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## **Developmental Disturbance of Oral and Para-oral Tissues**

**→ Dr Adel Abdel-Azim**

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# Developmental Disturbances of Face

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1. Treacher Collins syndrome
  2. Pierre Robin syndrome
  3. Crouzon syndrome
  4. Cleft lip and palate
  5. Oblique facial cleft
  6. Transverse facial cleft
  7. Macrostomia
  8. Microstomia
  9. Facial hemihypertrophy
  10. Facial hemihypoplasia
-

# Treacher Collins Syndrome (Franceschetti's Syndrome)

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## Definition:

- Developmental malformation of face often termed **mandibulofacial dysostosis**
  - The severe form of the disease is termed **Franceschetti's syndrome**
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# Treacher Collins Syndrome

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## Etiology and Pathogenesis:

- Hereditary, transmitted as dominant autosomal trait
  - The mutant gene is located on the long arm of chromosome 5 (5q32-33.1) and was termed treacle gene
  - The mutant gene results in:
    - Deficiency of blood supply of the 1<sup>st</sup> arch mesoderm, or
    - Failure of migration of neural crest cells to the area of first arch
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# Treacher Collins Syndrome

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## Clinically:

1. Colobomata of the lower eye lids with atrophy of the medial two thirds of the eyelid.
  2. Antimongoloid slant of the palpebral fissure.
  3. Hypoplasia of the zygomatic bone resulting in flattening appearance of the face.
  4. Hypoplasia of the mandible.
  5. Deformity of the ear pinna.
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# Treacher Collins Syndrome

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## Clinically (continued):

6. Deformity or absence of the external auditory meatus leading to deafmutism.
  7. Abnormal growth of hair in front of the ears.
  8. Flattened frontonasal angle.
  9. Cleft lip and/or palate.
  10. Incomplete forms of the disease may occur.
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# Pierre Robin Syndrome (Sequence)

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- A developmental malformation characterized by mandibular hypoplasia, cleft palate and glossoptosis.
  - Until 1974, the triad was known as Pierre Robin syndrome; however, the term syndrome is now reserved for those errors of morphogenesis with the simultaneous presence of multiple anomalies caused by a single etiology. The term sequence has been introduced to describe any condition that includes a series of anomalies caused by a cascade of events initiated by a single malformation
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# Pierre Robin Syndrome, Pathogenesis

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Two theories exist:

- Hereditary theory, **autosomal recessive** and **X-linked** inheritance have been proposed.
  - Mechanical theory (oligohydramnios theory) assumes that **oligohydramnios** will force the fetus body on the vertex of the head thus allowing the mandible to come in forceful contact with the shoulder and the sternum. This condition will in turn results in failure of the tongue to descend into its normal lower position with consequent clefting of the palate. The presence of a **shoulder impression** on the body of the mandible and the characteristic **U-shaped cleft palate** lend support to this theory.
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# Pierre Robin Syndrome, clinically

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1. Hypoplasia of mandible. Sometimes the mandible is not hypoplastic but there is right angle gonial angle instead of being obtuse. There is also a shoulder impression on the body of the mandible.
  2. Glossoptosis (ptosis in Greek means downward displacement) in which the tongue may fall back to touch the posterior wall of the pharynx producing asphyxia.
  3. Cleft palate.
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# Jacqueline Kennedy Onassis

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Former First Lady

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# Craniosynostosis – Generic Term

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## ■ Links to craniosynostosis:

- <..\Students\DevDistSoft\CRANIOSYNOSTOSIS.doc>
- <..\Students\DevDistSoft\CraniosynostGenTerm.pdf>
- <..\Students\DevDistSoft\Craniosynostosis01.htm>

## ■ Links to Crouzon syndrome:

- <..\Students\DevDistSoft\Crouzon Syndrome01.htm>
- <..\Students\DevDistSoft\Crouzon01.pdf>

## ■ Links to Apert syndrome:

- <..\Students\DevDistSoft\Apert syndrome01.htm>
- <..\Students\DevDistSoft\Apert syndrome02.htm>
- <..\Students\DevDistSoft\Apert01.pdf>

## ■ Links to Pfeiffer syndrome:

- <..\Students\DevDistSoft\Pfeiffer01.pdf>
  - <..\Students\DevDistSoft\Pfeiffer02.pdf>
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# Craniosynostosis – Generic Term

