Craniofacial Surgery and Syndromes

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Talk Overview

- Craniofacial Disorders
  - Focus on basic facts and possible areas for examination
  - Guides to further reading
- Exam Tips

Craniofacial Patients

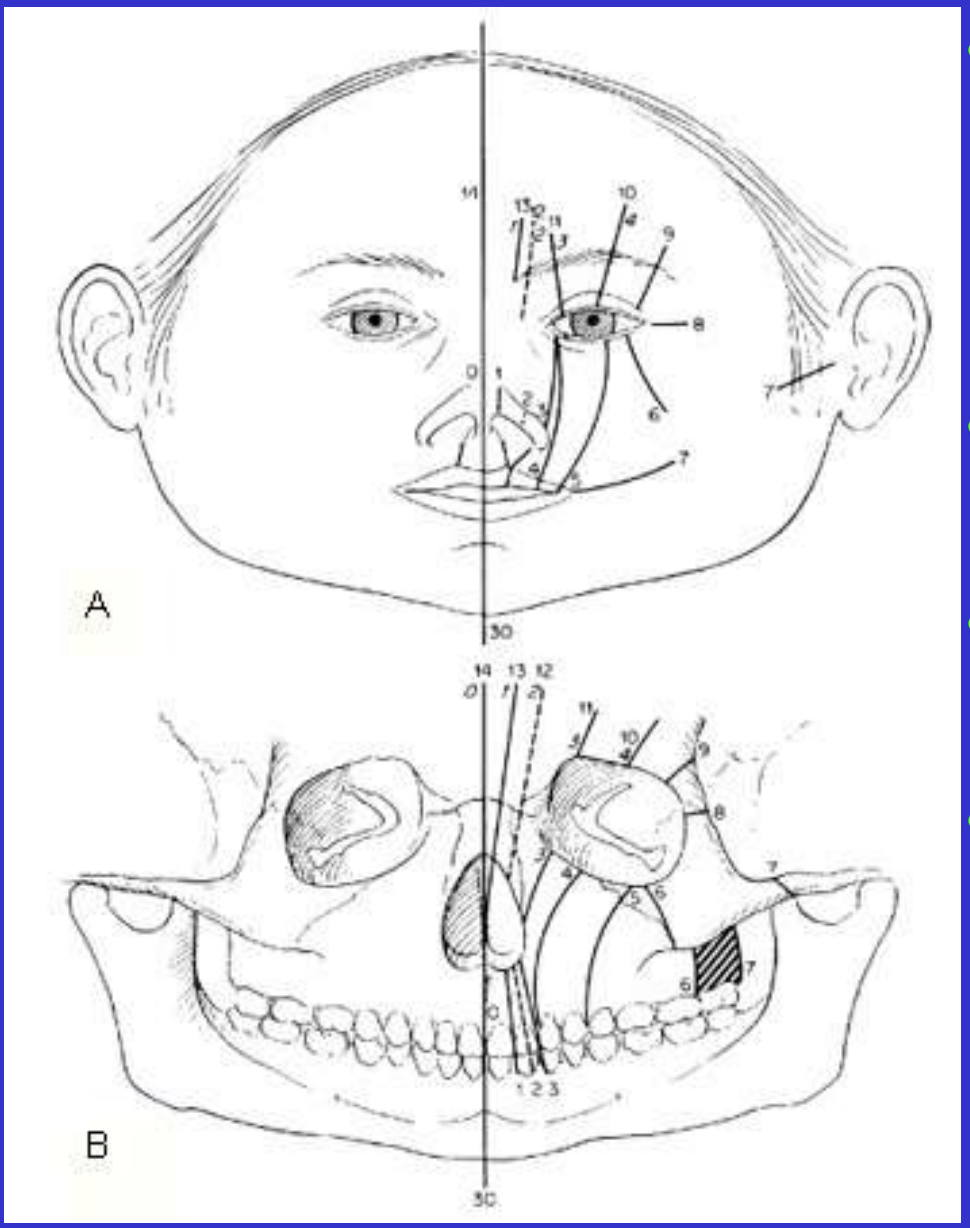
- Functional versus cosmetic issues
- Multidisciplinary approach
- Patients/ parents involved in decisions and timing of interventions

Craniofacial Disorders

(1981 Committee on Nomenclature and Classification of Craniofacial Anomalies of the American Cleft Palate Association)

I. Facial Clefts/ encephalocoeles and dysotoses
II. Atrophy/ hypoplasia
III. Neoplasia/ hyperplasia
IV. Craniosynostosis
V. Unclassified

I. Facial Clefts



- Tessier described classification (left) in 1976 (Van der Meulen alternative classification)
- Clefts of bone and soft tissue may not co-exist
- May cause tissue deficiency or tissue excess
- Often facial/cranial clefts add up to 14 (hairline pointer)

