The contents of all presentations are password protected. The copy is protected by copyright of the Authors. Consent has been given for the express purpose of educating attendees of the March 2010 Registrars’ Conference in Sydney.

You **MAY NOT COPY OR DISTRIBUTE** the contents or images in any form.

You **MAY PRINT** the document for your own personal use as an educational resource.
Craniofacial Surgery and Syndromes

Damian Marucci MBBS BA PhD FRACS
Plastic and Reconstructive Surgeon
Craniofacial Surgeon
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. Encephaloceles

- 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
  - Encephaloceles
  - 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

- Variously 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 – Radiation 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 births
- 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 ventricular dilatation
4. Obstructive sleep apnoea
   - CO₂ cerebral vasodilator
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
- Syndacdyly
- Brachydactyly
Pfeiffer's Syndrome

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