CRANIOFACIAL DYSOSTOSIS (CROUZON SYNDROME)

Dept.of Oral Pathology & Microbiology

Learning Objectives

At the end of the lecture student should be able to describe

- Clinical features, oral manifestations,

  radiographic features,& surgical management of

  Craniofacial Dysostosis, Treacher Collins Syndrome,&

  Down syndrome

Introduction

- Craniosynostosis syndromes constitute a group of conditions, each characterized by premature craniosynostosis occurring in association with a variety of other abnormalities
- Most common of Craniosynostosis syndromes without syndactyly is **Crouzon's Disease**

- Described in 1912 by Crouzon: as a variation of Craniofacial Dysostosis
- Caused by premature obliteration & ossification of two or more sutures, most often coronal and sagital

Clinical features:

- Due to early synostosis of sutures

Facial deformity observed at birth

- Coronal and sagittal sutures obliterated, fontannels remain open
- Lateral and anteroposterior flattening of acrocranium observed: growth only in vertical axis
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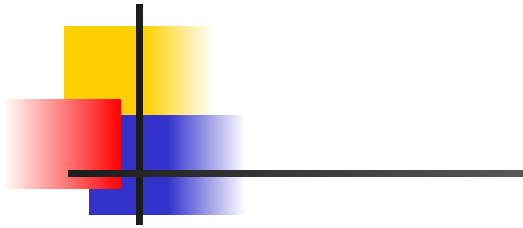
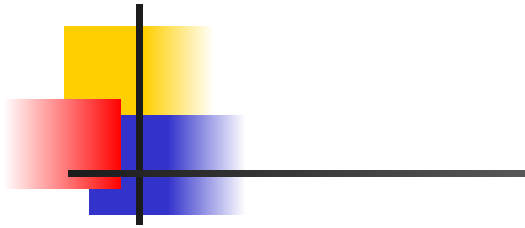
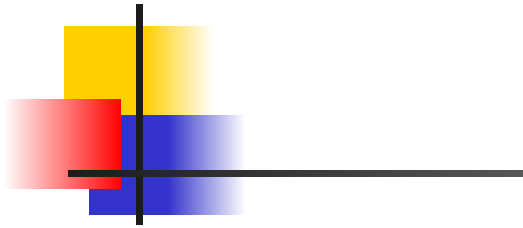


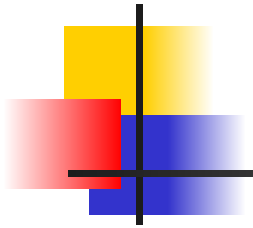
- Anteroposterior diameter smaller than transverse diameter: forehead is high & wide. Wide face & hypoplastic maxilla produce psuedoprognathism
- Deviation of nasal septum, narrowed ant nares, wide beaked nose



- Dysplasia of skeleton is caused by malformations of the mesenchyme & ectoderm
- Inherited as autosomal dominant pattern
- Mutation of FGFR- 2 and 3 gene could be responsible for this syndrome



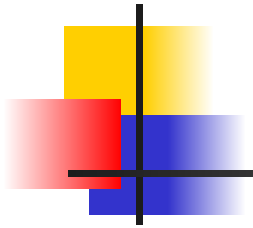
- Hypertelorism, divergent squint, eyelid seem antimongoloid, upper eyelid mimicking **“frog face”**
- Upper lip shortened, sometimes cleaved
- Progressing optic nerve atrophy due to intracranial hypertension leads to vision impairment



- Hearing impairment: middle ear disorders
- Malocclusion
- Short stature, dark skin
- Headache, convulsions, mental retardation

Radiographic features:

- Skull, spine, hand radiographs are necessary to confirm diagnosis
- Skull X ray:
- obliterated sutures (mostly sagittal, coronal)
- Shallow eye sockets (exopthalmous clinically)
- Shortened ant cranial fossa
- Underdeveloped lateral nasal sinuses

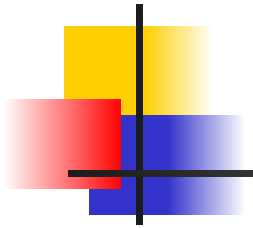


- Tympanic mem fixed obliquely
- Narrowed ext auditory canals, small pyramids with sclerosis