I. Facial Clefts - causes

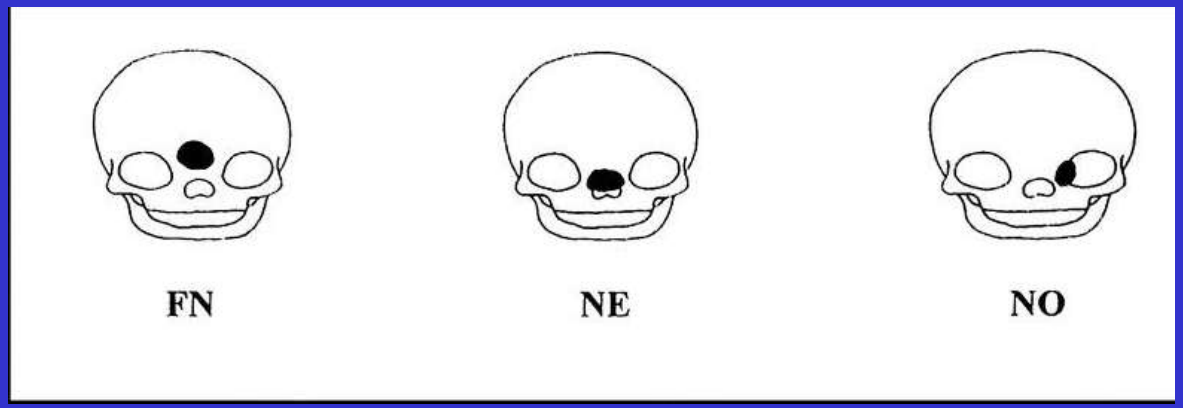
1. Classic Theory (Dursy and His)
   - Failure of fusion
2. Mesodermal Penetration Theory (Pohlman, Veau, Stark and Saunders)
   - Facial development is one of mesenchymal penetration of bilaminar ectodermal membrane
3. Other theories/factors
   - Amniotic bands
   - Environmental cleftogens
     1. Radiation
     2. Infections – eg toxo, rubella and CMV
     3. Maternal idiosyncracies – eg DM, phenyketonuria
     4. Chemicals – Vit deficiencies, Vit A excess, smoking

I. Encephalocoeles

- Protrusion of part of the cranial contents through a defect in the skull
- May contain
  - » Meninges (meningocoele)
  - » Meninges/ brain (meningoencephalocoele)
  - » Meninges/ brain/ ventricle (meningoencephalocystocoele)
- Classified as per position on skull
  - Basal
  - Convexity
  - Sincipital
    - Frontoethmoidal
      - » Nasofrontal
      - » Nasoethmoidal
      - » Naso-orbital
    - Interfrontal
    - Associated with clefts

- Antenatal diagnosis based on u/s or AFP levels
- DDX frontal midline masses
  - – Encephalocoeles
  - – Teratomas
  - – Gliomas
  - – Dermoids

  (ie you need a CT +/- MRI)



I. Encephalocoeles

- Pathogenesis of frontoethmoidal encephalocoeles
  - Diverticula of dura projects through fonticulus nasofrontalis; may become adherent to skin
  - Diverticular normally regresses and bone closes at foramen caecum anterior to crista galli
  - With encephalocoeles, diverticulum doesn't regress
- Causes (unknown but...)
  - Racial/ genetic/ environmental and paternal factors
- Epidemiology
  - 1:5000 live births
  - Western Europe/ Australia/ Nth America/ Japan – mainly occipital
  - Russia/ SE Asia – mainly frontoethmoidal

I. Facial dysotoses

- Hemifacial microsomia
- Treacher Collins Syndrome
- Nager Syndrome
- Binder Syndrome
- Pierre Robin Sequence

I. Facial Dysostoses - Hemifacial microsomia

- “Craniofacial microsomia”; “First and second branchial arch syndrome”; “Tessier 7 cleft”; “lateral facial dysostosis”
- 1:4000 live births
- Bilateral around 10%
- Cause ? Haematoma of embryonic stapedial artery
- Debate about whether disease is progressive

Hemifacial Microsomia – Classification OMENS-Plus

- Orbit (>75% have N orbit)
  1. AbN size
  2. AbN position
  3. AbN size & position
- Mandibular (Mulliken/Kaban modification of Pruzansky)
  - I. Mild hypoplasia of ramus
  - II. A. Small condyle/ramus with functioning TMJ
  - II. B. Small condyle/ramus with non-functioning TMJ
  - III. Absent ramus
- Ear (Meurman)
  1. Hypoplasia/cupping
  2. Absent EAC and variable conchal hypoplasia
  3. AbN lobule with absent auricle
- Nerve
  1. Upper CNVII involved
  2. Lower CNVII involved
  3. All CNVII involved
- Soft Tissue
  1. Mild
  2. Moderate
  3. Severe

Hemifacial Microsomia – Classification OMENS-Plus

- Plus! (>35%)
  - >20% have skeletal anomalies
  - <10% have anomalies in other systems (CNS, renal, CVS etc)

I. Facial Dysostoses - Goldenhar Syndrome

- “Oculoauriculovertebral dysplasia”
- Variant of HFM
- Epibulbar dermoids and vertebral abN (make sure c-spine is OK before GA)

I. Facial Dysostoses – Treacher Collins Syndrome

- Variably expressed symmetrical bilateral mandibulofacial dysostosis
- Tessier 6, 7 & 8
- 1:25,000-50,000 autosomal dominant (TCOF1 gene encoding Treacle protein on Ch5)
- ? Related to AbN Vit A metabolism
- Associated with advanced paternal age
- Very narrow airways at birth – OSA & neonatal death. May need trachy or mandibular DO.

