Cranial facial Syndromes

- Crouzon/ apert/ Pfeiffer
- Treacher Collin syndrome
- Pierre Robin Syndrome
- Stickler syndrome
- Goldenhar syndrome

1st branchial arch

- ✓ Precursor of maxilla and mandible
- ✓ Muscles/ nerves of mastication (V)
- ✓ Ear formation

- ✓ 1st branchial arch defects
- Crouzon syndrome/ apert syndrome
- Treacher Collins syndrome

2nd Branchial arch

- ✓ Ear formation
- ✓ Muscles/nerves of facial expression

- ✓ 1st + 2nd Branchial arch defects
- Goldenhar Syndrome

TREACHER COLLINS SYNDROME

- ✓ '*absent cheek bone*'

- ✓ Similar to Crouzon/apert syndrome (no exophthalmos)
- ✓ AD inheritance
- ✓ Intelligence normal
- ✓ +/- facial nerve affected

General

- Looks well

Around Bedside

- Hearing aids/glasses

Dysmorphic features

Original Notes prepared by Dr Elizabeth Stockley

- PROMINENT FEATURES

Head

- +/- brachcephaly

Eyes

- Down-slanting palpebral fissures
- Drooping of the lateral lower eyelid
- Squints
- Coloboma/ aplasia of lower lid lashes
- +/- hypertelorism

Ears

- Low set / malformed/absent ears/ preauricular sinuses
- Hearing problems

Face

- Micrognathia
- Hypoplastic maxilla

Mouth

- Lip/ Cleft palate

Neck

- +/- tracheostomy
- Scar

Limbs

- Fusion of radius + ulnar

To complete my examination

- Plot height/weight/ head circumference
- Cranial nerve examination (VII)
- Respiratory/ ENT examination – OSA/airway problems (tracheostomy)
- CVS - VSD
- Formal hearing assessment

GOLDENHAUR SYNDROME

- ✓ Mode of inheritance not known
- ✓ 1st and 2nd Branchial arch defects

General

- Well

Around bedside

- Hearing aids/glasses

Dysmorphic features

- PROMINENT FEATURES: facial asymmetry + hemi-facial microsomia

Head

- Frontal bossing

Eyes

- Epibulbar dermoid
- Coloboma

Ears

- Low set ears/ +/- abnormal position (angle of mandible)
- Malformation/abnormal outer ear
- Hearing problems

Nose

- Poorly developed nostrils

Face

- Asymmetry of the face +/- hemi facial microsomia

Mouth

- Lip/cleft palate

Neck

- Short neck (Fused verbebrae)

- ✓ Associated with renal and cardiac abnormalities
- ✓ Associated with vertebral anomalies (synostosis, spina bifida, hemi-vertebrae)
- ✓ +/- Mental retardation

Original Notes prepared by Dr Elizabeth Stockley

To complete my examination

- Plot head circumference/ height/ weight
- CVS/ renal examination
- Formal hearing assessment

Causes of Micrognathia

1. Treacher Collin syndrome
2. Craniosynostosis
3. Pierre Robin Syndrome
    - High arched palate/ cleft palate
    - Micrognathia
    - Posterior displacement of the tongue
    - Respiratory obstruction (position of tongue/ small mandible)
4. Stickler Syndrome
    - Pierre Robin Syndrome
    - Severe myopia + cataract
    - Deafness
5. Cornelia de Lange syndrome (?)
    - Brushy eye brow/ low hair line/ hypertrichosis on the limbs/trunk
    - Low set ears
    - Micrognathia
    - Limb defects: micromelia (small hands/feet)/ phocomelia
    - CVS: ASD, VSD
    - Mental retardation
6. Russell Silver syndrome
7. Noonan/turner's syndrome

Eye (Coloboma/dermoids)

1. Treacher Collin Syndrome (AD)
    - Coloboma
2. Goldenhar syndrome (?)
    - Coloboma
    - Epibulbar dermoids
3. CHARGE (?)
    - Coloboma (heart defect, choanal atresia, atresia of growth/mental retardation, genitourinary + ear abnormalities
4. Wardenburg (AD)
    - Heterchromia iridis
    - Pigmented changes in skin + hair (white forelock)
    - Sensorineural deafness