(ENT Perspectives) on Pediatric CranioMaxilloFacial (CMF) Abnormalities

NICU Grand-Rounds 9/14/23

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Disclosures

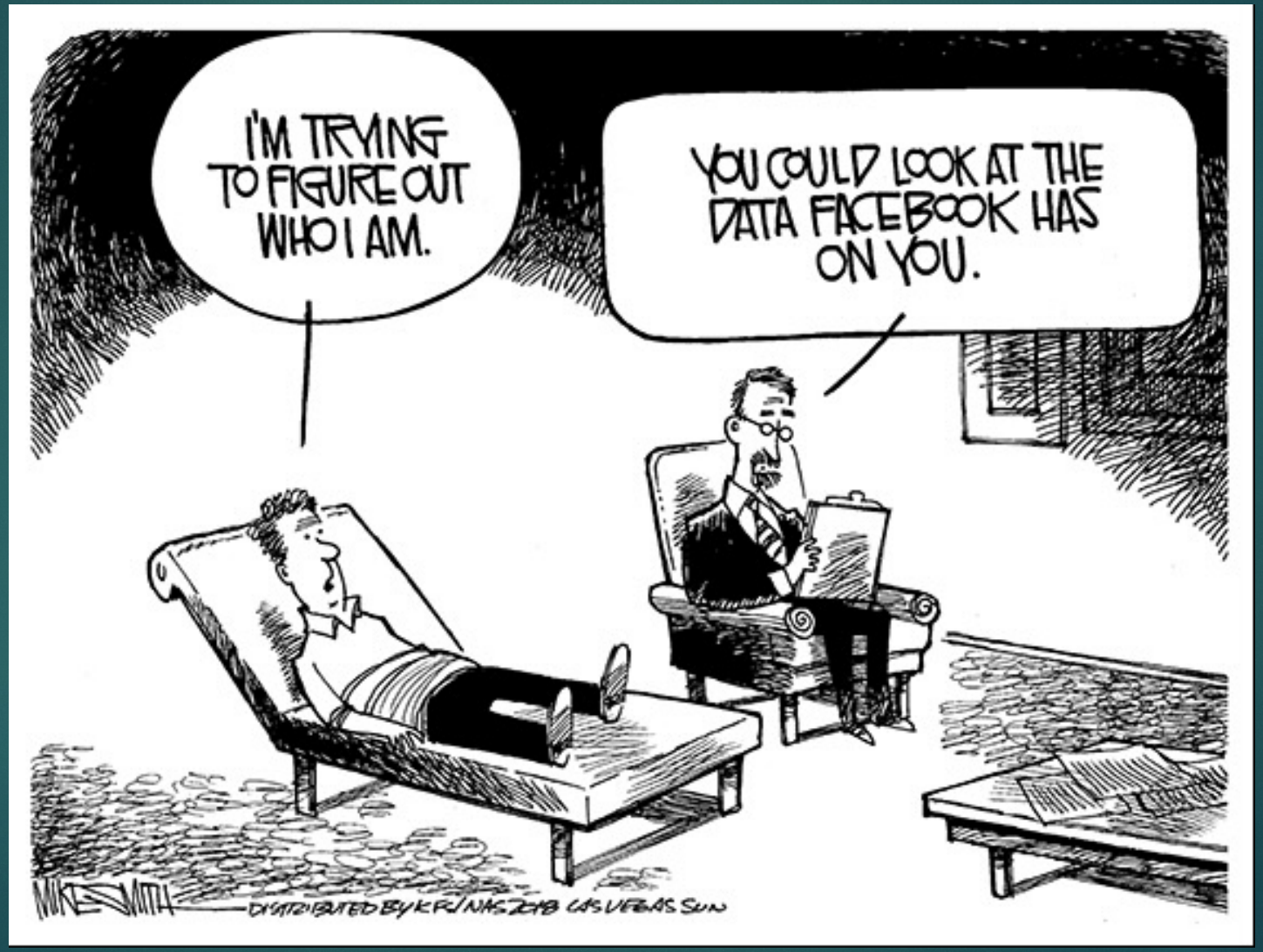
- (every now and then) I perform consultation work for *Karl Storz*

Goals

- At the conclusion of this activity, you should be able to:

1. Identify different types of pediatric craniomaxillofacial (CMF) abnormalities
2. Identify potential difficult airway in a child with CMF abnormalities
3. Learn mechanism to manage and secure a potential difficult pediatric airway
4. Hearing difficulties and hearing restoration in children with CMF abnormalities

Who am I?