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- Craniosynostosis is the early ossification of one or more of the cranial sutures resulting in abnormal head shape
  - Skull deformity occurs due to retarded skull growth perpendicular to the affected suture associated with increased growth along the same direction that the suture follows
  - Craniosynostosis is classified into primary and secondary
  - Primary craniosynostosis results from abnormal suture biology and is further classified into syndromic and non-syndromic (isolated)
  - Syndromic craniosynostosis occurs as a result of many syndromes, the major ones are: Crouzon syndrome, Apert syndrome, Pfeiffer syndrome and Carpenter syndrome
  - Most of these syndromes show autosomal dominant inheritance
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# Craniosynostosis – Generic Term

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- Most of these syndromes are due to mutations in one of the FGFR genes
  - Non-syndromic (isolated form) is of unknown etiology
  - Secondary craniosynostosis shows normal biology of sutures but there is abnormal internal or external forces resulting in early closure of sutures
  - Secondary craniosynostosis usually results from failure of brain growth and thus results in microrcephaly
  - No medical treatment exists to stop an early ossification of a cranial suture. Infants may require a series of surgical procedures to reduce the intracranial blood pressure or for cosmetic or other functional reasons
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# Crouzon Syndrome

## Craniofacial Dysostosis

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- Definition

A developmental disorder characterized by **craniosynostosis** and **facial malformation**

- Etiology

- Inherited in an autosomal dominant pattern
  - It is suggested that mutation of fibroblast growth factor receptor **FGFR-2** gene could be responsible for Crouzon syndrome
  - Moreover, the mutation in the transmembrane region of **FGFR-3** was detected in this syndrome
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## Crouzon Syndrome - Clinically

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- early ossification of coronal and sagittal sutures
  - Broad face and skull
  - Hypoplastic maxilla
  - Optic nerve atrophy leads to vision impairment because of the intracranial hypertension.
  - Impairment of hearing
  - Malocclusion
  - Short upper lip with possible cleft lip
  - Widely spaced eyes (hypertelorism), shallow orbits and protruding eyeballs
  - Possible presence of crossbite
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# Crouzon Syndrome

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- X-Ray:
    - Digital markings of skull
    - Maxillary hypoplasia
    - Small paranasal sinuses
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# Apert Syndrome

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- Apert's syndrome is a developmental malformation characterized by: craniosynostosis, a cone-shaped calvarium, midface hypoplasia, pharyngeal attenuation, ocular manifestations, and syndactyly of the hands and feet.
  - The prodromal characteristic for the typical craniofacial appearance is early craniosynostosis of the coronal suture, the cranial base, and an agenesis of the sagittal suture.
  - These characteristics predispose the patient to maxillary transverse and sagittal hypoplasia with concomitant dental crowding, a maxillary pseudocleft palate, and a skeletal and dental anterior open bite.
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# Apert Syndrome

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- The oral cavity of Apert patients is also characteristic. The findings include a reduction in the size of the maxilla, particularly in the anteroposterior direction. This reduction may result in tooth crowding. Cleft palate or bifid uvula is found in approximately 75 percent of those affected. Dental anomalies such as impacted teeth, delayed eruption, ectopic eruption, supernumerary teeth, and thick gingiva are also common
  - See also:
    1. <..\Students\DevDistSoft\Apert syndrome01.htm>
    2. <..\Students\DevDistSoft\Apert syndrome02.htm>
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# Noonan Syndrome - Clinically

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- Noonan syndrome is a clinically variable developmental disorder defined by short stature, facial dysmorphism and a wide spectrum of congenital heart defects.
  - The distinctive facial features consist of a broad forehead, hypertelorism, down-slanting palpebral fissures, ptosis, high-arched palate and low-set, posteriorly rotated ears.
  - Cardiovascular abnormalities, primarily pulmonic stenosis and hypertrophic cardiomyopathy, are present in up to 85% of affected individuals.
  - Additional relatively frequent features are multiple skeletal defects (spine and chest), webbed neck and mental retardation.
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# Noonan Syndrome

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## ■ Definition

- Is a genetic syndrome associating a short stature and some other minor anomalies.