I. Facial Dysostoses – Treacher Collins Syndrome

- Downsloping palpebral fissures
- Coloboma of outer portion lower lid
- Absent eyelashes medial 1/3 lower lid
- Hypoplasia facial bones esp malar and mandible. Class II and AOB.
- Macrostomia, high arch palate, malocclusion
- Pre auricular tags and sinuses
- Abnormal hair growth around ears

I. Facial Dysostoses – Nager Syndrome

- "Acrofacial dysostosis"
- Autosomal recessive
- Similar to Treacher Collins BUT
  - Lower eyelid colobomas not frequent
  - Cleft palate almost 100%
  - Developmental delay
  - Pre-axial reduction defects of upper (sometimes lower) limb
    - Hypoplasia/ agenesis of the thumbs and radius and one or more metacarpals

I. Facial Dysostoses – Binder Syndrome

- "Maxillonasal dysplasia" due to hypoplasia of the anterior nasal floor and symmetrical maxillary hypoplasia
- Short nose with flat nasal bridge
- Absent frontonasal angle
- Absent anterior nasal spine
- Limited nasal mucosa
- Short collumella and acute nasolabial angle
- Perialar flatness
- Convex upper lip and Class III malocclusion

- ?Autosomal recessive with incomplete penetrance

I. Facial Dysostoses – Pierre Robin Sequence

- Sequence
  - Retrogenia (post displacement of chin)
  - Glossoptosis
  - Airway obstruction
- 50% High arch cleft soft palate
- Glossoptosis causes airway obstruction, increased resp effort, exhaustion, poor feeding, cardiac failure and death
- Treatment is to hold infant prone

II. Atrophy/ hypoplasia

- Parry-Romberg Disease (Progressive Hemifacial Atrophy)
- Radiation Induced Craniofacial Deformity

II. Atrophy/ hypoplasia – Parry-Romberg Disease

- "Progressive hemifacial atrophy"
- F>M, commences 1st or 2nd decade
- Unilateral 95% of cases
- Lymphocytic vasculitis affecting soft tissue and maybe bone
- Cause unknown
  - ? scleroderma
  - ? Infection
  - ? Trigeminal peripheral neuritis
  - ? Cervical sympathetic loss
- Coup de sabre – involvement of frontal and maxillary dermatomes

II. Atrophy/ hypoplasia – Raditaion Induced Craniofacial Deformity

RTX may cause profound disturbances in growth of craniofacial hard and soft tissues

III. Neoplasia/ hyperplasia

- Fibrous Dysplasia
- Neurofibromatosis
- Craniofacial Tumours

III. Neoplasia/ hyperplasia -Fibrous Dysplasia

- Non malignant osseous tumour (malignant deterioration in 0.5% but higher if given RTX)
- AbN activity of bone forming mesenchyme with arrest of maturation in woven bone stage
- May be progressive until adulthood
- Mono- (ribs, femur, tibia, cranium [frontal, sphenoid], maxilla mandible) or Poly-ostotic
- Albright Syndrome
  - Polyostotic FD
  - Abnormal skin pigmentation
  - Precocious puberty
  - hyperthyroidism
- Monostotic form 4X> common than polyostotic form and 30X> common that Albright syndrome
- Clinical problems related to nerve entrapment (esp optic nerve) and cosmesis

III. Neoplasia/ hyperplasia -Fibrous Dysplasia

- Familial fibrous dysplasia (“cherubism”) genetic disorder affecting maxillae and mandible of giant cell type. Self limiting disease of childhood that spontaneously regresses (!!)

III. Neoplasia/ hyperplasia -Neurofibromatosis

- Hereditary AD
- 1:3000 live briths
- NF2 – bilateral acoustic neuromas (Ch22)
- NF1 (Ch 17)
  - more common
  - Benign tumour of skin/ subcutaneous tissue and bone
  - May be neuroorbital – defect in sphenoid bone causing pulsatile proptosis with/without visual loss
  - Sarcomatous degeneration is rare

III. Neoplasia/ hyperplasia –Craniofacial Tumours

- Basically, tumours of base of skull may be approached using “craniofacial techniques” – basically means approaching from above (craniotomy) and below
- Combined neurosurgery/ plastics/ ENT/ H&N
- Must prevent communication between paranasal sinuses and intracranial space (pericranial flaps) at end of procedure

IV. Craniosynostosis

- (Positional plagiocephaly)
- Single suture
  - Sagittal
  - Metopic
  - Unicoronal
  - Bicoronal
  - Lambdoid
- Syndromic Vs Non syndromic
- Specific Syndromes
  - Muenke
  - Crouzon
  - Apert
  - Sathre-Chotzen
  - Pfeiffers
  - Carpenters

Cranial Sutures

- Fibrous union between skull bones
- Allow deformation during delivery, skull expansion with brain protection
- Major growth centres for skull
- Cranial suture complex is dura, bone plates, intervening mesenchyme and overlying periosteum
- Evidence from rats points to dura determining fate of overlying suture
- Intrauterine constraint also produces CS

