturbances in form of teeth.

## ■ Dilaceration

- Tooth severely bent along its long axis, trauma
- Maxillary incisor

## ■ Taurodontism

- Pulp chamber higher, with no constriction in amelocemental junction
- Failure of Hertwig's sheath invaginate at the proper horizontal level
- Sporadic or assoc. with Klinefelter and poly-X chromosomes syndromes

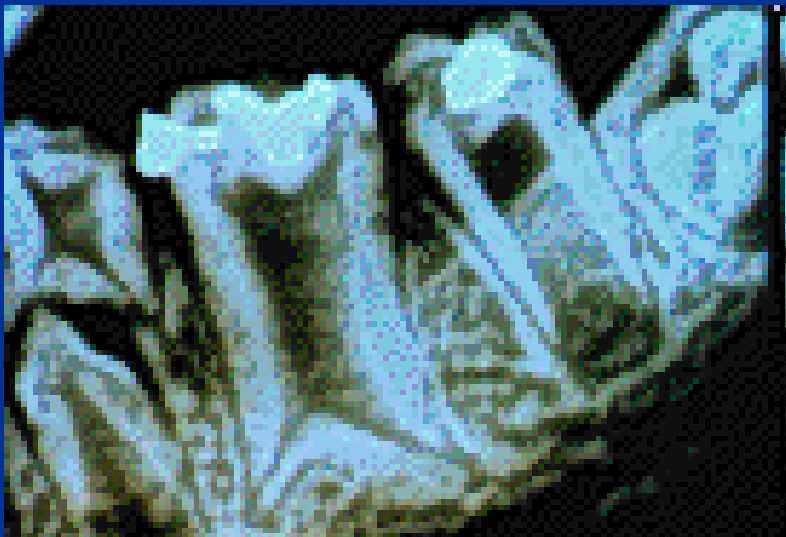
## ■ Double teeth

- Developmental anomaly, teeth joined together (crowns, roots, or both (with/without joining of the pulp)
- More often in primary dentition
- Fusion (the union of two or more separate developin)
- Gemination (incomplete division of teeth)

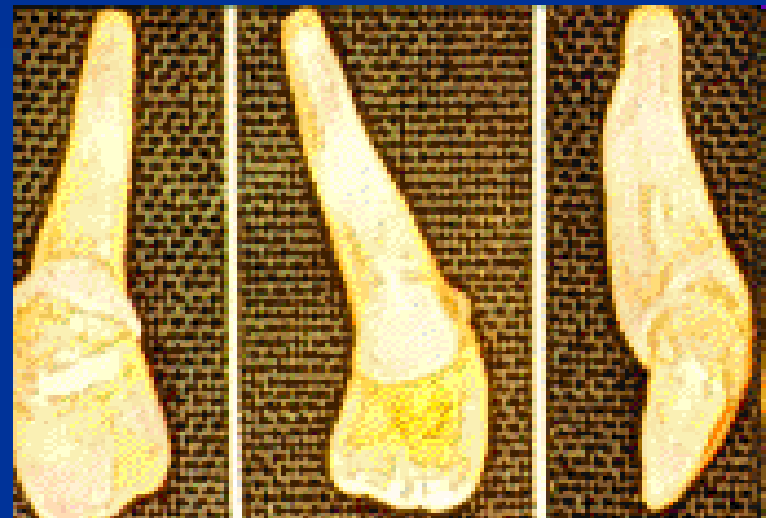
## ■ Concrecence

- Acquired disorder, affects more often permanent dentition
- Teeth united by cementum (anatomically close teeth (2nd and 3rd maxillary molar, hypercementosis in inflammation)

