Help! I've never heard of this syndrome….

Corina Lee

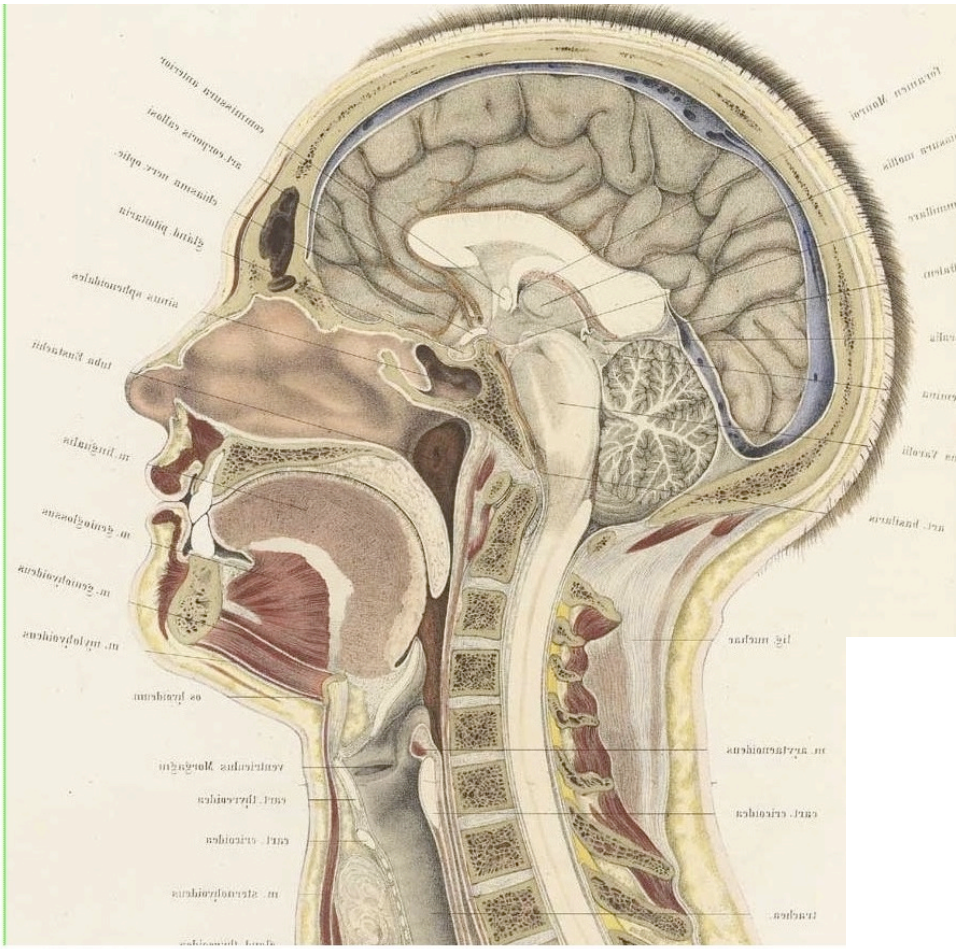
November 2014

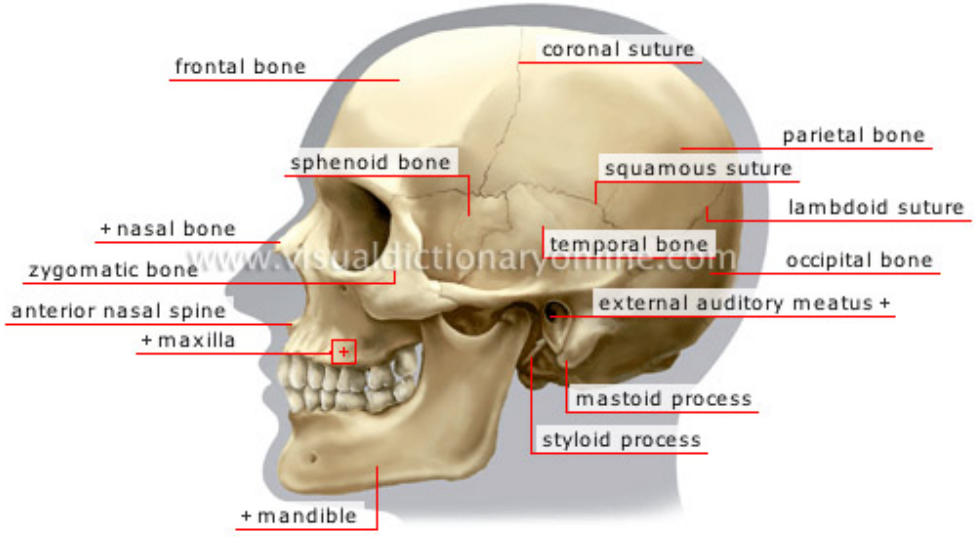
Anytime, anywhere…

- Any geographical location
- Any hospital
- Elective/emergency
- Surgery/medical stabilisation
- For investigations
- In adulthood…

NAP 4

- 13 paed case (7%)
- 3 deaths (A&E, ICU, theatres)
- 3 congenital abnormalities
- 9/13 < 4 years old




frontal bone
coronal suture
parietal bone
sphenoid bone
squamous suture
lambdoid suture
+ nasal bone
temporal bone
occipital bone
zygomatic bone
external auditory meatus +
anterior nasal spine
+ maxilla
mastoid process
styloid process
+ mandible
www.visualdictionaryonline.com