Craniosynostosis

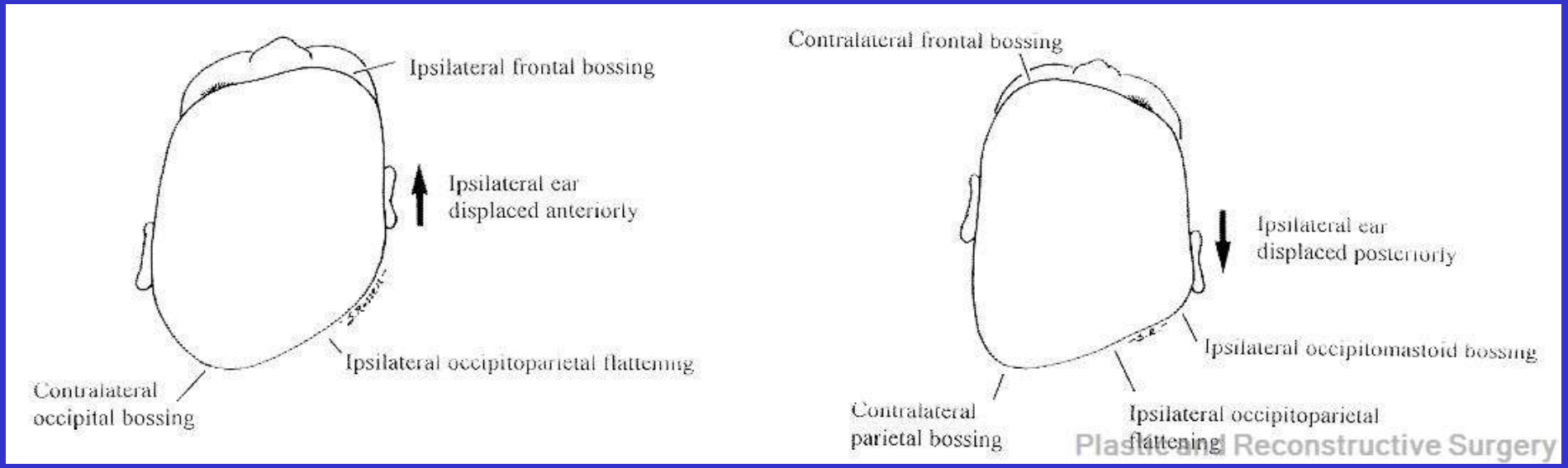
- Craniosynostosis affects 1:2000
- Cosmetic implications to neuro- and vicero-cranium
- Functional issues – visual impairment, deafness and cognitive deficits

- Over 100 known syndromes (esp FGFR1-3 genes and transcription factors TWIST and MSX2
- Phenotype (non syndromic):
  - Sagittal 40-55%
  - Unicoronal 20-25%
  - Metopic 5-15%
  - Lambdoid 0-5%
- Remember Virchow's Law!

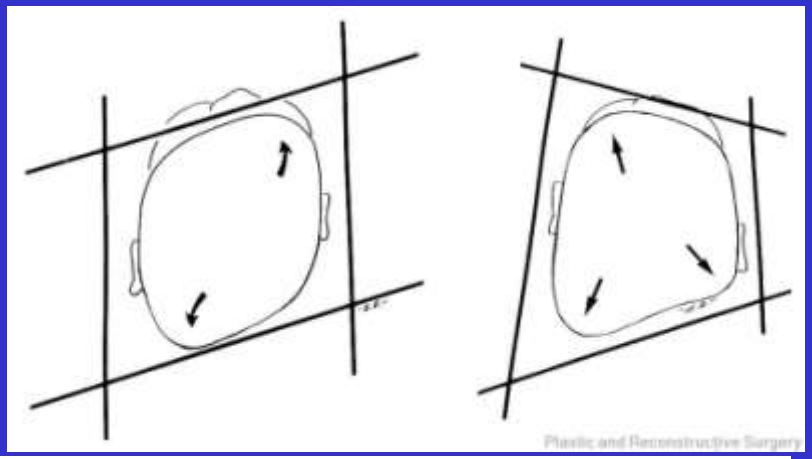
Positional Plagiocephaly
(Deformational Plagiocephaly, Plagiocephaly without Synostosis)

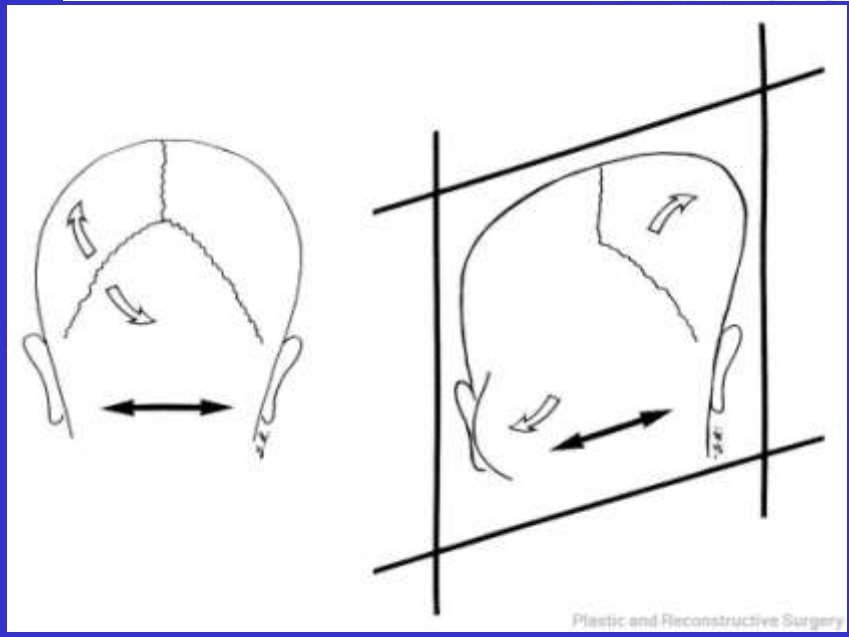
- Very common
- Associated with
  - Multiple births
  - Prolonged labour
  - AbN foetal positioning
  - Hypotonia
  - NICU
  - Torticolis
- Back to sleep campaign
- Parallelogram head
- ? No functional implications
- Treatment is controversial