## ■ Etiology

- Inherited in an autosomal dominant pattern

## ■ Neoplastic Risk

- Children with Noonan syndrome are predisposed to malignancies, usually juvenile myelomonocytic leukemia (JMML)
  - JMML, formerly termed juvenile chronic myeloid leukemia or chronic myelomonocytic leukemia, is a myeloproliferative/myelodysplastic disorder of childhood characterized by excessive proliferation of immature and mature myelomonocytic cells that originate from a pluripotent stem cell.
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# Nefertiti – The Beautiful

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The right Statue is from Berlin Museum, Germany and the left one is from Fournce Museum, Italy



# Face Development

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[Click This Hyperlink](#)

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# Cleft Lip & Palate - Classification

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## 1. Association with other syndromes

### 1. Syndromic

1. Treacher collins syndrome
2. Pierre robin syndrome
3. Apert's syndrome

### 2. Non-syndromic

## 2. Anatomical classification

1. Prealveolar clefts (lip clefts)
  2. Alveolar clefts
  3. Postalveolar clefts (palatal clefts)
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# Cleft Lip and Palate

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- Definition
  - Pathogenesis - multifactorial
  - Classification
  - Clinically
    - Cleft lip
    - Cleft alveolus
    - Cleft palate
  - Submucous clefts
-

# Facial Hemihypoplasia (hemifacial atrophy)

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## ■ Definition

- A characteristic loss in the soft tissues below the skin, usually on one side of the face, in some severe cases, the underlying bone is also affected.

## ■ Clinically

- Starts in 1st or 2nd decade of life
  - Left side more commonly affected
  - Affected side may be hyperpigmented
  - Hollowing of cheek and the orbit
  - Jaw bones and roots of teeth on affected side may exhibit delayed development and retarded tooth eruption
-

# Facial Hemihypoplasia (hemifacial atrophy)

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- Radiographically
    - Deficient root development or root resorption
  - Etiology
    - Peripheral nerve dysfunction, trauma or infection of the growth centers, and genetic causes have been proposed
  - Prognosis
    - The condition progresses slowly for a few years and remains stable thereafter
-

# Developmental Disturbances Of Jaws

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1. Agnathia
  2. Micrognathia (mandibular dysostosis)
  3. Macrognathia
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# Condylar Hyperplasia

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- Is the condition created by excessive growth of one of the condyles
  - Occurs usually in young adults
  - The cause is unknown but the following is postulated:
    - Local circulatory disturbances
    - Endocrine disturbances
    - Trauma
  - Clinically, there is facial asymmetry, prognathism, cross bite or open bite
  - Histologically, during the active phase, proliferation of the condylar cartilage is noted, after which the condyle shows normal histologic appearance
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# Developmental Disturbance of Palate

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- Cleft palate
- Torus palatinus

# Developmental Disturbances of Lips

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1. Cleft Lip
  2. Double Lip
  3. Congenital lip pits and fistulae
  4. Chelitis glandularis
  5. Chelitis granulomatosa
  6. Milkerson-Rosenthal syndrome
  7. Xeroderma pigmentosa
  8. Peutz-Jegher's Syndrome
-

# Double Lip

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- Is the condition characterized by presence of a fold of tissue on the mucosal side of the lip
  - The condition usually affects the upper lip more often than the lower lip
  - The condition may be congenital or acquired later on life as in case of Ascher syndrome
  - Congenital cases may result from malformation of the labial sulcus
  - Treatment by surgical excision if necessary
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# Orofacial Granulomatosis

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## Definition:

- Orofacial granulomatosis comprises a group of lesions characterized by orofacial swellings associated with non-caseating and non-infective granulomas.
- These lesions include:
  1. Cheilitis granulomatosa
  2. Melkersson Rosenthal syndrome
  3. Sarcoidosis
  4. Oral lesions of Crohn's disease

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For more details, see infections of the oral cavity ([Orofacial Granulomatosis](#))

# Xeroderma Pigmentosa

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- A condition in which there is increased sensitivity of the skin to the ultraviolet rays of sun.
  - Hereditary, transmitted as a recessive autosomal trait. The inherited defect seems to be in the DNA repair mechanism.
  - Exposure to sun results in erythema of the skin associated with hyperpigmentation resembling freckles.
  - This is followed by atrophy of the skin and appearance of multiple papules, which may turn, into multiple squamous cell carcinomas or malignant melanoma.
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# Xeroderma Pigmentosa

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## ■ Histologically:

- Hyperkeratosis.
  - Atrophy of the prickle cell layer.
  - Atrophy of some rete pegs and elongation of others.
  - Melanin pigmentation of the basal cell layer.
-

## Peutz Jegher's Syndrome

(Multiple intestinal polyposis syndrome with melanin pigmentation)

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### Definition:

- This is a hereditary disease characterized by presence of multiple polyps in the small intestine associated with circum-oral, circum-nasal, circum-ocular melanin pigmentation. Melanin pigmentation may also be present in the oral mucosa and on the dorsum of hands and feet.

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For more details, see (<F:\Students\Malignant\Cancer Prone Diseases.pdf>)

# Peutz Jegher's Syndrome

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## Etiology:

- Transmitted as a simple Mendelian dominant autosomal trait.

