DEVELOPMENTAL DISORDERS

Lecture outline

- Glossary
- Dev Distb of teeth affecting
  - Number
  - Size
  - Shape
  - Position
  - Eruption
  - Structure
- Dev Distb affecting
  - Jaws
  - Lips
  - Palate
  - Oral mucosa
  - Gingiva
  - Tongue
  - Salivary glands
  - Oral Lymphoid tissue

Glossary of terms

1. Malformation
2. Deformation
3. Anomaly
4. Anomalad
5. Syndrome
6. Congenital
7. Hereditary
8. Familial
9. Teratogen
10. Chromosome
11. Gene
12. Locus

Malformation & Deformation

Malformation is a defect due to localized error of morphogenesis resulting in an abnormal shape or structure with interference in function e.g. Cleft lip, Cleft palate.

Deformation is an alteration in shape and/or structure of a previously normally formed part. E.g. Torticollis (wry neck).

Anomaly & Anomalad

Anomaly is a deviation from normal. Same as malformation but without interference in function. E.g. Peg-shaped lateral, Enamel hypoplasia.

Anomalad is a malformation and subsequently derived structural changes. E.g. Pierre-Robin anomalad.

Syndrome

Syndrome is a group of symptoms & signs of a disordered function related to one another by means of same anatomic, physiological or biochemical peculiarity presumably of same aetiology but not consequent of a single localized error in morphogenesis. E.g. Apert, Crouzon, Down, Klinefelter, & Marfan syndromes.

Eagle Syndrome
Developmental Disorders of Teeth

Related definitions

- **Congenital** is a term used for a condition that occurs at birth or noticed at birth. E.g. congenital syphilis.
- **Hereditary** refers to a genetic character transmitted from parent to the offspring. E.g. Hereditary ectodermal dysplasia.
- **Familial** is a term used for disease occurring more frequently in a family than would be expected by chance. E.g. familial fibrous dysplasia, familial hypophosphatia.

Cell & Chromosome

Teraogens - environmental aetiological factors responsible for developmental disorders independent of any known inheritance pattern. E.g. radiations. Chromosome is a linear thread like structure in the nucleus of a cell containing DNA which transmits genetic information.

Gene

Gene is the basic unit of heredity present on a chromosome. It is a sequence of DNA that carries the coded message for production of a particular type of protein polypeptide.

Locus, Allele, Homozygous & Heterozygous

Locus is the position occupied by a gene on a chromosome. Allele is one of the 2 or more different genes containing specific heritable characteristics that occupy loci on paired chromosomes. **Homozygous** means identical alleles (2 dominant or 2 recessive). **Heterozygous** means dissimilar alleles (1 dominant & 1 recessive).

Glossary of terms

- **Penetrance** is the capacity to express abnormal genes clinically in a carrier. Frequency with which a heritable characteristic /trait is manifested by individuals carrying the principle gene or genes conditioning it. It could be reduced penetrance or complete penetrance.
- **Expressivity** is the degree of variation in expression of severity of abnormal genes, given that a gene is penetrant.
- **Mutation** is the change of genetic constitution of an individual.
- **Genotype** is the genetic constitution of an individual.

Phenotype is the expression of the genetic constitution of an individual. Final product of a combination of genetic & environmental influences.

Incidence refers to number of new cases of a disease that develop during a given period per unit of population. Usually expressed as per year per 1,000, 10,000 or 100,000 population.

Prevalence refers to the total number of cases of a disease in the population, living on a given date. Usually expressed as per 1,000, 10,000 or 100,000 population.
Autosomal dominant

Autosomal recessive

X-linked inheritance

Dev Disorders affecting Tooth number
- Anodontia
- Hypodontia
- Oligodontia
- Supernumerary teeth (Hyperdontia)
- Pre-deciduous dentition
- Post-permanent dentition

Anodontia
- True anodontia
  - Assoc syndromes
    - Anhidrotic ectodermal dysplasia
    - Ectrodactyly, Ectodermal dysplasia, Clefting (EEC) syndrome
- Pseudoanodontia
- False anodontia
**Hypodontia & Oligodontia**