Anatomical Segments

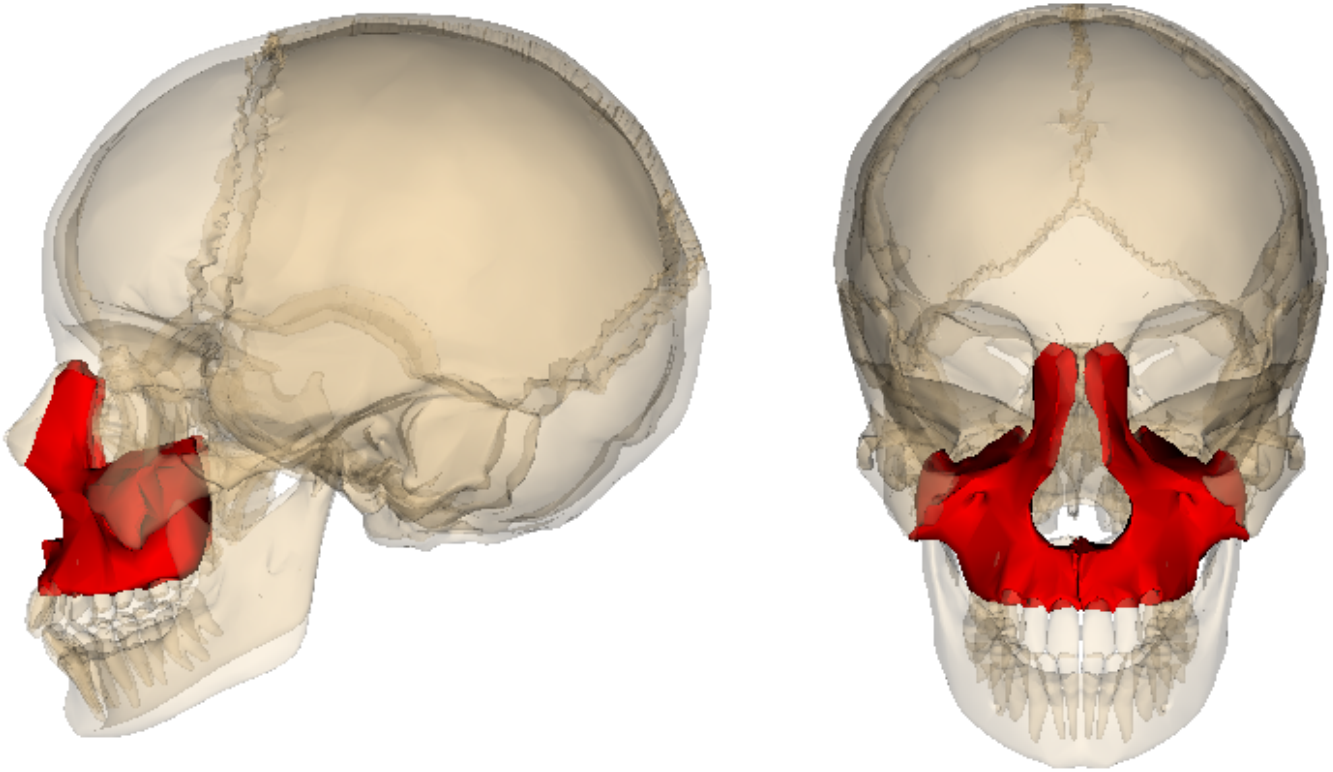
1. Maxillary Hypoplasia
2. Mandibular Hypoplasia
3. Combined maxillary and mandibular hypoplasia
4. Enlarged tongue

Airway Management in Children with Craniofacial Anomalies

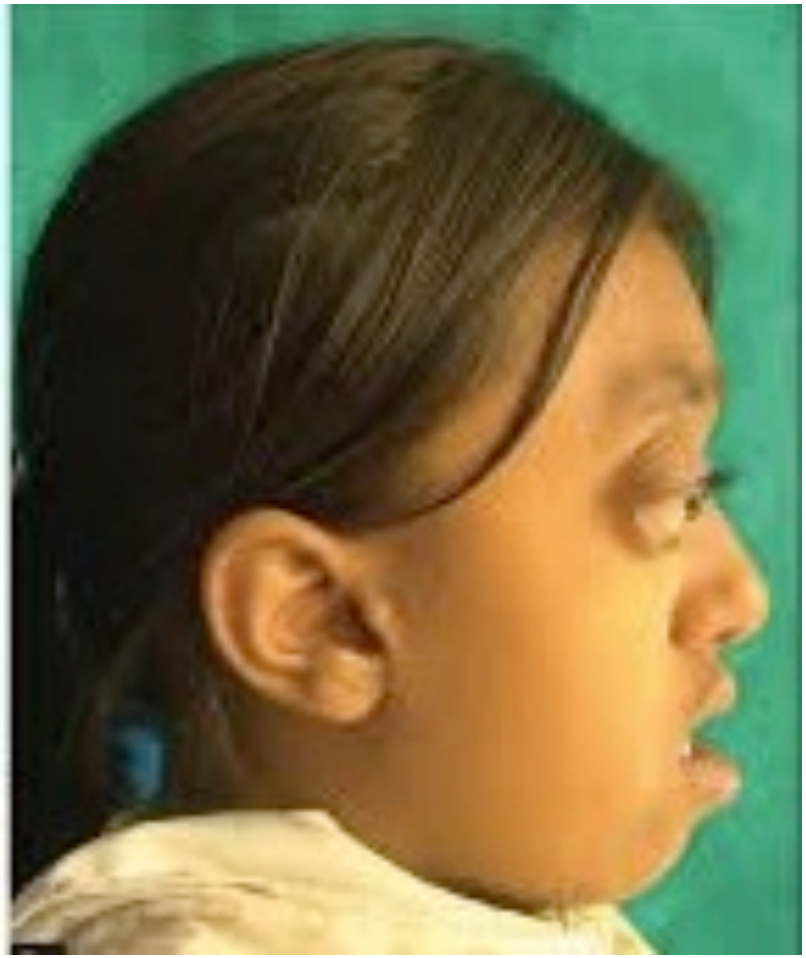
Jonathan A. Perkins, Kathleen C. Y. Sie, Henry Milczuk, Mark A. Richardson

The Cleft Palate-Craniofacial Journal: March 1997, Vol. 34, No. 2, pp. 135-140.