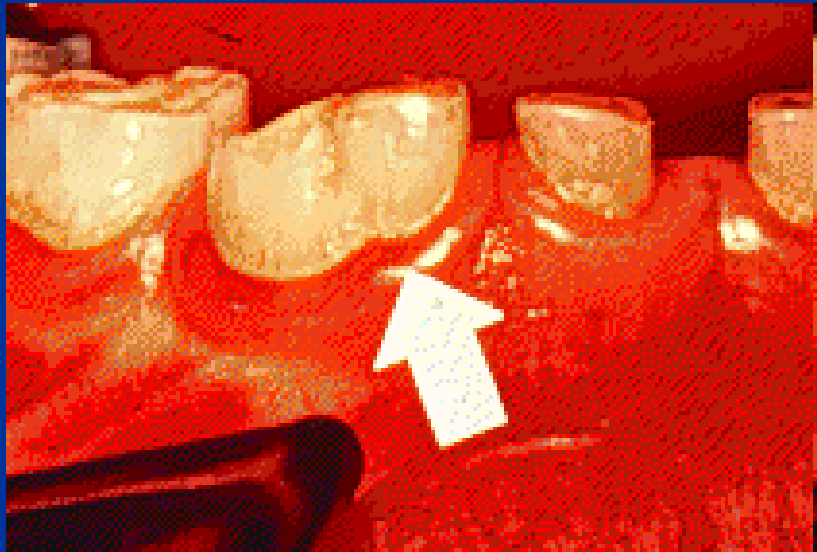




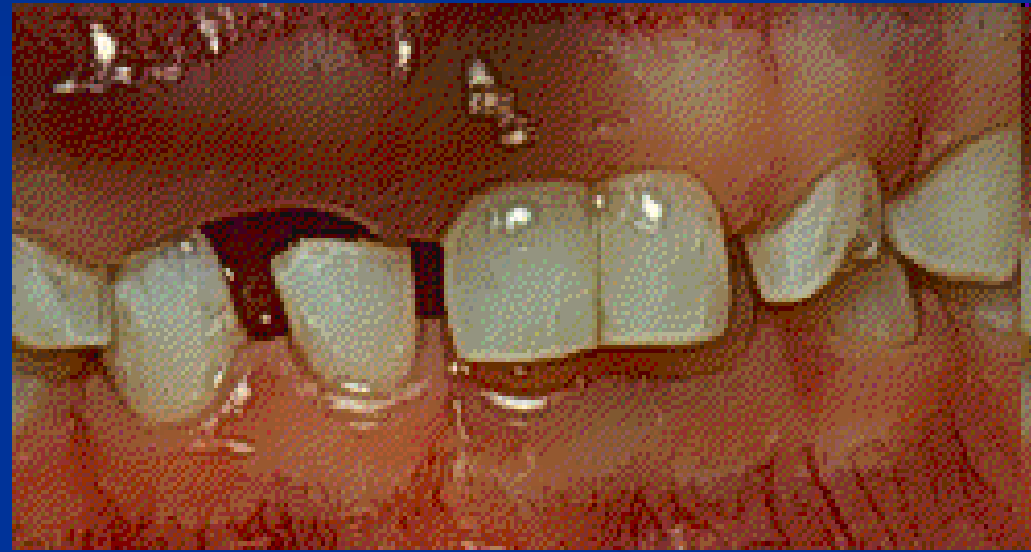
**Taurodontism**



**Dilaceration**



**Fusion**



**Gemination**

# Disturbances in structure of teeth

- Disturbances in structure of enamel
- Disturbances in structure of dentine
- Disturbances in structure of cementum

# Amelogenesis

## Secretory phase

Secretions of enamel matrix proteins by ameloblasts: amelogenin, enamelin, ameloblastin, tuftelin