Positional Plagiocephaly Versus Lambdoid Synostosis



Positional Plagiocephaly Versus Lambdoid Synostosis





Sagittal Synostosis

- Frontal bossing
- Occipital protuberance ("bullet")
- Narrow long head
- CI= max width/max length

Unicoronal Synostosis

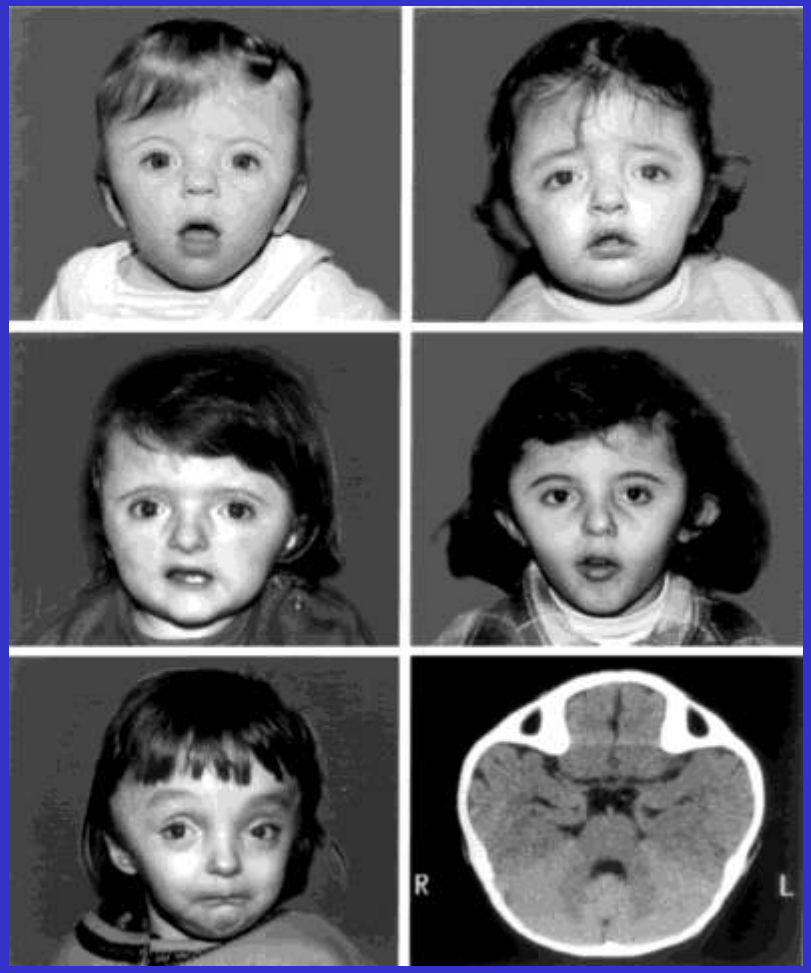
- More open eye more aesthetically pleasing but pathological
- Ipsilateral forehead recession
- Contralateral bossing
- Nasal root deviation to side of fusion
- Facial scoliosis
- Strabismus

Metopic Synostosis

- Prominent forehead ridge
- Trigonocephaly
- Hypotelorism
- Bilateral lateral forehead recession

Muenke Syndrome

- 1:30,000 newborns
- Bicoronal synostosis
- FGFR3 mutation AD
- Hearing loss in 10-30%
- Mild limb abnormalities



Crouzon Syndrome

- 1:60,000 live births
- Bicoronal synostosis
  - Raised ICP
- Maxillary hypoplasia
  - Exorbitism
  - OSA
  - Class III malocclusion
- Normal hands

Apert Syndrome

- Bicoronal synostosis
- Hypertelorism
- Maxillary hypoplasia
  - Exorbitism
  - Upper airway obstruction
  - Class III malocclusion
- Cleft palate
- Developmental delay
- Brain malformation
- Severe symmetrical complex syndactyly