- 109 patients
- symmetric mid-face and/or mandibular hypoplasia
- airway intervention
  - Positioning
  - Nasopharyngeal airway
  - Tracheotomy
  - Other
  - None



Mid-face Hypoplasia



42% airway intervention

- Crouzon
- Apert
- Pfeiffer
- Saethre-Chotzen
- CHARGE

Maxillary Hypoplasia

- OSA 50%
- Palate high and arched
- Normal mandible
- Choanal stenosis
- Mouth breathers
- Tracheal ring abormalities

Maxillary Hypoplasia

- Mouth open for BVM
- Face mask fit may be tricky
- Laryngoscopy usually straightforward
- May require smaller tube

ORIGINAL ARTICLE

Pediatric Anesthesia

Perioperative complications in children with Apert syndrome: a review of 509 anesthetics

Sarah Barnett, Claire Moloney & Robert Bingham

Great Ormond Street Hospital, London, UK

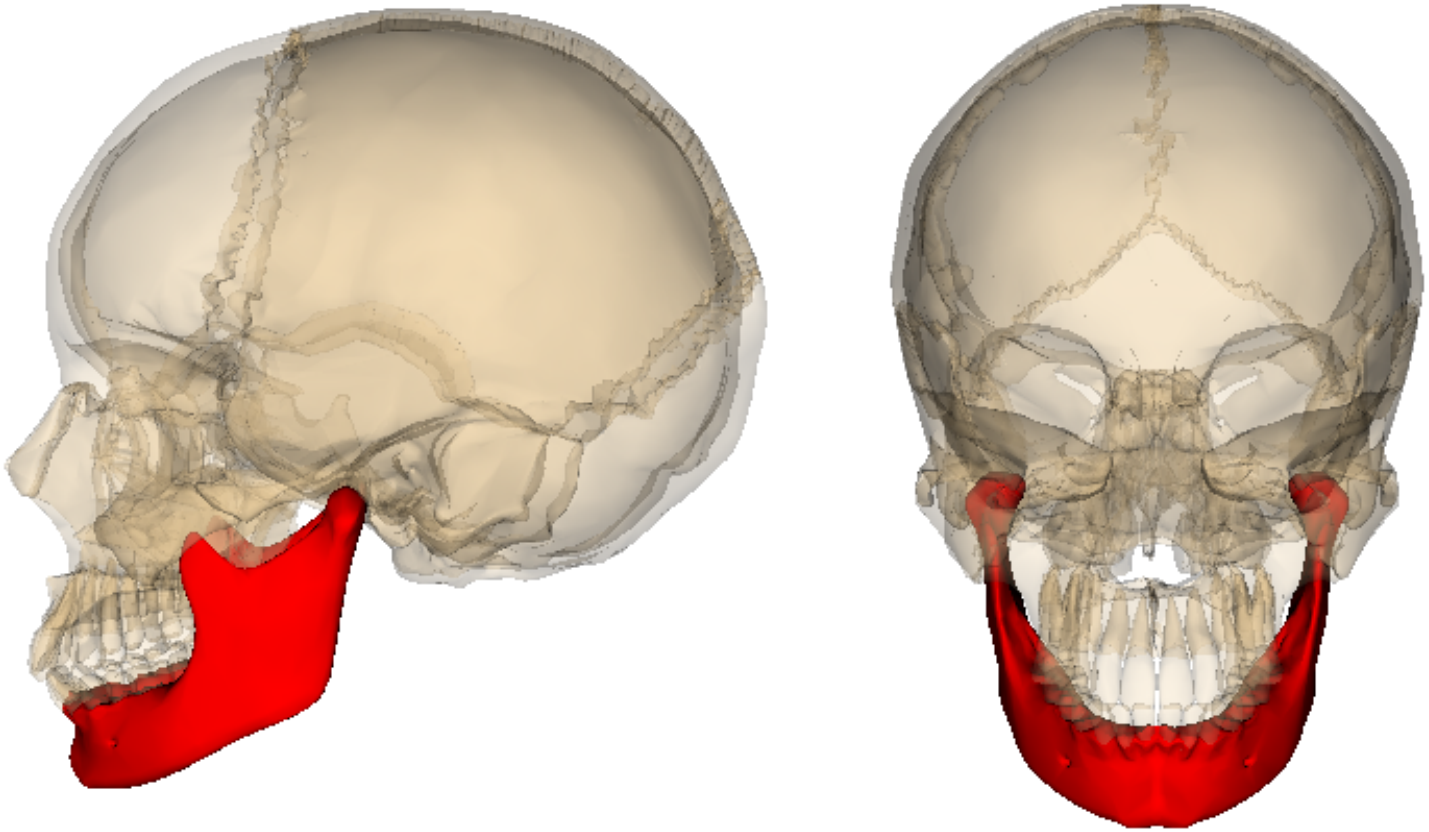
Facemask	68 (13%)
LMA	206 (40%)
Tracheal tube	199 (39%)
- **196 grade 1 or 2, 3 grade 3**	
Tracheostomy	36 (7%)

Pre-operative OSA 50%

Supraglottic obstruction at induction 19 (4%)