I'M TRYING TO FIGURE OUT WHO I AM.
YOU COULD LOOK AT THE DATA FACEBOOK HAS ON YOU.
MIKE SMITH
DISTRIBUTED BY KFI/NHS 2018 LAS VEGAS SUN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Greetings from Basel

Itinerary

- **Non-Syndromic H&N abnormalities**
  - Ear
    - Pinna / External ear malformations
    - Microtia / Anotia
    - Hearing Loss
  - Nose
    - Pyriform aperture stenosis
    - Nasal obstruction (e.g. lacrimal duct cysts)
    - Choanal atresia
  - Airway
    - Laryngomalacia
    - Vocal Cord Paralysis
    - Subglottic stenosis
  - Oral cavity
    - Common (benign) findings
    - Ankyloglossia and (maybe maxillary lip tie)
    - Cleft lip and palate
  - Head & Neck
    - Micrognathia
    - (benign) “Lumps and Bumps”
    - Branchial Cleft Cysts
    - Thyroglossal Duct Cyst
    - Vascular Malformations

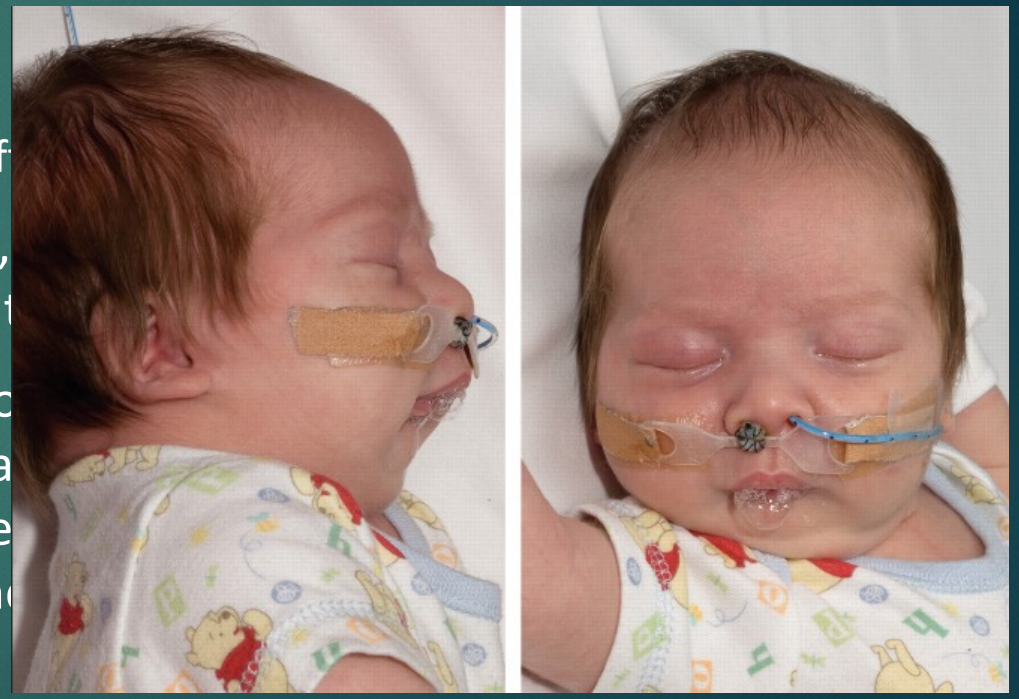
Itinerary Cont.