Treatment & Prognosis:

- Neurosurgery
- Plastic surgery of face: very effective cosmetic results
- Patient may lead normal life after treatment

TREACHER COLLINS SYNDROME (MANDIBULO-FACIAL DYSOSTOSIS)

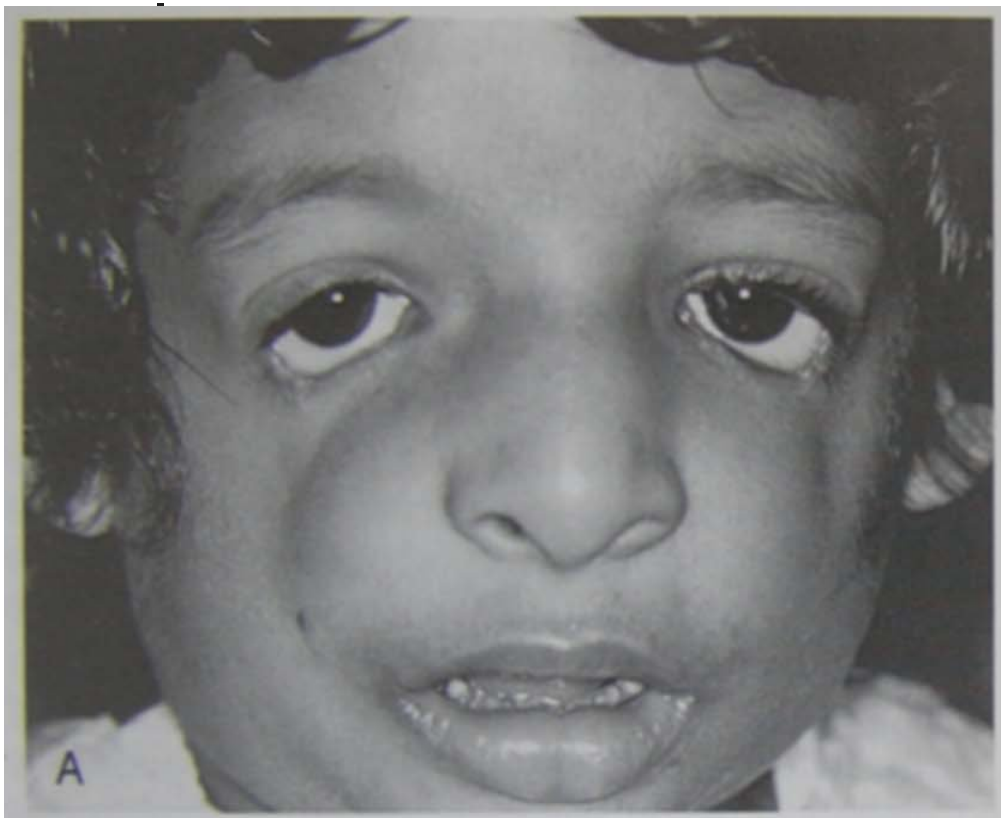


- TCS is primarily affects structures developing from 1st branchial arch, but also involves 2nd arch to a minor degree
- TCS is transmitted by an autosomal dominant mode of inheritance
- Incidence : between 0.5 – 10 cases in 10,000 births

Clinical features:

- Various degree of **hypoplasia** of max, mand, zygomatic process of temporal bone, external & middle ear
- Notched or colobomas of the lower eyelid,
- Lower eye-lashes are absent
- Malformation of external ear