- **ALL managed with simple airway manoeuvres**

No subglottic obstruction

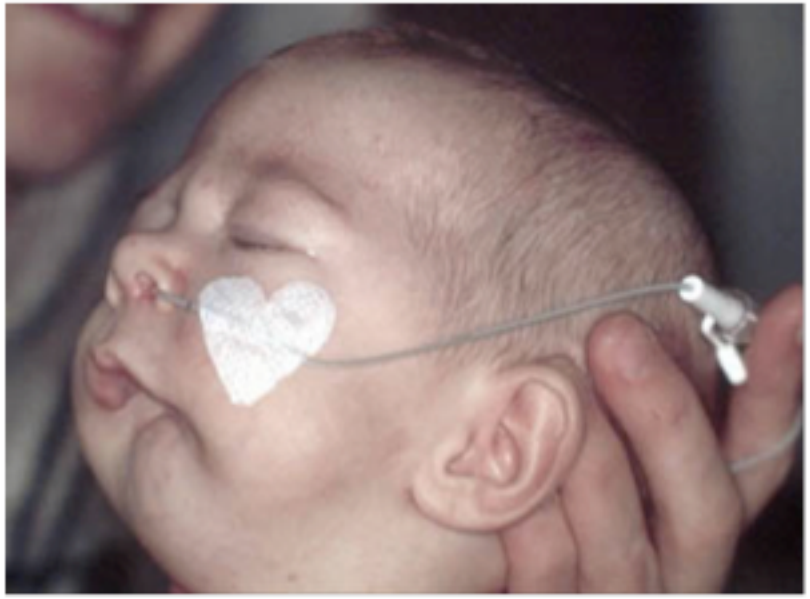


Mandibular Hypoplasia

72% airway intervention

- Pierre Robin Sequence
- Stickler's
- Nager

Mandibular Hypoplasia



- Reduced anterior mandibular space
- High anterior larynx
- Nowhere for tongue to be displaced
- Often cleft palate

Pierre Robin Sequence

- Micrognathia
- Glossoptosis
- Respiratory obstruction
- Cleft palate

The Laryngoscope

Airway Management for Intubation in Newborns With Pierre Robin Sequence

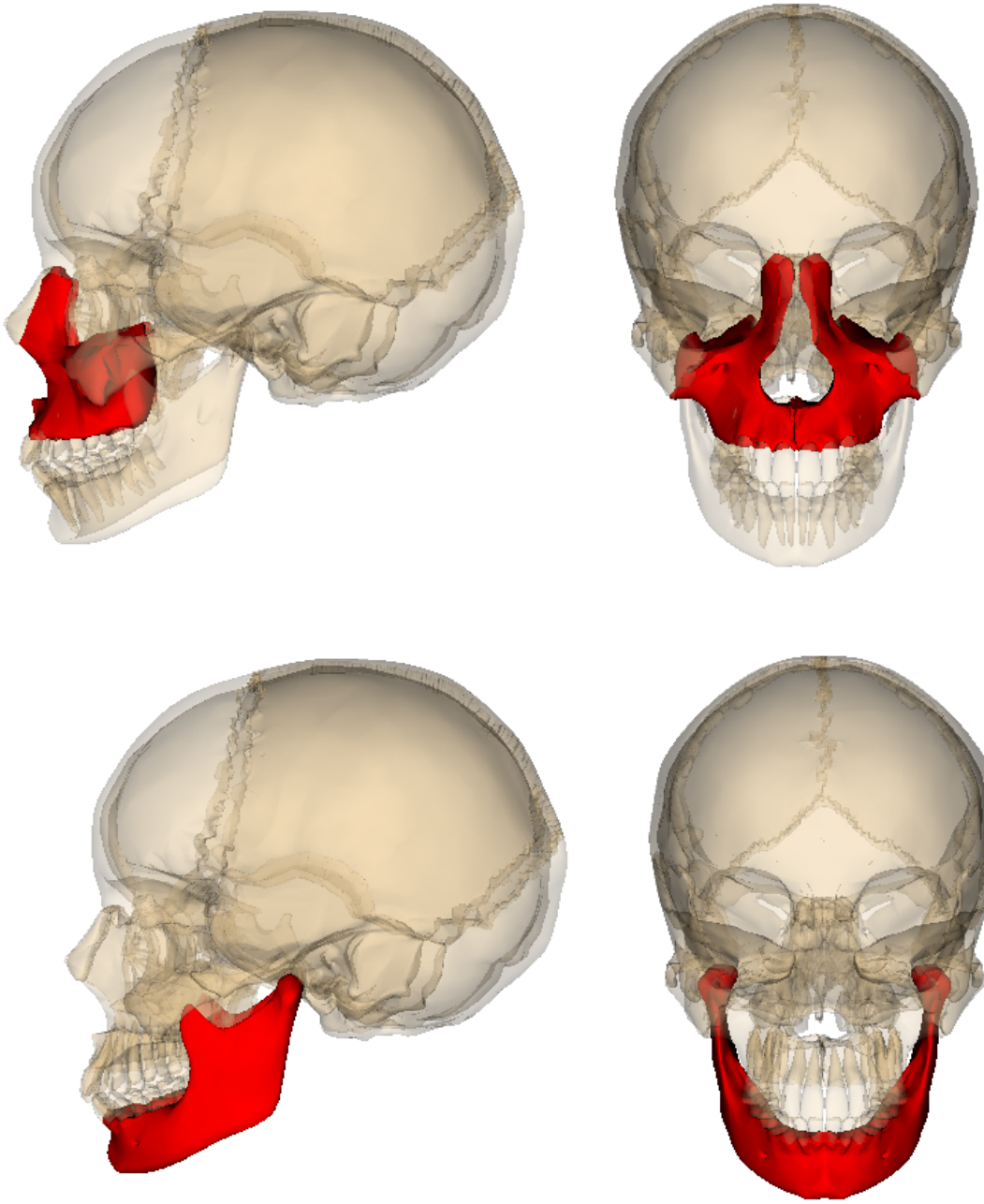
Alexander P. Marston, BA; Timothy A. Lander, MD; Robert J. Tibesar, MD; James D. Sidman, MD