- True – evolutionary, hereditary, infections
  - More common in perm dentition
  - More common in girls
  - Mandible : Maxilla =
    - 3rd molars, Lat incisor, mand. 2nd premolar
  - Assoc Syndromes: ED, EEC, Down, Crouzon
- Pseudo
- False

**Supernumerary teeth**

According to location
- Mesiodens
- Paramolars
- Distomolars
- Ectopic

According to shape
- Supplemental
- Conical
- Tuberculate
- Odontome-like

**Mesiodens**

**OPG showing multiple supernumeraries**
### Aetiopathogenesis & Features
- Hyperactivity of dental lamina
- Dichotomy of tooth germs of normal series
- Atavistic trend – 31, 1C, 4PM, 3M
- Genetic
- Incidence: 0.1 – 3.6%
  - > Men
  - > Maxilla
  - > perm dentition
- Assoc conditions:
  - Gardener’s syndrome
  - Cleidocranial dysostosis

### Predeciduous & Postpermanent dentition
- Hornified epithelial structures seen in infants
- 3rd set of dentition
- Multiple retained supernumerary teeth?
- Should be distinguished from premature eruption of deciduous teeth

### Dev Disorders affecting tooth size
- Microdontia
- Macrodontia
- Rhizomicry
  - Dentinogenesis imperfecta
  - Dentine dysplasia
  - Rhizomegaly

### Microdontia
- True generalized
- Relative generalized
- Focal

### Macrodontia
- Focal Macrodontia
- Fusion or Macrodontia?

### Dev Disorders affecting tooth shape
1. Gemination
2. Fusion
3. Concrescence
4. Dilaceration
5. Taurodontism
6. Dens invaginatus
7. Dens evaginatus
8. Talon cusp
9. Enamel pearls
10. Supernumerary cusps
11. Peg-shaped teeth
12. Shovel-shaped incisors
13. Moralization of premolars
Developmental Disorders of Teeth

- Gemination (Schizodontia)
- Fusion (Synodontia)
- Concrescence
- Dilaceration
- Taurodontism

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Vestigiality describes homologous characters of organisms which have seemingly lost all or most of their original function in a species through evolution. These may take various forms such as anatomical structures, behaviors and biochemical pathways. Some of these disappear early in embryonic development, but others are retained in adulthood. Examples include:

- vermiform appendix
- coccyx or tailbone (a remnant of a lost tail)
- the plica semilunaris on the inside corner of the eye (a remnant of the nictitating membrane)
- muscles in the ear
- formation of goose bumps in humans under stress is a vestigial reflex.

**Taurodontism**

**Dens invaginatus**
Dens evaginatus (Leong’s premolar)

Talon cusp

Rubinstein-Taybi Syndrome

Enamel pearls – Classification

Enamel pearls

Shovel-shaped incisors
**Protostylid**

- Extra cusp on buccal side of the lower molar crown
- Seen esp on the 1st or 3rd perm molars or in dec lower 2nd molars.
- may be seen in up to 40% of a population.

**Dev Disorders affecting tooth position**

- Ectopia
- Transmigration
- Transposition
- Rotation
- Inversion
Developmental Disorders of Teeth

**Transposition**

Maxillary Canine 2nd Premolar Transposition

**Rotation of teeth**

**Dev Disorders affecting tooth eruption**
- Premature eruption
- Delayed eruption
- Impacted and embedded teeth
- Premature loss of deciduous teeth
- Delayed shedding of deciduous teeth
- Eruption sequestrum
- Ankylosed (submerged) teeth

**Premature eruption**

Natal teeth

**Delayed eruption**

Single tooth
- Large crowns
- Early loss of dec teeth
- Ectopia
- Eruption cyst
- Thick operculum
- Normal biologic variation

Multiple teeth
- Hypopituitarism
- Hypothyroidism
- Cleido-cranial dysostosis
- Down's syndrome
- Crouzon's syndrome
- Hemifacial atrophy
- Rutherford syndrome