## Clinically:

- Frequent episodes of abdominal pain and signs of minor obstruction.
  - In the past, the intestinal polyps in Peutz-Jegher's syndrome were considered to be non-precancerous in contrast to the polyps of the colon noted in Gardner's syndrome
  - However, recent studies indicate that the condition is precancerous with increased risk of carcinomas affecting small intestine, stomach, colon and esophagus.
-

# Lip Fissure

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- Lip fissure is not a developmental disease
  - It is mentioned here because of its relevance to lip disorders
  - The exact etiology is unknown
  - However, it is postulated that the disease results from chronic exposure to sun or due to nutritional deficiency
  - The condition may run in a chronic course
  - Treatment is by lip protection or surgical excision of the fissure in resistant or chronic cases
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# Developmental Disturbance of Oral Mucosa

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1. Fordyce's granules
  2. Leukoedema
  3. White spongy nevus
  4. Hereditary benign intraepithelial dyskeratosis
  5. Darier's disease (keratosis follicularis)
  6. Epidermolysis bullosa
  7. Hereditary hemorrhagic telangiectasia
  8. Dyskeratosis congenita
  9. Fanconi's syndrome
  10. Pachyonychia Congenita
  11. Plumer-Vinson Syndrome
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# Leukoedema

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- Is a bilateral, diffuse, translucent white thickening of the oral mucosa
  - The thickening is soft, pliable and can not be wiped off
  - The thickening disappears upon stretching and reestablishing itself when released
  - The buccal mucosa is the most common site
  - It is a variation of normal, present in 90% of blacks and variable numbers of whites.
  - The cause of this normal variation is not precisely known
  - Histologically, there is acanthosis with intercellular edema of the spinous cell layer.
  - Treatment is unnecessary but reassurance may be required.
-

# White Spongy Nevus (Familial White Folded Gingivostomatitis, Cannon's Disease)

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- An autosomal dominant condition of the oral cavity characterized by soft, white or opalescent, thickened and corrugated folds of mucous membrane.
  - The condition resembles leukoplakia, but the lesion tends to be soft and spongy upon palpation.
  - The disorder may appear at birth, in infancy, or in childhood, and reaches full severity at puberty when it should remain stationary.
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# White Spongy Nevus

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- The lesion usually affects the buccal mucosa; however in rare cases the entire oral mucosa may be affected.
  - A nevus by definition is a developmental malformation of skin or mucous membrane arising from cells native to skin or mucous membrane and simulating a neoplasm.
  - Nevi in general are of three types, keratotic (white spongy nevus), melanotic and vascular (e.g. hemangioma and lymphangioma).
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# White Spongy Nevus

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- Nevi, in general, are considered to be one of the types of hamartomas.
  - The term hamartoma by definition is a developmental malformation appearing in a tumor like condition due to presence of normal cells in normal sites but in an exaggerated amount.
  - Treatment is unnecessary but reassurance may be required.
-

# White Spongy Nevus - Histologically

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- Acanthosis (increased thickness of prickle cell layer)
  - Inter and intra-cellular edema of the prickle cell layer giving the characteristic picture of basket weave appearance
  - Some degree of hyperkeratosis
  - Chronic inflammatory cell infiltration of the connective tissue
-

## Differences between leukoedema and white spongy nevus

Leukoedema	White spongy nevus
Etiology is unknown, may represent a normal variation of oral mucosa	Etiology is hereditary, autosomal dominant
Just thickening of the surface epithelium (acanthosis only)	Thickening with hyperkeratosis (acanthosis with hyperkeratosis)
Soft and pliable	More tough than leukoedema but less hard than leukoplakia
Disappear upon pressure	Does not disappear upon pressure
Smooth surface	Rough surface
No chronic inflammatory cells in the CT	There is chronic inflammatory cell infiltration in the CT

# Darier's Disease (keratosis follicularis)

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- Is a hereditary skin disease characterized by hyperkeratotic papules usually in seborrheic regions and various nail abnormalities.
  - Abnormal cell-cell adhesion and abnormal epidermal keratinization are the primary features of DD
  - Electron microscopy reveals loss of desmosomes and breakdown of desmosome-keratin intermediate filament attachment.
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# Darier's Disease (keratosis follicularis)

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- Mutations in the **ATP2A2** gene cause functional disruptions of the adhesion between keratinocytes and affect cellular differentiation in the epidermis
  - DD most commonly begins in the first and second decades of life.
  - The lesions may first appear as skin-colored papules, which soon become yellowish brown, greasy, warty papules. These lesions are especially common in the seborrheic areas, such as the forehead, the scalp margin, the scalp, the nasolabial folds, the ears, the chest, and the back.
  - Involvement of the hands is very common (about 95%).
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# Darier's Disease (keratosis follicularis)