- 33 PRS patients, 35 intubations
  - 13/35 (37%) direct laryngosocpy
  - 22/35 (63%) fibreoptic bronchoscope

- No problems BVM
- No tracheostomy

Mandibular Hypoplasia

- Consider oral/ pharyngeal airway
- ?OSA – mask ventilation
- Alternative airway techniques
- Fibreoptic
- Tracheotomy

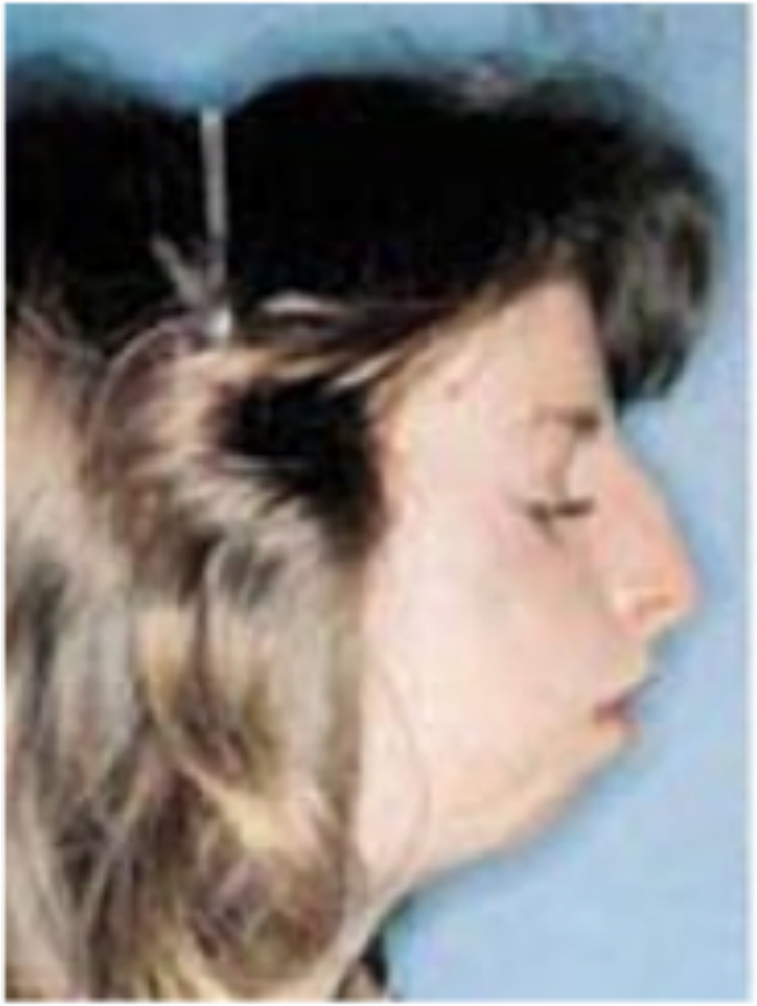


Hypoplastic Maxilla AND Mandible

71% airway intervention

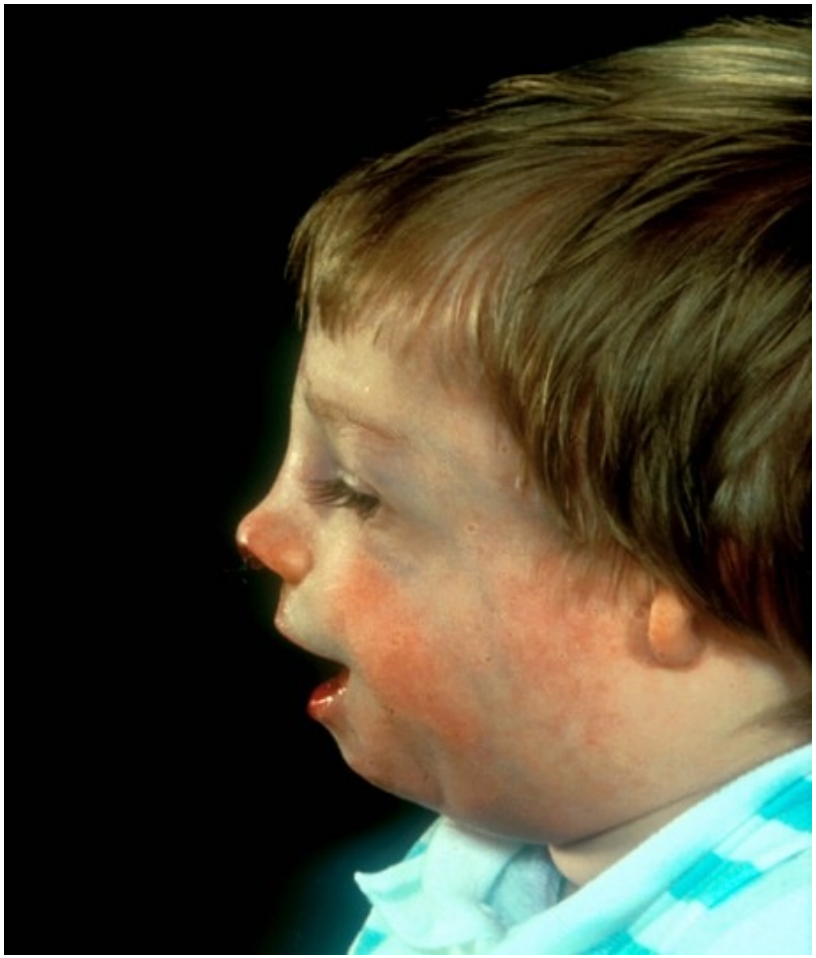
- Treacher Collins
- Bilateral hemifacial microsomia

Hypoplastic Maxilla AND Mandible



- Nasal obstruction
- OSA
- Reduced submandibular space

Treacher-Collins Syndrome



- Mandibular/malar/ zygomatic hypoplasia
- Down-slanting palpebral fissures
- Eyelid colobomas
- Malformation external pinna and ear canal
- Cleft palate/ velopharyngeal