**Premature loss of deciduous teeth**

Generalized
- Papillon-Lefevre syndrome
- Juvenile diabetes
- Cyclic neutropenia
- Hypophosphatasia

Localized
- Caries & its sequelae
- Periodontal disease

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 Delayed shedding of deciduous teeth

- Generalized
  - Heredity
  - Oligodontia / anodontia of permanent teeth
  - Delayed eruption of permanent teeth

- Localized
  - Absence or late development or malpositioned perm tooth germ
  - Periapical infection of deciduous tooth
  - Submerged deciduous teeth

 Ankylosed (submerged) teeth

- Also called Reimpaction

 Eruption sequestrum

- Tiny irregular spicule of bone overlying the crown of an erupting permanent molar found just prior to or immediately following the emergence of tips of cusps through oral mucosa.

 Impacted teeth

 Dev Disorders affecting tooth structure

- Environmental enamel hypoplasia
- Amelogenesis imperfecta
- Dentinogenesis imperfecta
- Dentin dysplasia (Rootless teeth)
- Regional odontodysplasia (Ghost teeth)
- Acementogenesis
Aetiology of Enamel Hypoplasia

1. LOCAL CAUSES
   a) Periapical infection of a deciduous tooth
   b) Trauma to the deciduous tooth
   c) Direct injury to the permanent tooth bud
      • Turner’s Hypoplasia of the underlying permanent tooth

2. GENERAL CAUSES
   a) Hereditary
      • Amelogenesis imperfecta
      • Ectodermal dysplasia
      • EEC syndrome
      • Chondroectodermal dysplasia
      • Ehlers-Danlos syndrome

   b) Neonatal disturb
      • Physical injury at birth
      • Hypoxia at birth
      • Haemolytic dis of new born
      • Premature birth
      • Cerebral palsy + kernicterus
      • Infantile tetany
      • Congenital allergies

   c) Infections
      • Congenital syphilis
      • Exanthamatous fevers: Measles, Scarlet fever, Chicken pox

   d) Nutritional Deficiencies
      • Def of Vit A, B, C, D
      • Def of Minerals Ca, P, Mg

   e) GIT disturb
      • Non-Specific diarrhoea
      • Coeliac syndrome
      • Incorrect feeding correct feeding

   f) Intoxications
      • Fluorosis
      • Tetracycline

   g) Endocrinopathies
      • Hypoparathyroidism
      • Hypothyroidism
      • Hypogonadism

   h) Miscellaneous
      • Calcinosis
      • Nephrotic syndrome
      • Congenital heart disease

Environmental Enamel hypoplasia

Generalized

Focal

Fluorosis: Dean’s Index

<table>
<thead>
<tr>
<th>Score</th>
<th>Type</th>
<th>Features</th>
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</thead>
<tbody>
<tr>
<td>0</td>
<td>Normal</td>
<td>No white patches pits or other abnormalities</td>
</tr>
<tr>
<td>0.5</td>
<td>Questionable</td>
<td>When factors other than fluorosis are suspected to have caused enamel hypoplasia.</td>
</tr>
<tr>
<td>1</td>
<td>Very mild</td>
<td>White spots or flecks on enamel</td>
</tr>
<tr>
<td>2</td>
<td>Mild</td>
<td>White patches on enamel</td>
</tr>
<tr>
<td>3</td>
<td>Moderate</td>
<td>Pitting + brownish staining of enamel</td>
</tr>
<tr>
<td>4</td>
<td>Severe</td>
<td>Corroded enamel surface, with brownish staining and greater wear rate</td>
</tr>
</tbody>
</table>
Fluorosis (Mottled enamel)

Congenital syphilis

Congenital syphilis occurs when an infected woman becomes pregnant or when a pregnant woman becomes infected.