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- Mucosal lesions are detected in about 15% of patients, and they appear as white papules with a central depression.
  - These lesions are most commonly found in the mouth, and they give the mucosa a **sandpaper texture (cobble stone appearance)** At times, these lesions may affect the salivary glands and cause obstruction
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# Darier's Disease (Histologically)

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- Hyperkeratosis
  - Acanthosis
  - Benign dyskeratosis. Two types of dyskeratotic cells are present: corps ronds and grains. Corps ronds are predominantly located in the prickle and granular cell layer. Corps ronds are characterized by an irregular eccentric and sometimes pyknotic nucleus, a clear perinuclear halo, and a brightly eosinophilic cytoplasm. Grains are mostly located in the stratum corneum, and they consist of oval cells with elongated cigar-shaped nuclei and abundant keratohyalin granules
  - Acantholysis (loss of attachment between epithelial cells). Acantholysis results in the formation of suprabasilar clefts which may contain many acantholytic cells
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# Epidermolysis Bullosa

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- A group of inherited chronic skin diseases in which large bullae and erosions result from slight mechanical trauma affecting skin and oral mucosa. Tooth brushing increases bullae formation. Dystrophy of nails was described in some cases
  - There are more than one mode of inheritance for EB with many gene mutations, thus EB has a highly variable molecular etiology and represents a collection of different diseases. The majority of cases are **autosomal dominant**, but **recessive inheritance** was also described for some cases
-

# Epidermolysis Bullosa

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- Onset of EB is at birth or shortly after.
  - Dental manifestations include:
    - Pitted or thin enamel which may lack prismatic structure
    - Smooth amelodentinal junction
    - Delayed or failure of eruption of teeth.
    - Vigorous tooth brushing can elicit bullae formation
  - EB is a genetic disease and no drugs are known to correct the underlying molecular defects.
-

# Papillon-Lefèvre Syndrome

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## Definition

An autosomal recessive disorder characterized by severe destructive periodontal disease affecting both the primary and permanent dentitions and hyperkeratosis of the palms of the hands and soles of the feet (palmoplantar keratosis).

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# Papillon-Lefèvre Syndrome

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## Clinically

- First and fifth years of life
  - No gender predilection
  - Premature shedding of both deciduous and permanent teeth
  - Early development of aggressive periodontitis
  - Hyperplastic and hemorrhagic gingivitis
  - Progressive destruction of the alveolar bone
  - Palmoplantar hyperkeratosis; skin of the knees, elbows, and over the joints of the hands and feet can also be affected
  - Mobility and migration of teeth
  - Painful mastication
-

# Papillon-Lefèvre Syndrome

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## ■ X-ray

- teeth appear to float in the soft tissue.

## ■ Histologically

- Increased vascularity of the connective tissue
- Mixed inflammatory cellular infiltrate

## ■ Treatment

- Retinoids for skin lesions
  - Periodontal therapy and antibiotics
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# Hereditary Hemorrhagic Telangiectasia

## "Rendu-Osler-Weber Disease"

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- Autosomal dominant disease of blood vessels.
  - Onset may be delayed until adult life
  - Pinhead or spider-like telangiectases of the mouth, skin and sometimes of viscera
  - Severe epistaxis often early
  - Sometimes oral bleeding from lips or tongue
  - Haemostatic function is normal
  - Little risks with extractions
  - Laser therapy is adopted for treatment
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# Dyskeratosis Congenita

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X-linked recessive disease which occurs only in males and characterized by the triad:

1. Premalignant leukoplakia of the oral mucosa.
  2. Dystrophy of nails.
  3. Pigmentation, atrophy and telangiectasia of the skin.
-

# Fanconi's Syndrome

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- A condition similar to dyskeratosis congenita, in addition the patient suffers from hypersplenism and anemia. Oral and anal lesions may progress into carcinomas.
  - Fanconi's syndrome:
    1. Dyskeratosis congenita
    2. Hypersplenism (splenomegaly)
    3. Anemia
-

# Pachyonychia Congenita

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- Pachyonychia congenita (PC) is a rare form of hereditary palmoplantar keratoderma (PPK).
  - Müller made the first documented observation in 1904
  - The next reports were published in 1905 by Wilson
  - In 1906 detailed report was made by Jadassohn and Lewandowsky
  - In the dermatologic literature, PC is better known as Jadassohn-Lewandowsky syndrome
-