- **Syndromic H&N abnormalities**
  - Common triple chromosome anomalies (Trisomy 21, 18 and 13)
  - Pierre Robin Sequence (PRS)
  - Craniofacial dysostosis
    - Apert Syndrome
    - Crouzon Syndrome
    - Pfeiffer Syndrome
  - Treacher Collins Syndrome
  - Goldenhar's Syndrome
  - Klippel-Feil Syndrome
  - Beckwith-Wiedemann syndrome
  - Mucopolysaccharidosis (MPS)

… there are most likely many more, but impossible to cover (in one lecture…)

Pediatric Airway – In General

- (!) The incidence of difficult airway is higher in children with craniofacial syndromes than normal children.
- **Common challenges:**
  - uncooperative child
  - unreliability of the Mallampati scoring for prediction of the dif
- **History: -** snoring / apneas, croup, stridor, voice hoarseness, smoking, and most importantly a history of previous difficult
- **General airway examination**: a baseline oxygen saturation c position (prone position must be red-flagged as upper airwa of any obstruction manifested by intercostal/suprasternal re opening, teeth, tongue size and Mallampati score, shape an distance, and neck length/mobility.



- (!) Take a **lateral "profile" look** of the mandible which could spot a micrognathia or a retracted mandible.

Pediatric Airway - Evaluation

- **Risk factors which may potentially contribute to difficult airway management:**

1. An extremely short thyro-mental distance with an overbite, with micro/retrognathia such as seen in Pierre Robin sequence and Treacher-Collins syndrome;
2. A fixed neck such as in Klippel-Feil syndrome;
3. A small oral opening and large tongue such as seen in Beckwith-Wiedemann syndrome;
4. Obstructive sleep apnea +/− secondary pulmonary hypertension;
5. Stiff subcutaneous tissues as seen in Mucopolysaccharidosis (high risk of difficult ventilation);
6. Midface hypoplasia as seen in Apert and Crouzon syndromes;
7. Obstruction of the airway when in supine position and the need to continuously maintain in prone positioning;
8. History of a previous failed airway;
9. Soft tissue tumors and vascular malformations with significant obstruction of the airway

Syndromic Children and Airway

Anatomical malformation	Syndrome
Overbite	Pierre Robin-Treacher Collins-Goldenhar
Mandibular prognathism	Apert
Small mouth	Pierre-Robin, Treacher-Collins, Goldenhar, Down's syndrome
Large mouth	Mucopolysaccharidosis, Beckwith-Wiedemann
Palate (high arched)	Down, Crouzon, Apert, Pierre Robin, Treacher Collins, Mucopolysaccharidosis
Cleft Palate	Pierre Robin, Treacher Collins, Goldenhar, Down's, Apert, Crouzon
Tongue abnormality (Large or Glossoptosis)	Beckwith-Wiedemann, Mucopolysaccharidosis, Pierre Robin.
Short thyromental distance (less than 3 finger breadths used by the own child's fingers)	Pierre Robin, Treacher Collins, Goldenhar,
Subglottic narrowing	Down's syndrome
Tracheal abnormality (tortuosity)	Mucopolysaccharidosis
Cervical spine abnormalities	Down's syndrome, Klippel-Feil, Mucopolysaccharidosis, Goldenhar