Raised ICP in Apert Syndrome

Craniocerebral Dysproportion

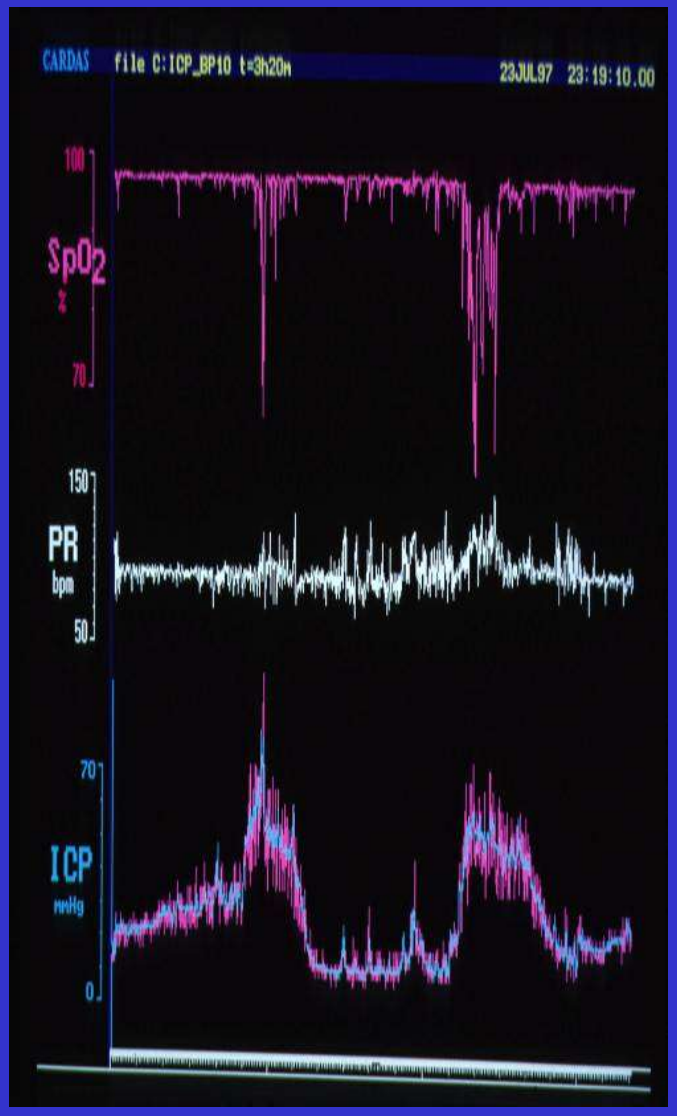
BUT

- Intracranial volume may be NORMAL or INCREASED
- Widely patent sagito-metopic

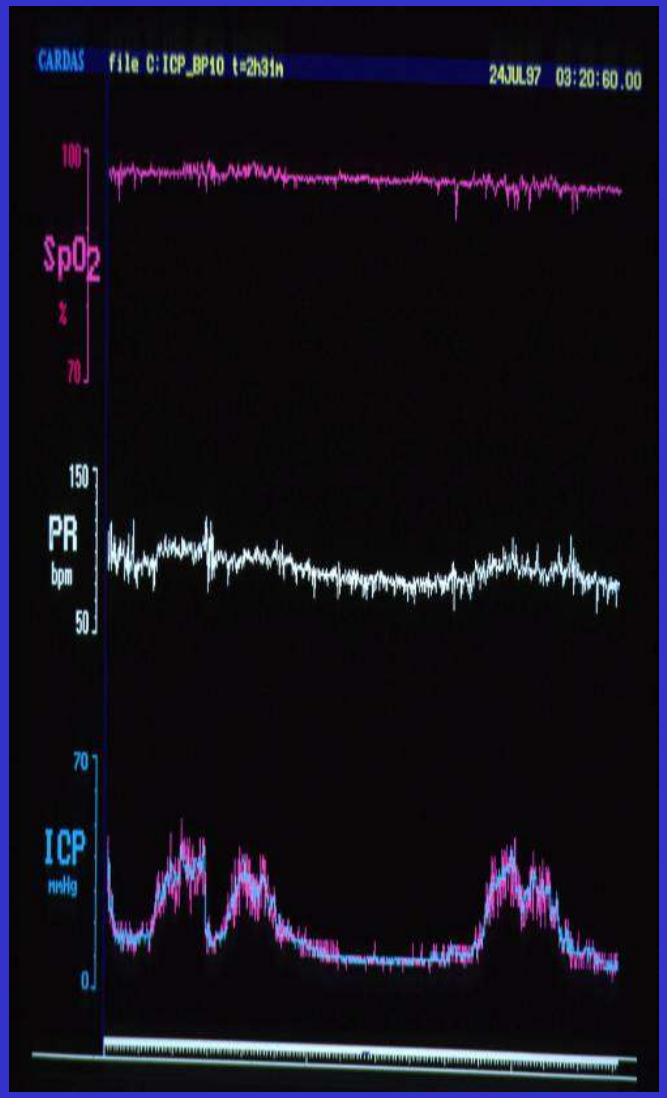
Raised ICP in Apert Syndrome

1. Craniocerebral dysproportion
2. Anomalous venous drainage
   - Impaired venous outflow (impairs CSF reabsorption)
3. Hydrocephalus
   - 40 – 90% have ventricu
4. Obstructive sleep apno
   - CO_2 cerebral vasodilato