Enamel matrix proteins – maturation initiation

Crystallites growing mainly in length, little in width or thickness

Amount of matrix produced determines thickness of enamel and crown morphology

## Maturation phase

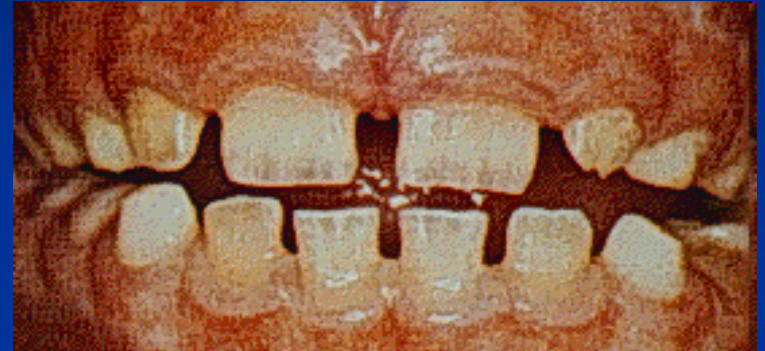
Secretion of matrix protein ceases

Growth in length of crystallites is terminated

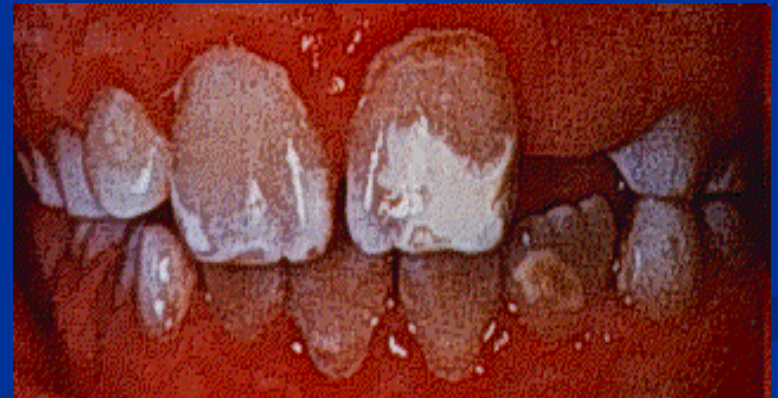
Secretion of proteolytic enzymes and degradation of matrix proteins

Crystallites growing in width and thickness

- Defective matrix production – *enamel hypoplasia*



- Defective maturation/mineralisation – *hypomineralized enamel*



## Local causes of developmental abnormalities of enamel

Infection, trauma, radiotherapy. Idiopathic.

## General causes

### Environmental/systemic causes (chronological dysplasias)

#### Prenatal

Infections: rubeolla, syphilis, ...  
Maternal diseases  
Excess fluoride ions

#### Neonatal

Hemolytic disease of newborn  
Hypocalcaemia  
Premature birth/prolonged labour.

#### Postnatal

Infections (viral exanthemata)  
Heart diseases, endocrinopathies, GIT diseases  
Avitaminosis (D)  
Chemotherapy  
Excess fluoride ions

## Genetic causes

#### Teeth affected

Amelogenesis imperfecta

#### + generalized defects

Ectodermal dysplasia syndromes , Down syndrome

# Genes encoding enamel proteins.

- Amelogenin
- Enamelin
- Ameloblastin
- Tuftelin

# Amelogenesis imperfecta.

## ■ 2 types:

### - hypomineralized/hypomaturation type

(normal tooth morphology when first erupt, soft chalky enamel easily lost, exposing dentine)

### - hypoplastic type

(enamel of normal hardness, variable thickness)

## ■ AD most often; rare XR (amelogenin)



## Local causes of developmental abnormalities of dentine

Trauma, radiotherapy, Turner teeth (due to trauma/infection of primary teeth)

## General causes of developmental abnormalities of dentine

### Dentinogenesis imperfecta

Typ I      assoc. with osteogenesis imperfecta

Typ II      Teeth only affected, AD, both dentitions affected, discoloration (amber like), obliteration of pulp

Typ III      Racial isolate in USA, type II like

### Dentinal dysplasia

Typ I      Radicular (rootless teet)

Typ II      Coronal

### Environmental/systemic causes

Avitaminosis D

Hypophosphatemia

Hypophosphathasia

Juvenile hypoparathyroidism

Other mineral deficiencies, drugs, chemotherapeutics,...

# Turner tooth

- enamel hypoplasia involving a solitary permanent tooth; related to infection in the primary tooth that preceded it or to trauma during odontogenesis.
- enamel discoloration, abnormal coalescence or enamel missing; in severe cases dentine and cementum also affected