ORIGINAL ARTICLE

Pediatric Anesthesia

Anesthesia for Treacher Collins syndrome: a review of airway management in 240 pediatric cases

Jane Hosking[1], David Zoanetti[1,2], Alison Carlyle[3], Peter Anderson[2,4] & David Costi[1,2]

35 children, 240 anaesthetics

BVM	easy	63 (26%)
	difficult	22 (9%)

- **all** became easy with LMA

trache in situ	42	(17%)
no comment	113	(47%)

ORIGINAL ARTICLE

Anesthesia for Treacher Collins syndrome: a review of airway management in 240 pediatric cases

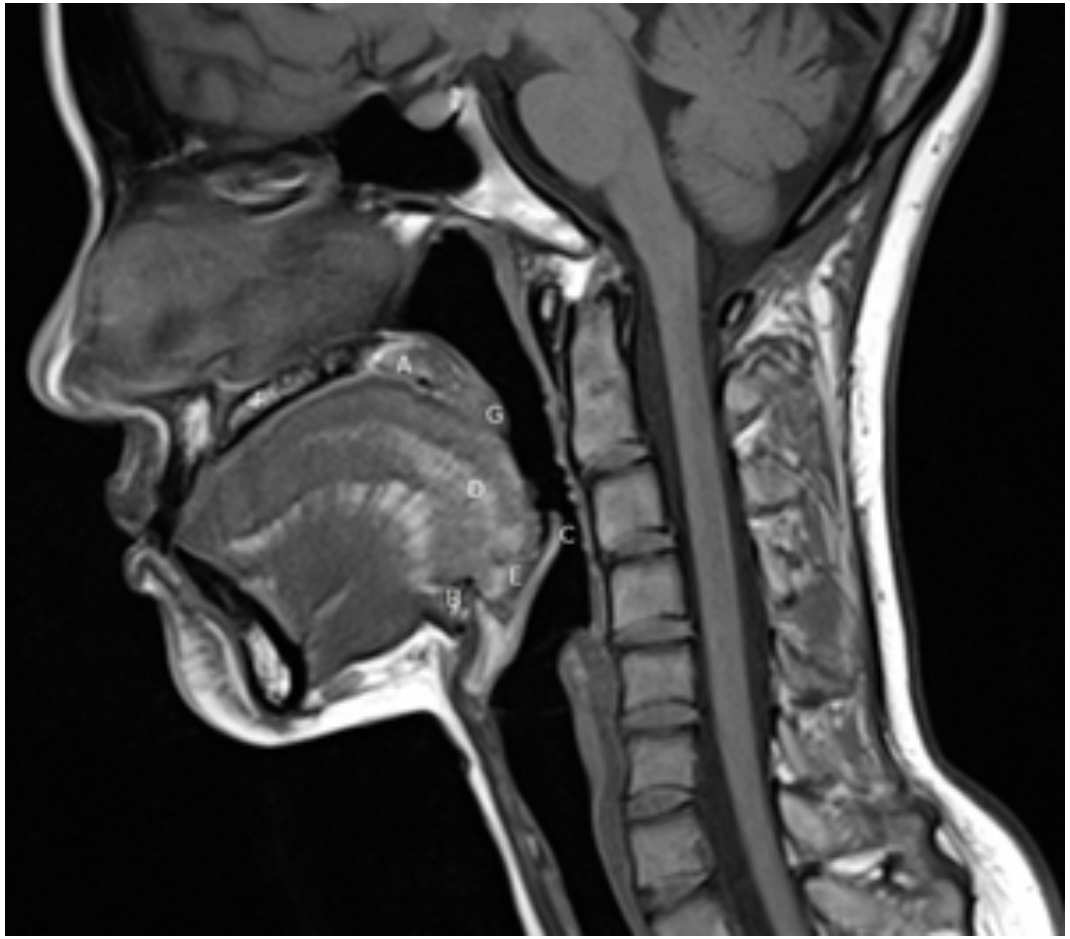
Jane Hosking[1], David Zoanetti[1,2], Alison Carlyle[3], Peter Anderson[2,4] & David Costi[1,2]

Intubation:

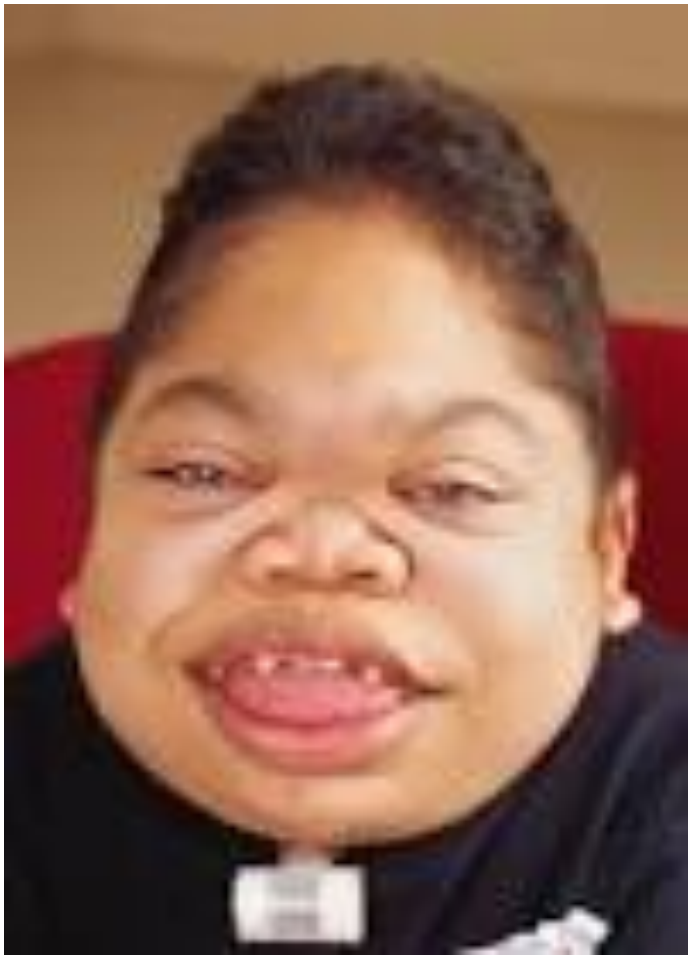
- Failed intubation rate 5%
- 41% used alternative technique from direct laryngoscopy
- Increasing age associated with higher Cormack-Lehane Grade