TREACHER COLLINS SYNDROME



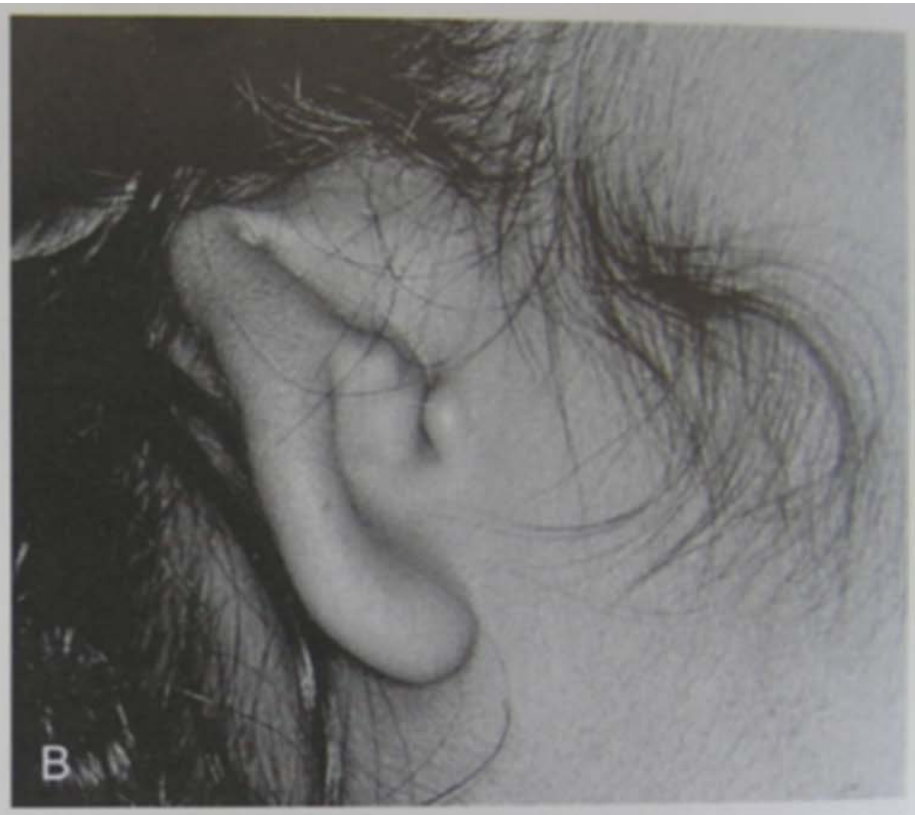


Figure 15–23. *A,* Treacher Collins syndrome. Note the characteristic facial appearance, including downward sloping of the palpebral fissures and colobomas of the lower eyelids. *B,* Microtia, or underdeveloped ear, and a narrow extension of hair over the preauricular region, known as a "hair lick," are common in patients with Treacher Collins syndrome.

- Hair growth is *Tongue-shaped* in the region of pre-auricular area (known as “Hair-lick”)
- Macrostomia, high palate, cleft palate & malocclusion of teeth
- Facial clefts & skeletal deformities are seen in few individuals
- Characteristic appearance are described as being **“Bird like” or “Fish-like”**

Radiography:

- Underdeveloped or complete agenesis of malar bones and also mandible are seen
- Clefting may be seen
- Paranasal sinuses are grossly underdeveloped

DOWN SYNDROME (Trisomy 21 syndrome; Mongolism)

•DS is a disease associated with subnormal mentality in which an extremely wide variety
of anomalies & functional disorders may occur (cranial & facial deformities)

Etiology:

- Many factors like --- advanced maternal age, & uterine & placental abnormalities, have been regarded as causes of the disease.
- **Recent concept: chromosomal abnormality**

Three forms of DS:

1. One in which there is a typical trisomy 21 with 47 chromosomes (95% cases)
2. Another is a translocation type, in which there appears to be 46 chromosomes, although the extra-chromosomal material of no 21 is translocated to another chromosome (3% cases)
3. Another results from chromosomal mosaicism (2% cases)

Clinical features:

•Flat face, large anterior fontanel, open sutures, small slanting eyes with epicanthal folds,
eye defects (refractive errors, nystagmus, cataracts etc..);
open mouth with frequent prognathism, sexual underdevelopment, cardiac abnormalities & hypermobility of the joints,
Congenital heart diseases is commonly seen.