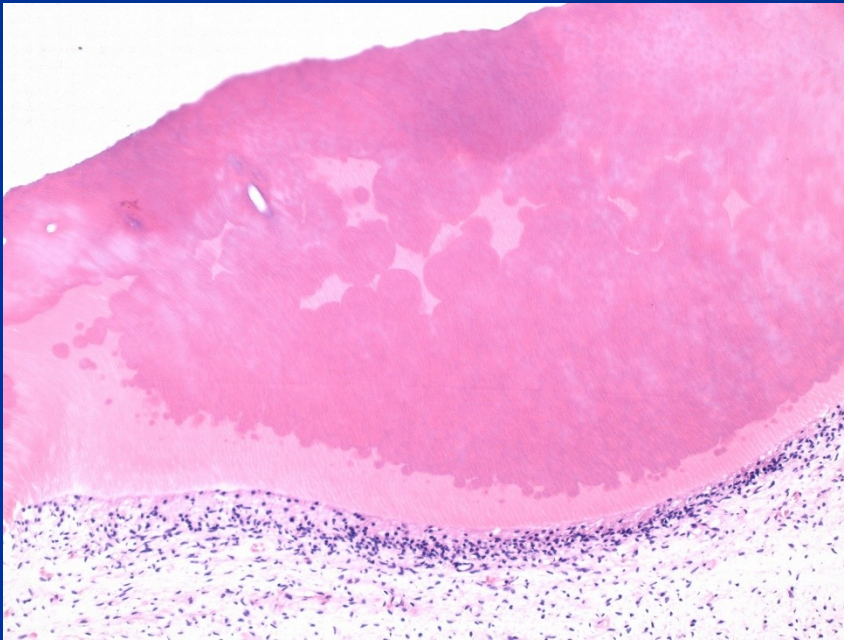


# Regional odontodysplasia („ghost teeth“)

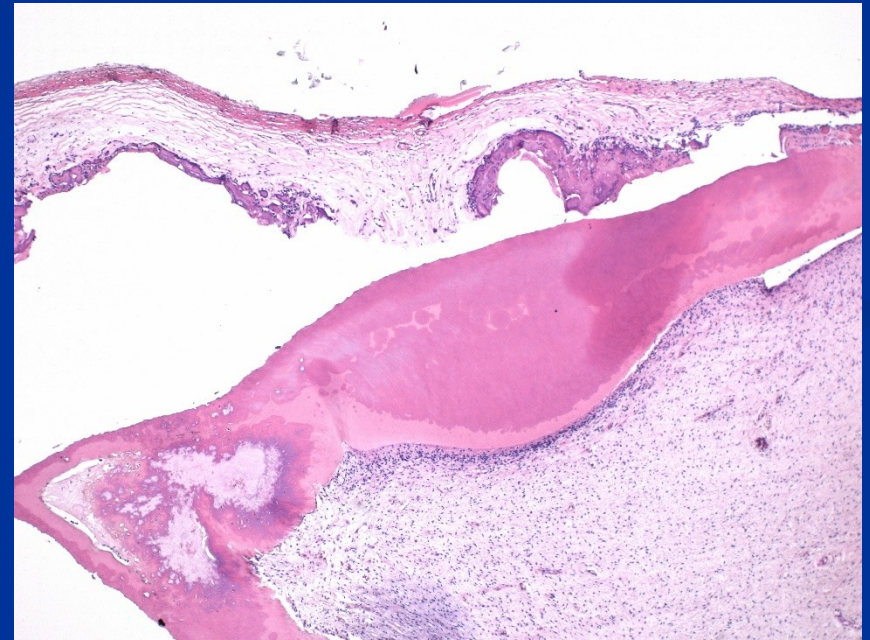
- Unknown etiology
- Abnormalities of enamel, dentin, pulp, dental follicle
- Both dentition affected
- Delayed eruption of abnormally formed tooth
- Reduced radioopacity of the teeth with lost of distinction between enamel and dentine („ghostly“ appearance)



# Regional odontodysplasia („ghost teeth“)

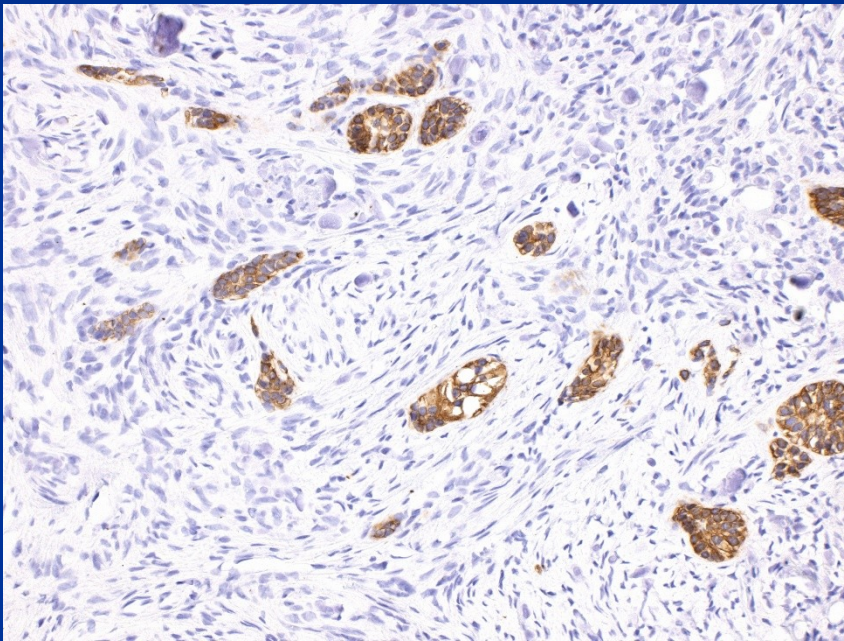


Dentine, mostly atubular, with fields of amorphous dentine with globular formations.