Hemifacial microsomia

- As severity mandibular deformation increases, intubation difficulty increases
- Often more difficult after surgery
- Tracheal intubation AND mask ventilation may be difficult

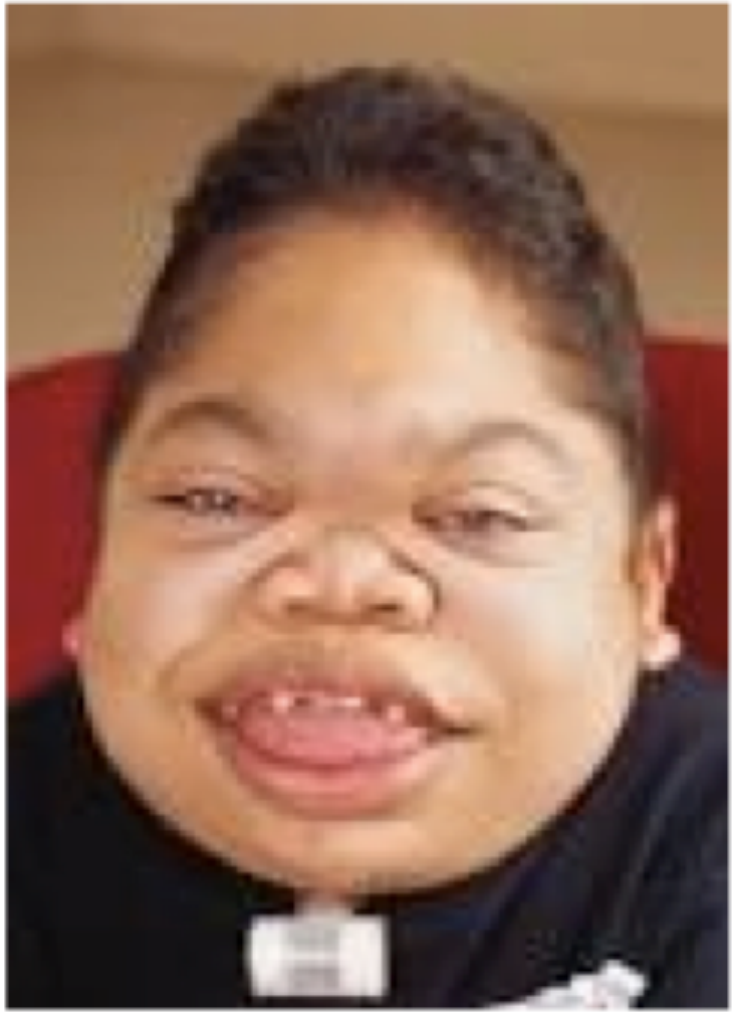

A
G
D
C
E
B

Macroglossia



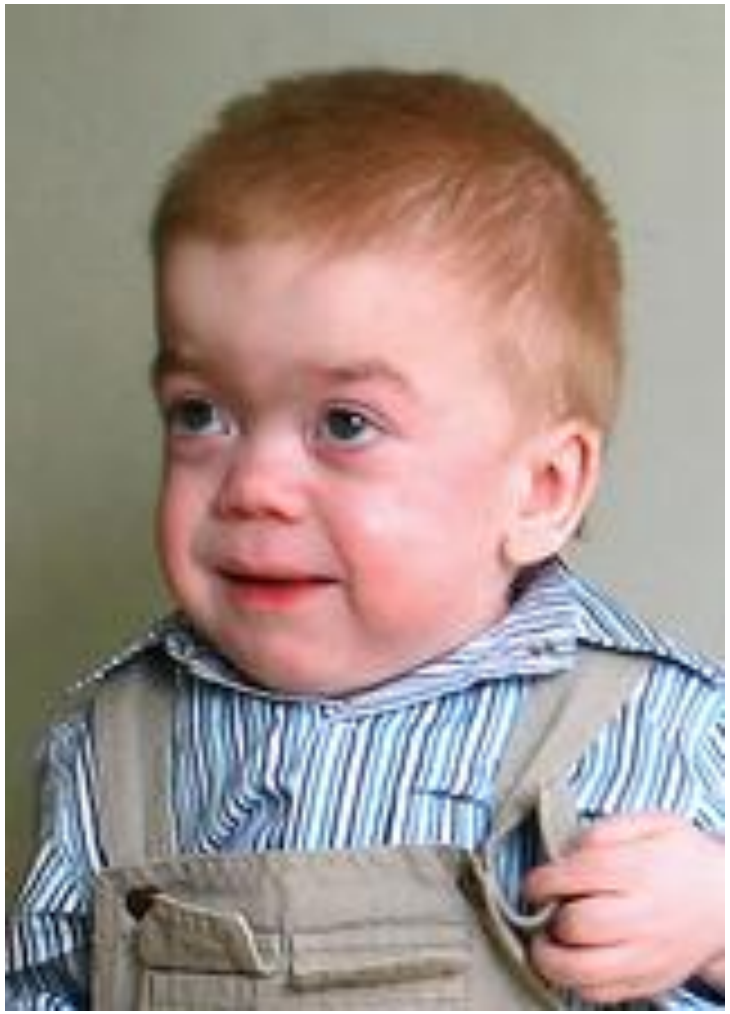
- Mucopolysaccharidoses (Hunter's/Hurlers)
- Beckwith-Wiedemann
- Down's