An Approach to the Airway Management in Children with Craniofacial Anomalies

WRITTEN BY

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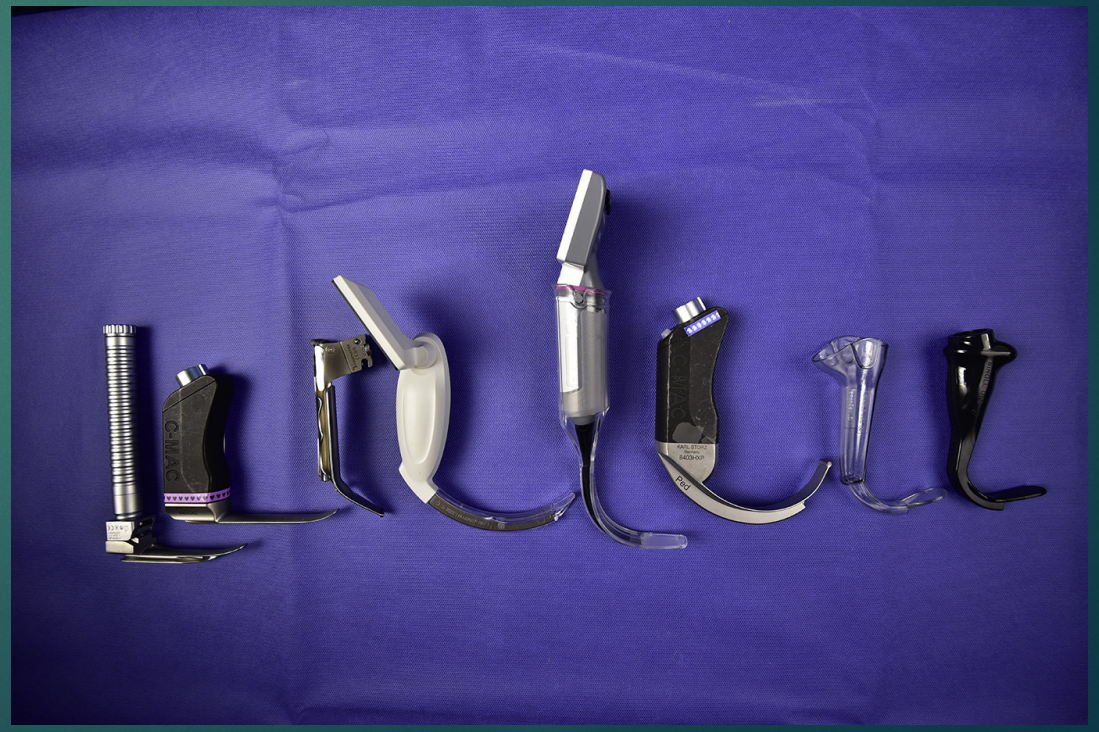
Airway Management in Children with Craniofacial Syndromes

- Have a Plan (A, B, C, ….)!

- **Plan A:** *Preserve spontaneous ventilation*
- Induction using IV sedative medication: propofol infusion or boluses (also dexmedetomidine, ketamine)
  Or
- Inhalation induction with Sevoflurane
- A wide range of airway tools and techniques have been described for the intubation. However, it is crucial that the airway operator sticks with the tool that he is mostly familiar with. It is noteworthy to keep in mind that an airway tool is not a plan.



Intubation Techniques in Syndromic Children:

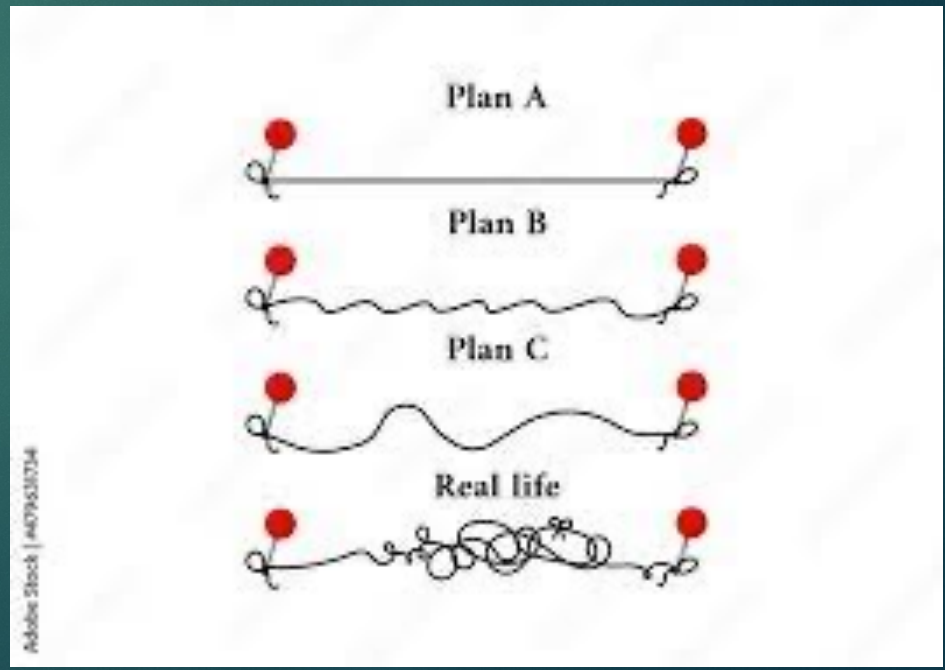
1. **Direct laryngoscopy**: a regular laryngoscope blade (a Macintosh curved or a Miller straight blade).
2. The paraglossal approach: The laryngoscope's straight blade is inserted through the trench between the tonsils and the base of the tongue
3. Intubation with direct laryngoscopy may be facilitated with gum elastic bougie or a stylet.
4. **Video laryngoscopy**: Storz video laryngoscope (C-MAC) or the Glidescope
5. **Fiberoptic bronchoscope intubation**: Can be performed nasally, orally or through an LMA. Limitations: Tube Size (e.g. Storz slimline only fits a >3.5ETT)
6. **Fiberoptic bronchoscope + GlideScope**: The fiberoptic bronchoscope can be used as a guiding bougie for intubation, while the view is provided by the Glidescope. This technique will require two personnel: with one handling the Glidescope while the other manipulating the F.O.B. It is called the video-assisted fiberoptic intubation (VAFI) technique.