OSA & ICP



Pre airway Rx



Post airway Rx

Management of OSA

- NPA
- Adenotonsillectomy
- CPAP
- Midfacial Surgery
- Tracheostomy

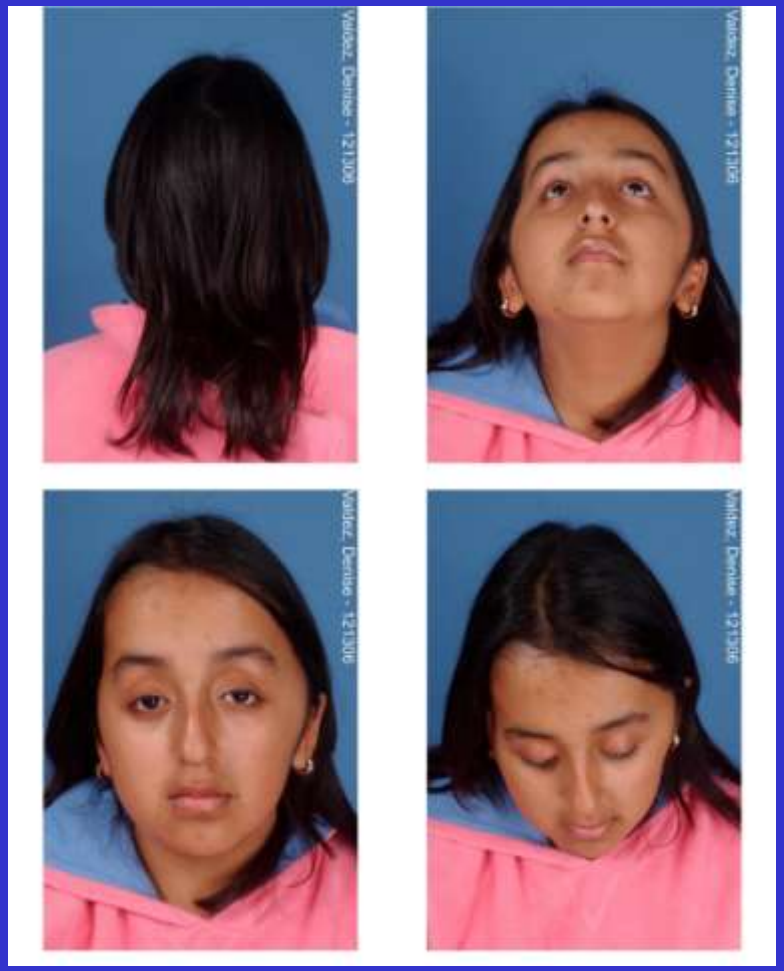
Raised ICP in Apert Syndrome

1. Craniocerebral dysproportion
2. Anomalous venous drainage
3. Hydrocephalus
4. Obstructive sleep apnoea

83% of patients with Aperts developed raised ICP by the age of 5 in recent GOSH study

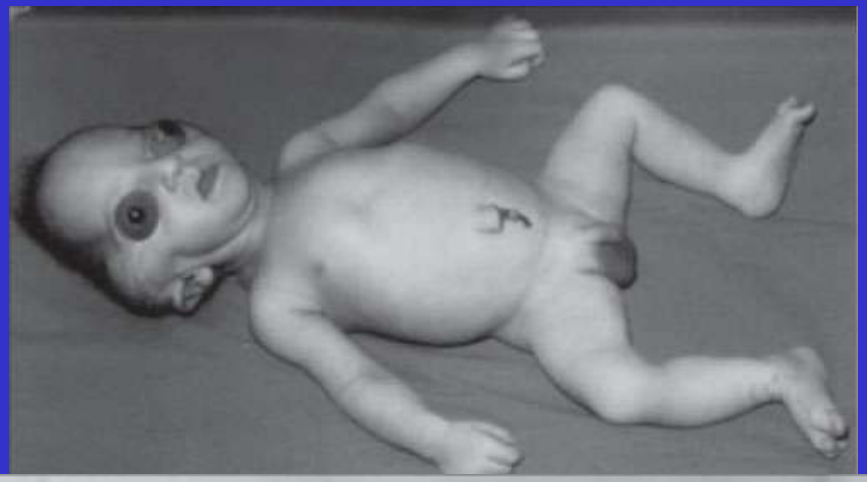
Sathre Chotzen Syndrome

- AD TWIST gene
- Uni or bicoronal synostosis
- High forehead
- Low frontal hairline
- Ptosis
- Ear abnormalities
- Syndacytyly
- Brachydactyly



Pfeiffers Syndrome

- Bicoronal synostosis
- Midfacial hypoplasia
- Broad toes and thumbs
- Variable soft tissue syndactyly
- AD complete penetrance
- May be associated with clover leaf skull