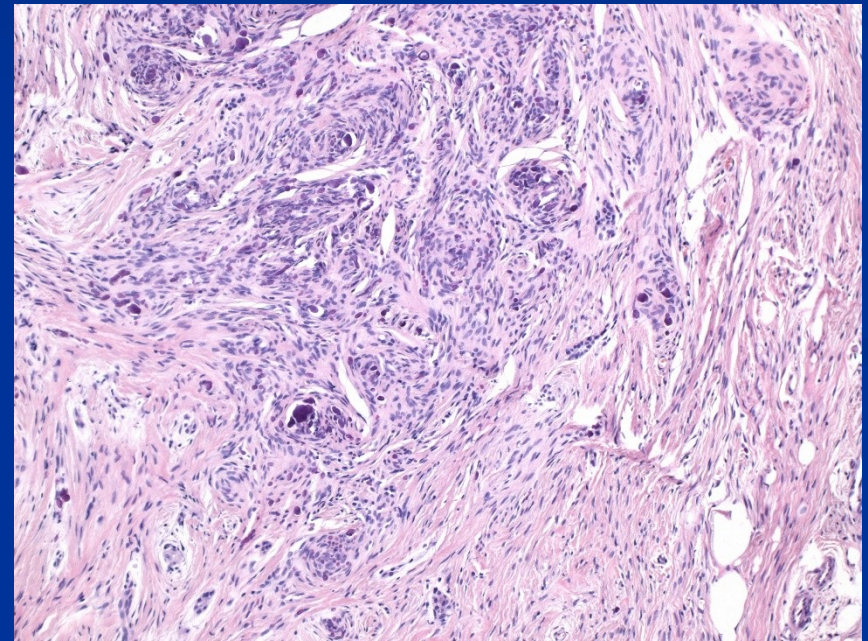


Hypoplastic enamel with globular calcifications, mostly atubular dentine, with clefts.

# Regional odontodysplasia („ghost teeth“)



Follicular tissues of unerupted tooth with remnants of odontogenic epithelium (immunohistochemically with positive expression of cytokeratins – epithelial tissues markers).



Follicular tissues of unerupted tooth with remnants of odontogenic epithelium, fibrous tissues and calcifications of soft tissues.

# Disturbance in structure of cementum.

- Coronal third covered by a narrow layer of acellular (primary) cementum
- apical 2/3 covered by an additional thicker layer of cellular (secondary) cementum
- **Hypercementosis**
  - Idiopathic or known causes
  - Ankylosis, concretion
  - causes: periapical inflammation, mechanic stimulation, functionless/unerupted tooth, Paget's disease of bone
- **Hypocementosis**
  - In hypophosphatasia, in cleidocranial dysplasia,.....

## **Causes of macroglossia**

### **Congenital and hereditary**

**Vascular malformations**

**Hemihyperplasia**

**Cretinism**

**Beckwith-Wiedemann syndrome (omphalocele, visceromegaly, gigantism, hypoglycemia)**

**Down syndrome**

**Mucopolysaccharidoses**

**Neurofibromatosis**

**Multiple endocrine neoplasia, type 2B**

### **Acquired**

**Edentulous patients**

**Amyloidosis**

**Myxedema**

**Acromegaly**

**Angioedema**

**Tumors**

- **Microglossia**

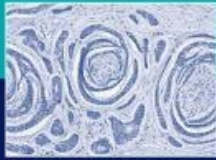
- **Aglossia**

(in oromandibular-limb hypogenesis syndrome)

- **Ancyloglossia (tongue-tie)**

(short, thick lingual frenum)





# Oral Pathology

Fourth Edition

J. V. Soames and J. C. Southam

OXFORD

# Oral & Maxillofacial PATHOLOGY



SECOND EDITION

Neville  
Damm  
Allen  
Bouquot



- Oral pathology textbook not necessary
- Material from lectures obligatory!