Can't Ventilate, can't Intubate…

- **Plan B:** If Plan A fails and ventilation becomes problematic at any step of the airway management, then an LMA should be immediately inserted.
- If "Cannot ventilate" through LMA, then the operator should immediately move to plan C.

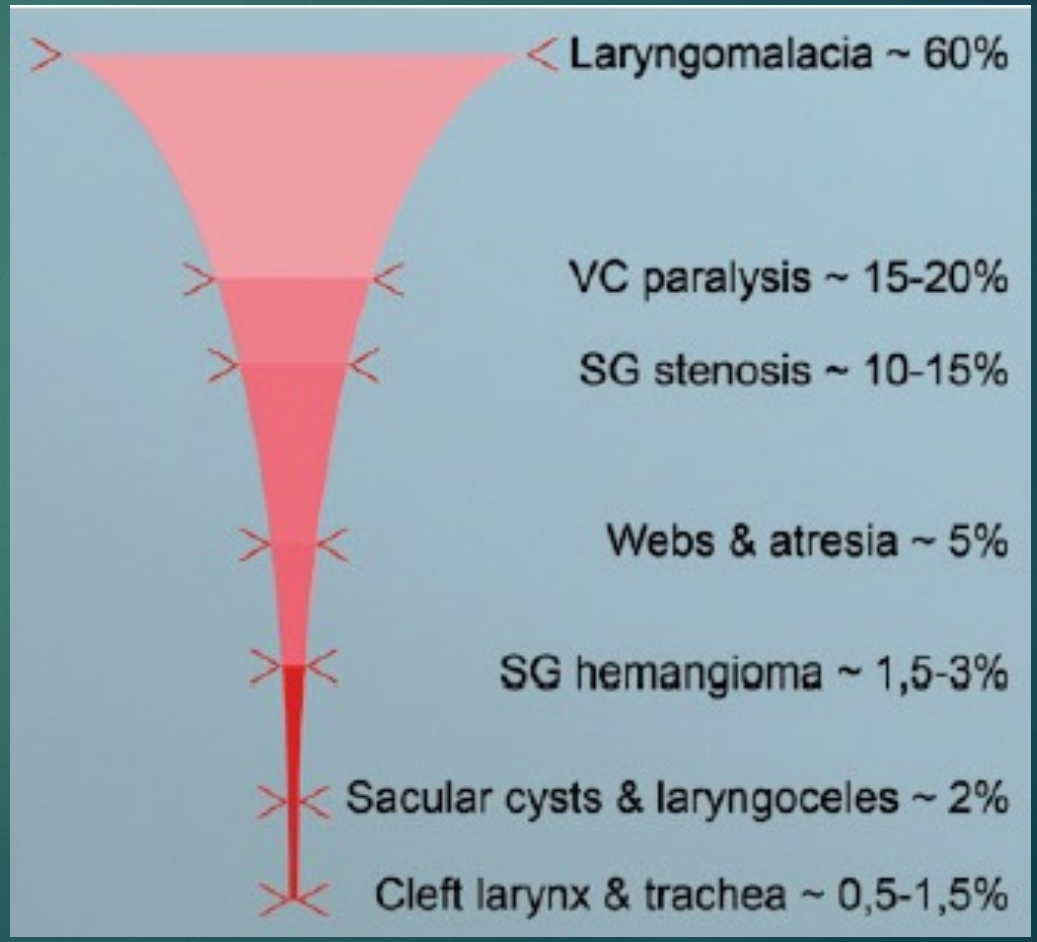
- **Plan C:** If attempts at ventilating via "Bag-Mask" and LMA fail, one attempt at direct laryngoscopy can be performed aiming at intubating if possible.