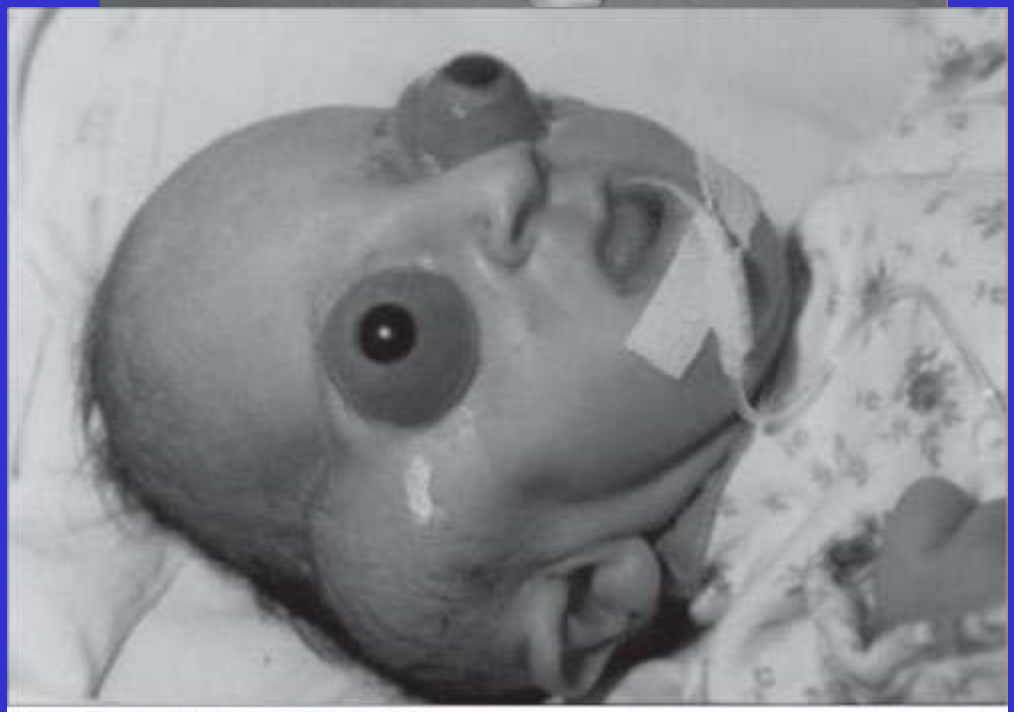
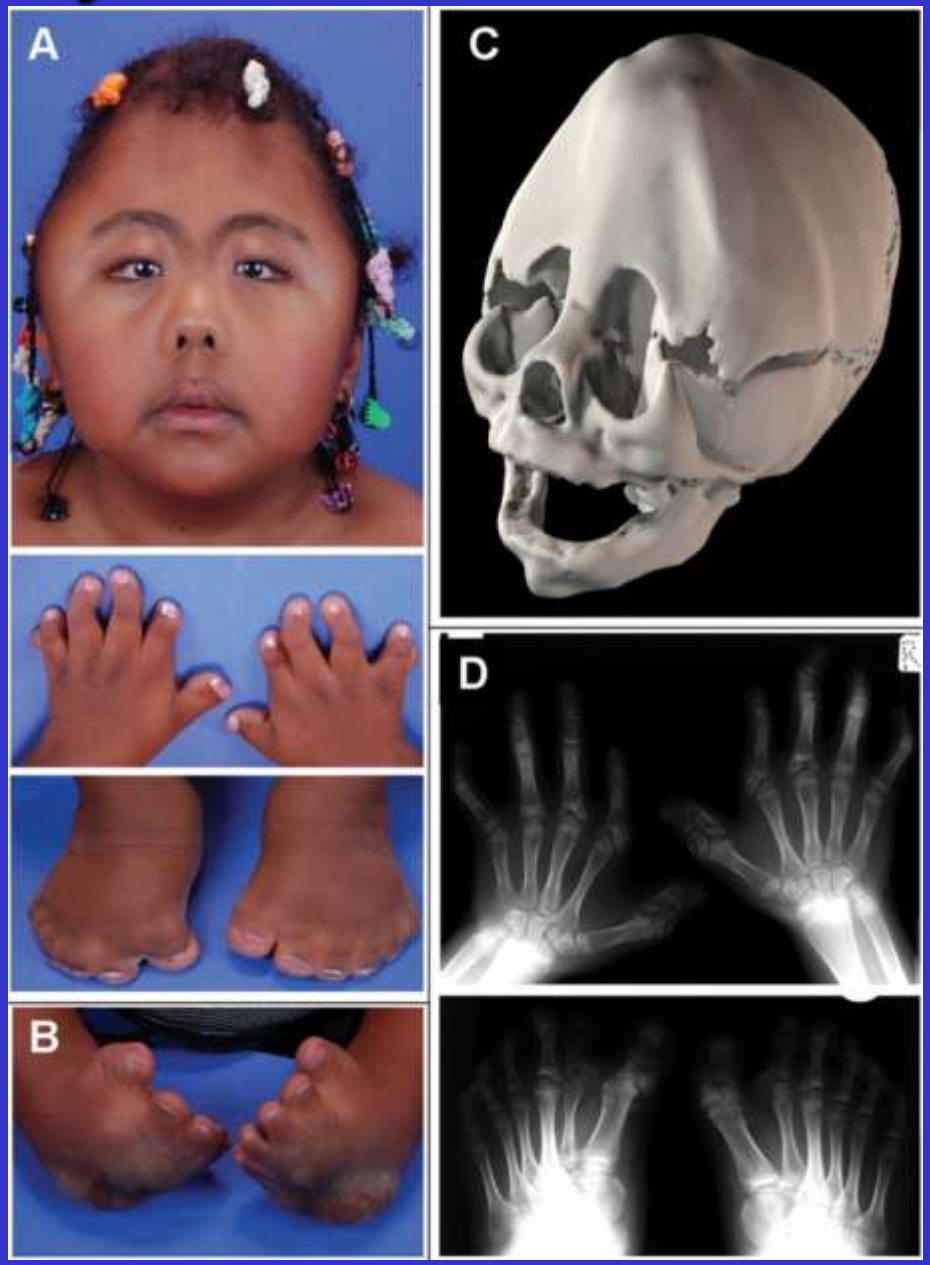


Figure 2. Cloverleaf skull, exorbitism and low-set ears.

Carpenters Syndrome

- Bicoronal synostosis
- Pre axial polysyndactyly
- Short fingers with clinodactyly
- AUTOSOMAL RECESSIVE!



The Exam/Lake Analogy

You need to have enough knowledge to cover everything a little bit, with deeper knowledge in some areas

My Exam Tips

You need to act and sound like a junior consultant talking to a group of senior consultants (who are considering you to be their locum for a month)

DO NOT INVENT AN OPERATION DURING THE EXAM. IF YOU'VE NEVER HEARD OF A PROCEDURE, DON'T SAY YOU WOULD DO IT

You must be able to draw cleft lip/palate repairs and all forms of upper/lower lip, upper/lower eyelid, ear and nose reconstruction/flaps

My Exam Tips

Answer the question

and if you don't know, say so and move on. You can't bluff the examiners. Don't waffle.

Surgical name-dropping is good.

Find out what the patient wants

Rock the examiners to sleep with soothing answers and then don't startle/ wake them

My Exam Tips

The examiners **want** to pass you – so let them.