# Pachyonychia Congenita

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- PC results from mutations in the genes encoding epidermal keratinocyte keratins, specifically K6a, K6b, K16, and K17
  - These genes are located on chromosomes 16 and 17
  - In most cases, an autosomal dominant mode of inheritance is described
  - However, autosomal recessive inheritance is also mentioned in the literature
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# Pachyonychia Congenita

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- Oral Leukokeratosis (white patches), not precancerous as in leukoplakia
- Hyperkeratosis on the palms and soles
- Thickening of nails
- Natal teeth

Oral lesions of pachyonychia congenita should be differentially diagnosed from natal candidiasis, while those of pachyonychia tarda should be differentially diagnosed from oral leukoplakia

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# Plumer-Vinson Syndrome

## *(Paterson-Kelly Syndrome)*

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- Iron deficiency anemia
  - Atrophy of the mucosa of the oral cavity, pharynx and esophagus
  - Atrophy of tongue papillae with glossitis
  - **Dysphagia** due to atrophy causing burning sensation of the esophagus associated with degeneration of the esophageal muscles
  - Atrophy of the epithelium is thought to be due to iron depletion
  - The disease is a risk factor for oral and pharyngeal cancer
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# Developmental Disturbance of Tongue

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1. Aglosia
  2. Microglossia
  3. Macroglossia
  4. Ankyloglossia
  5. Lingual Varicosities
  6. Prominent Circumvallate Papillae
  7. Lingual Tonsils
  8. Cleft tongue
  9. Fissured tongue
  10. Geographic tongue
  11. Hairy tongue
  12. Black tongue
  13. Furred tongue
  14. Median Rhomboidal Glossitis
  15. Thyroglossal tract Cyst
  16. Lingual thyroid nodule
-

# Aglossia

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- Very rare
  - Usually associated with severe fatal syndromes
  - Tuberculum impar may be present as a small nodule in the floor of the mouth
-

# Microglossia

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- Means small tongue
  - No clinical significance
-

# Macroglossia

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- Large tongue
  - Etiology:
    - Congenital: Mongolism, cretinism, hemangioma and lymphangioma
    - Acquired: Amyloidosis, myxedema, inflammation and tumors
-

# Macroglossia

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- Complications:
    - Spacing of teeth
    - Noisy breathing
    - Drooling of saliva
    - Difficulty in eating
    - Lispings speech
    - Crenated lateral border
    - Constant protruding results in ulceration, infection
    - Airway obstruction in severe case
-

# Macroglossia

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- Treatment:
    - Depends on cause & severity
    - Mild cases no treatment, speech therapy may be necessary
    - Severe cases – Reduction glossectomy
-

# Ankyloglossia

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1- Superior

2- Inferior

A- Complete

B- Partial

Treatment: Frenectomy, postponed till 6 month and should not be delayed to avoid speech problems

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# Lingual Tonsils

(Prominent Foliate Papillae, Foliate Papillitis)

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## Etiology

Trauma from teeth

Trauma from dentures

Reactive lymph node reaction

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# Black Tongue

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- Black tongue without elongation of filiform papillae
  - Due to drugs as iron therapy
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# Furred Tongue

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- A coat of debris and desquamated epithelial cells
  - Due to severe fevers
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# Lingual Thyroid Nodule

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- Diagnosis is best established by thyroid scan using iodine isotopes
  - (CT) and (MRI) can be helpful in delineating the size and extent of the lesion.
  - Biopsy is often avoided because of the risk of hemorrhage.
  - In some cases, incisional biopsy may be needed to confirm the diagnosis or to rule out malignant changes.
  - No treatment except periodic follow-up for asymptomatic patients. If symptomatic, suppressive therapy with supplemental thyroid hormone can reduce the size of the lesion. If hormone therapy does not eliminate symptoms, surgical removal or ablation with radioactive iodine-131 can be performed. If the mass is surgically excised, autotransplantation to another body site can be attempted to maintain functional thyroid tissue and to prevent hypothyroidism.
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# Developmental Disturbances of Gingiva

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1. Fibromatosis gingivae (Elephantiasis gingivae, Hereditary gingival fibromatosis)
  2. Gingival cyst of adult
  3. Gingival cyst of newborn (Bohn's nodules)
  4. Epstein's pearls ?????
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# Fibromatosis Gingivae (Elephantiasis Gingivae)

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- Definition:
    - Diffuse fibrous enlargement of the gingiva.
    - Frequently associated with the syndrome known as hypertrichosis.
    - The condition is inherited as an autosomal dominant trait.
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# Fibromatosis Gingivae (Elephantiasis Gingivae)