- **Plan D**: Call ENT ☺
  - Rigid laryngo-bronchoscopy (a 2.7mm scope can fit a 3.0 ETT)
  - Open airway / emergency tracheostomy



Congenital Anomalies of the Larynx and Trachea

- The prevalence of congenital airway anomalies has been estimated to range between 1 in 10,000 and 1 in 50,000 live births



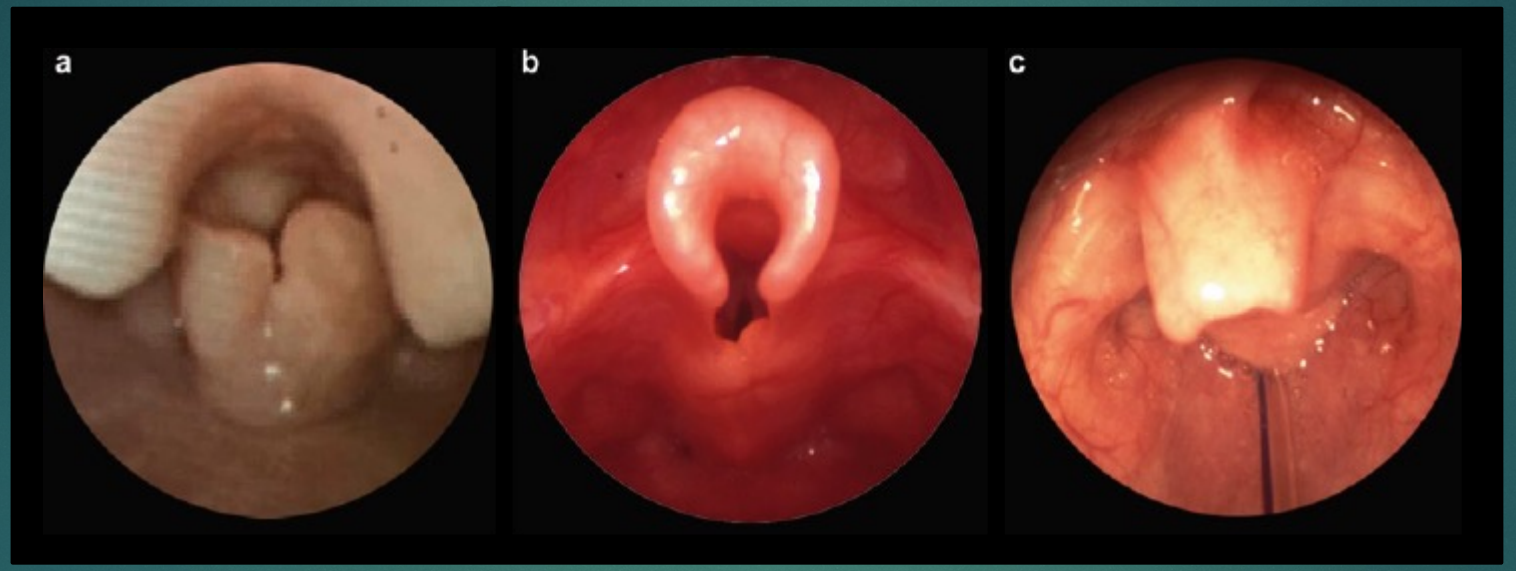
Laryngomalacia

- Most common (~60%) congenital laryngeal anomaly
- Most common source of stridor in newborns
- Male to female ratio 2:1
- **I**nward collapse of supraglottic structures on inspiration
- High-pitched fluttering inspiratory stridor exacerbated by crying, feeding, agitation and supine position.
- Self-limiting condition:
  - Onset: 2–4 weeks after birth
  - Progression: up to 6–8 months after birth