Macroglossia



- Obstruct airway
- Hypoxia may lead to cor pulmonale
- Difficult BVM and intubation

Mucopolysaccharidoses



- Enlarged tongue
- Thickening soft tissue oropharynx, larynx
- Profuse secretions
- Blockage nasal passages
- Progressive airway obstruction
- Limited neck movement
- Learning disabilities

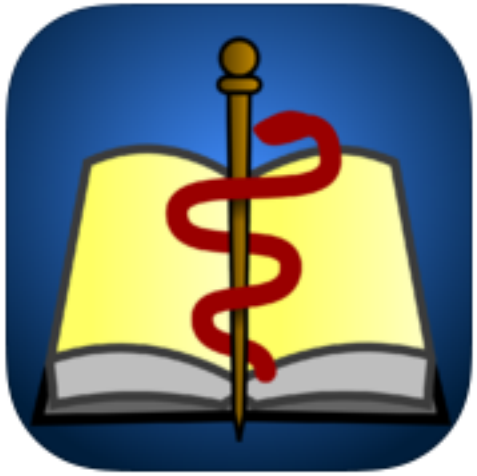
Mucopolysaccharidoses – the bottom line!

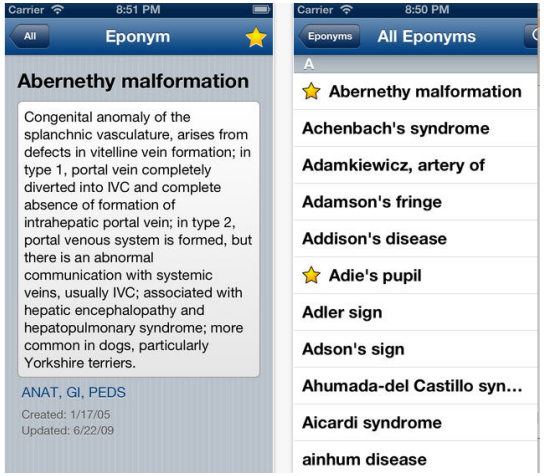
- Difficult facemask ventilation – 14.2%
  - No Rx 26.7%, BMT 4.2%, ERT 12.5%
- Failed intubation – 1.4 - 3%
- Difficult intubation (all MPs) – 28%
  - Odds ratio BMT 0.36, ERT 3.95
- BMT < 2year age decreases airway management problems compared ERT and no treatment

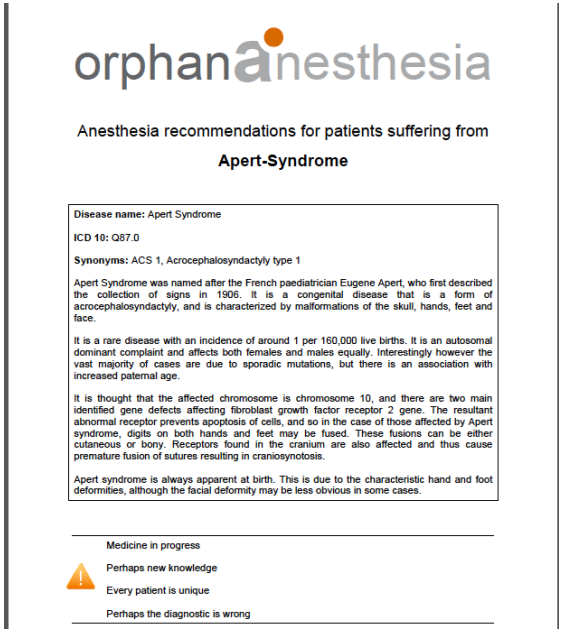
Frawley et al. A retrospective audit of anesthetic techniques and complications in children with mucopolysaccharidoses. Pediatric Anesthesia 22(2012) 737-744

Resources

- Eponyms app
- www.orphananesthesia.eu






orphananesthesia

Anesthesia recommendations for patients suffering from

Apert-Syndrome

Disease name: Apert Syndrome

ICD 10: Q87.0

Synonyms: ACS 1, Acrocephalosyndactyly type 1

Apert Syndrome was named after the French paediatrician Eugene Apert, who first described the collection of signs in 1906. It is a congenital disease that is a form of acrocephalosyndactyly, and is characterized by malformations of the skull, hands, feet and face.

It is a rare disease with an incidence of around 1 per 160,000 live births. It is an autosomal dominant complaint and affects both females and males equally. Interestingly however the vast majority of cases are due to sporadic mutations, but there is an association with increased paternal age.

It is thought that the affected chromosome is chromosome 10, and there are two main identified gene defects affecting fibroblast growth factor receptor 2 gene. The resultant abnormal receptor prevents apoptosis of cells, and so in the case of those affected by Apert syndrome, digits on both hands and feet may be fused. These fusions can be either cutaneous or bony. Receptors found in the cranium are also affected and thus cause premature fusion of sutures resulting in craniosynotosis.

Apert syndrome is always apparent at birth. This is due to the characteristic hand and foot deformities, although the facial deformity may be less obvious in some cases.

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

Butler MG et al. **Specific Genetic Diseases at Risk for Sedation/ Anesthesia Complications**. Special Article. Anesth Analg 2000;91:837-55

Recommendations

- On-line, fast resources / apps
- Can often predict
- Think about anatomical segments
- Equipment – remember NP airways, LMAs
- Team – consider early ENT involvement
- Tracheostomy may be required
- Practice combined techniques