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## ■ Etiology

- Autosomal dominant
  - may be a part of a number of other rare syndromes e.g.:
    - Hypertrichosis
    - Laband syndrome
    - Rutherford syndrome
-

# Fibromatosis Gingivae (Elephantiasis Gingivae)

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- Clinically:
    - The gingiva starts to enlarge in infancy or even as late as the ninth year.
    - The enlargement usually affects both jaws; however, it is greater in the maxillary than in the mandibular gingiva.
    - The enlargement is diffuse, firm, smooth or nodulated with no apparent signs of inflammation.
    - The condition may interfere with the eruption of teeth and can be regarded as a potential cause of partial or total anodontia.
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# Fibromatosis Gingivae (Elephantiasis Gingivae)

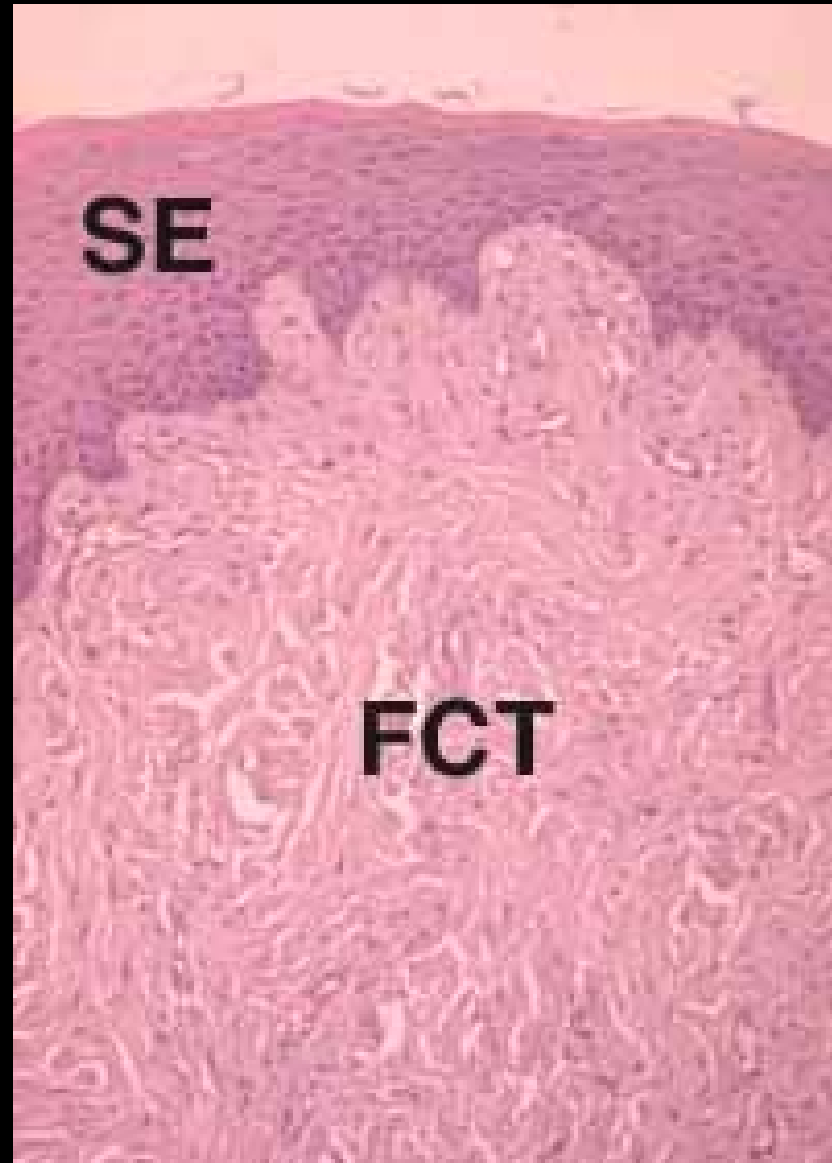
---

- Differential Diagnosis:
    - Leukemic gingival hyperplasia
    - Granulomatous gingivitis
    - Plasma cell gingivitis
-

# Fibromatosis Gingivae (Elephantiasis Gingivae)

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- dense or moderately dense, rather vascular, bland collagenic connective tissue with scattered chronic inflammatory cells
- extreme elongation of rete processes