  - Resolution: 18 (range: 12–24) months after birth

Laryngomalacia

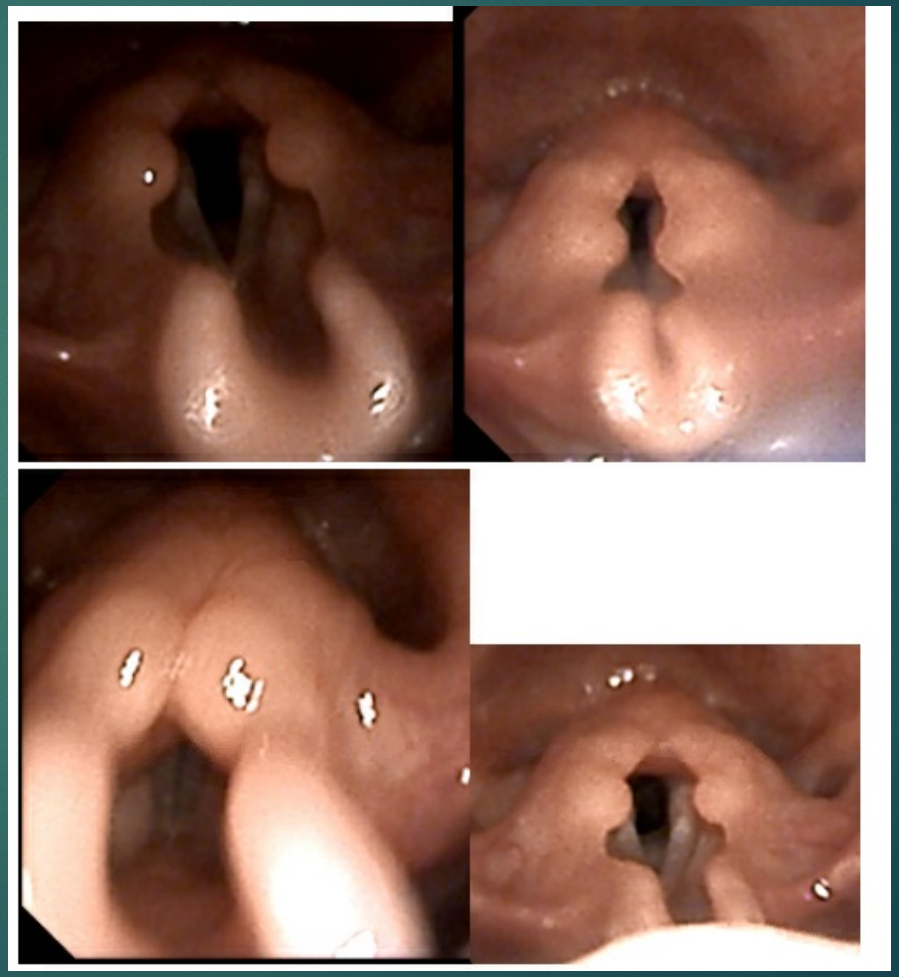
- Diagnosis made using awake transnasal flexible laryngoscopy (TNFL):
  - Three main types of obstruction
- Associated gastro-esophageal reflux in up to 80 % of cases.
- Severity of the disease:
  - Mild to moderate in 80 % of cases
  - Severe in 15 % of cases; supraglottoplasty required
  - Very severe in 1–3 % of cases; tracheotomy required
- Prevalence of synchronous airway anomalies varies widely in published literature
- Severe stridor, feeding difficulties, failure to thrive, obstructive apnea, dyspnea with easy fatigability and severe suprasternal or intercostal retractions warrant a surgical intervention