- Foetal transmission
  - Haematogenous spread
  - Direct contact with infected genital lesions (uncommon)
- Only permanent central incisors and 1st molars are affected

Classification of AI (Witkop, 1976)

I. Hypoplastic AI
   A. Pitted, AD
   B. Localized, AD
   C. Localized, AR
   D. Smooth, AD
   E. Smooth, XL-D
   F. Rough, AD
   G. Agenesis, AR

II. Hypomaturation AI
   A. Pigmented, AR
   B. XL-AR
   C. Snow-capped

III. Hypocalcified AI
   A. AD
   B. AR

IV. With others
   A. AI + taurodontism
   B. AI + epilepsy & MR
   C. AI + Nephrocalcinosis

Amelogenesis imperfecta

Type I: Hypoplastic
- Quantitative defect
- Reduced thickness
- Well mineralized

Type II: Hypomaturation
- Qualitative defect
- Easily chipped off
- Yellow to brownish
- Normal thickness
- No contrast between enamel and dentine on radiographs

Type III: Hypocalcified
- Qualitative defect
- Soft & friable enamel
- Dull & lustre-less
- Honey-coloured; stains easily
- Normal thickness

Type IV:
- Hypomaturation-Hypoplastic
- Taurodontism

AI- Hypoplastic type
Dentinogenesis imperfecta

- Definition: Hereditary defective dentine formation with opalescent teeth in absence of systemic disease.
  - Shields Type I: Assoc with Osteogenesis imperfecta
    - Inherited defect in collagen formation
    - More common in primary teeth
    - Amber translucent tooth colour; bulbous crowns with cervical constriction
    - Periapical lesions, obliteration of pulp chambers, root tips
  - Shields Type II: Hereditary Opalescent Dentine
    - Primary and permanent dentition
    - Features same as Type I but no Osteogenesis imperfecta
  - Shields Type III: Brandywine / Maryland type / Shell teeth
    - Bell-shaped crowns
    - Multiple pulp exposures

AI- Hypoplastic type

AI- Hypocalcified type

AI- Hypomaturation type

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Dentinogenesis imperfecta

Dentine dysplasia
- Shields Type I: Radicular dentine dysplasia (Rootless teeth)
- Shields Type II: Coronal dentine dysplasia

Dentine dysplasia – Radicular type
- Normal crown morphology
- multiple periapical radiolucencies
- short roots; pulp chambers absent (obliterated)
- Wide variation in root formation because dentinal disorganization may occur at different stages of tooth development
- Deciduous teeth more severely affected

Dentine dysplasia Type I: Radicular
Dentine dysplasia Type I: Radicular

- Root length normal in both dentitions
- Primary teeth
  - Clinically resemble dentinogenesis imperfecta - Amber coloured
  - Radiographically have similar appearance to Type I
- Permanent teeth
  - Normal colour
  - Pulp chambers enlarged with apical extension-thistle tube - shaped or flame-shaped

Dentine dysplasia Type II: Coronal

- “Thistle tube” pulp chamber
- Thistle tube

Regional odontodysplasia (Ghost teeth)

- Localized, non-hereditary developmental abnormality of teeth with extensive adverse effects on formation of enamel, dentin and pulp — Ghost-like appearance on the radiographs
- Occurs in region or quadrant

Features

- Aetiology unknown - Localized arrest in tooth development due to regional vascular developmental anomaly
- Occurs in both dentitions
- If present in primary dentition, permanent teeth in area usually affected
- More in Maxilla 2.5:1
- Many affected teeth fail to erupt
- Erupted teeth have small irregular yellow-brown crowns
- Short roots, enlarged pulp & shell-like crowns “egg-shell” appearance
- Large diffusely calcified pulp chamber
- Open apical foramina
Regional odontodysplasia (Ghost teeth)

Ghost teeth – “egg shell” appearance

Developmental Disorders of teeth: Recap

- **Number:** Ano~, Hypo~, Oligo~dontia, Supernumerary teeth
- **Size:** Macro~, Micro~dontia
- **Shape:** Gemination, Fusion, Concrescence, Taurodontism, Dilaceration, Dens invaginatus, talon cusp, enamel pearls etc
- **Position:** Ectopic, Transposition, Rotation
- **Eruption:** Premature & delayed, Impaction
- **Structure:** En Hypoplasia, AI, DI, DD, Ghost teeth

THANK YOU!!