Oral manifestations:

- Hypoplasia of maxilla
- Macroglossia with protrusion of tongue, fissured tongue, open mouth, frequent mouth breathing causes drying & cracking of lips, palatal width is decreased & bifid uvula & cleft palate are seen
- Teeth : delayed eruption of permanent dentition, dental caries, microdontia, enamel hypocalcification , severe PDL destruction

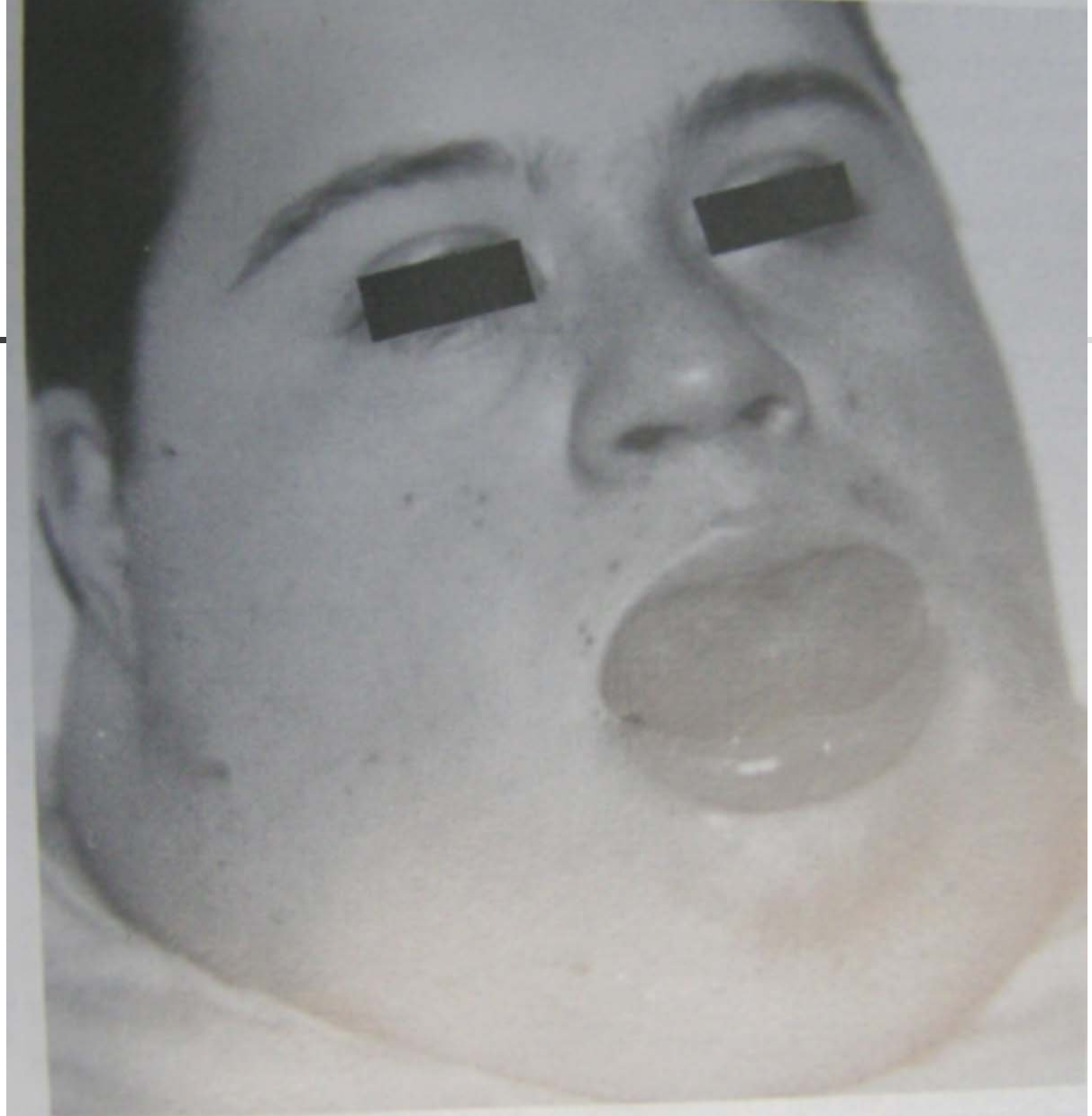


FIGURE 1–18. Macroglossia. Large tongue in a patient with Down syndrome. (Courtesy of Dr. Sanford Fenton.)

Summary

- Clinical features, oral manifestations, radiographic features,& surgical management of Craniofacial Dysostosis, Treacher Collins Syndrome,& Down syndrome

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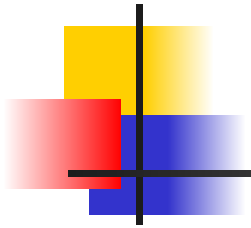

Thank You