# Epstein's Pearls

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- Epstein's pearls are small whitish swellings found on the soft tissues of the midpalatine raphe.
  - Epstein's pearls arise due to cystic degeneration of the non-odontogenic epithelium entrapped at the line of fusion of the two palatine processes.
  - Although, Epstein's pearls are not gingival disease, they are mentioned here because they are frequently confused with Bohn's nodules.
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# Retrocuspid Papilla

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- A sessile nodule on the gingival margin of the lingual aspect of the mandibular cuspids (canines).
  - Etiology: Developmental
  - Clinically:
    - 2-4 mm sessile nodule of mandibular alveolar mucosa
    - located on gingival margin lingual to mandibular cuspids
    - more common in children
    - may be unilateral or bilateral
  - Histologically: A mass of fibrous connective tissue covered with normal epithelium.
  - Treatment: Not necessary
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# Developmental Diseases of Salivary Glands

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- Aplasia
- Atresia
- Aberrancy
  - Latent bone cyst (Stafne's bone cyst)

# Multisystem Disorders Affecting Oral And Para-oral Tissues

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1. Cleido-Cranial dysostosis
  2. Gardener's syndrome (Polyposis Coli)
  3. Ehlers-Danlos syndrome (floppy joint)
  4. Epidermolysis bullosa
  5. Hypophosphatasia
  6. Early onset idiopathic hypoparathyroidism
  7. Mongolism (Down's syndrome, Trisomy 21)
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# Cleidocranial Dysplasia - Definition

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- Is a developmental bone disease characterized by defective formation of the clavicles with other dental and cranial abnormalities
  - Was initially thought to involve only membranous bone but now is known also to affect endochondral ossification and to represent a generalized disorder of skeletal structures.
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# Cleidocranial Dysplasia - Etiology

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- Autosomal dominant
  - 40% of cases appear to represent spontaneous gene mutation
  - The defect was mapped to the CBFA1 gene of chromosome 6p21
  - This gene normally guides osteoblastic differentiation and bone formation
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# Cleidocranial Dysplasia - Clinically

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- Partial or complete absence of the clavicles
  - Short stature
  - Large head with pronounced frontal and parietal bossing
  - Ocular hypertelorism
  - Depressed nasal bridge
  - Delayed closure of fontanelles with presence of many wormian bones
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## Cleidocranial Dysplasia – Oral Manifestations

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- Delayed shedding of deciduous teeth
  - Delayed eruption of permanent teeth
  - many unerupted supernumerary or supplemental teeth
  - Many of the unerupted teeth have hooked roots.
  - Many teeth may show different degrees of enamel hypoplasia.
  - Some teeth will assume a conical shape.
  - Multiple dentigerous cysts
  - Gemination may be observed.
  - The maxilla is narrow and v-shaped with a high arched palate.
  - Absence of cellular cementum is usually found
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# Gardener Syndrome's (Polyposis Coli)

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- A developmental hereditary disease characterized by presence of multiple polyps in the large intestine particularly the colon area.
  - Autosomal dominant inheritance was established in many cases.
  - Caused by mutations of the tumor suppressor APC gene
  - Unlike Peutz-Jegher's syndrome, the polyps of Gardener's syndrome show definite tendency toward malignant transformation.
  - Begins usually in late childhood; polyps increase in numbers, causing symptoms of chronic colitis, and carcinoma of the colon almost invariably develops in untreated cases.
  - Also characterized by presence of multiple osteomata and fibromata affecting the skull, jaw bones and sometimes the oral mucosa.
  - There are also multiple supernumerary and supplemental teeth, many of them remain impacted.
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# Achondroplasia

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A child with achondroplasia showing short limbs, a relatively large head, thoracic kyphosis, a sharply angled upper lumbar lordosis, and protrusion of the abdomen.



# Trisomy 21 (Down syndrome)

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## Mongolism

- most frequent - 1:700 births; parents have normal karyotype
  - maternal age has a strong influence: <20 y. 1:1550 live births, >45 y. 1:25 live births
  - most frequently is abnormality in ovum (ovum is under long-time influence of environment)
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# Trisomy 21 - Clinical symptoms

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- mental retardation
  - Mongoloid slant of the eyes
  - congenital heart defects
  - neck skin folds
  - skeletal muscle hypotonia
  - increased risk of acute leukemias
  - mortality 40% until 10Y (cardiac complications)
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# Trisomy 21 – Oral Manifestations

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1. Macroglossia
  2. Macrogathia
  3. Microdontia
  4. Hypodontia
  5. Fissured tongue
  6. High arched palate
  7. Enamel hypoplasia
  8. Excessive salivation
  9. Decreased incidence of dental caries
  10. Increased incidence of periodontal disease
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# Non-disjunction in Down's Syndrome

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