Types of laryngomalacia



- Type I: inward collapse of the aryepiglottic folds.
- Type II: tubular epiglottis with short aryepiglottic folds.
- Type III: retroflexed epiglottis with prolapse into the laryngeal inlet

The noisy breather…

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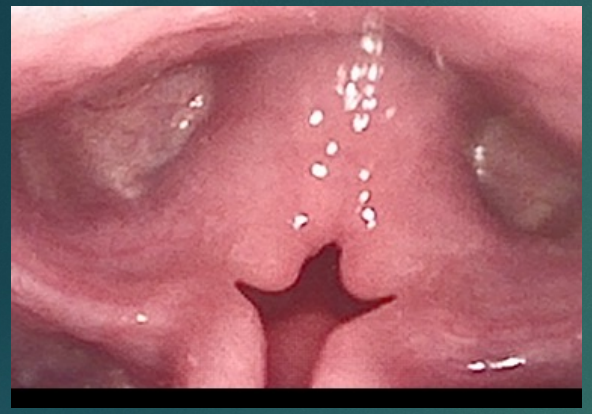
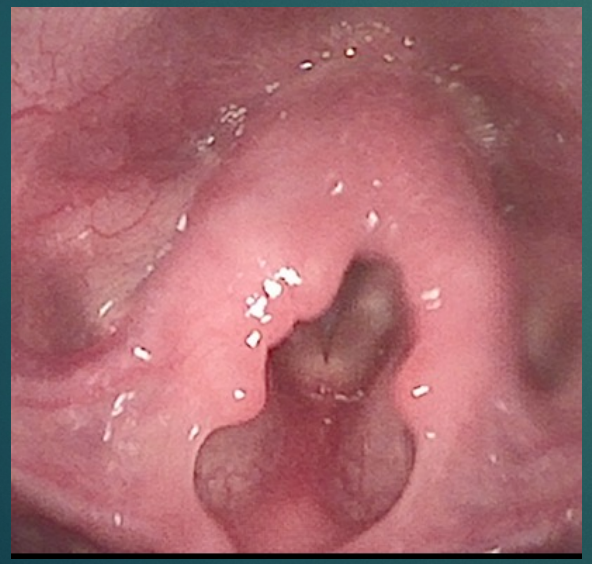
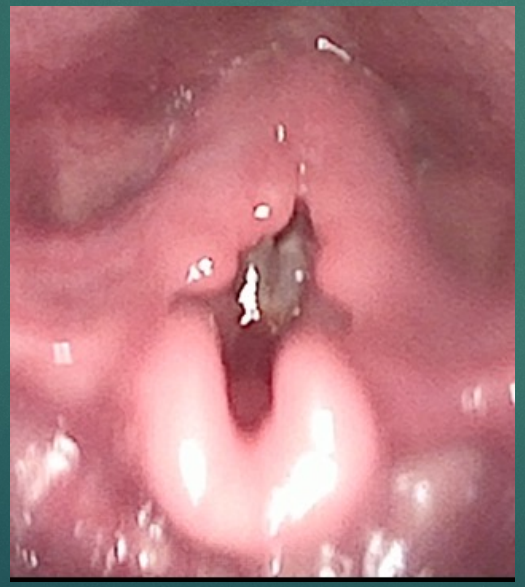
Laryngomalacia – Typical Findings







- Redundant arytenoid tissue/mucosa with mild prolapse and hooding over the TVC's during inspiration
- Shortened AE folds and omega shaped epiglottis
- (!) Awake fiberoptic flexible laryngoscopy (FFL):
  - In nearly all cases, the infant is crying during exam
  - Can miss mild LM or lead to overdiagnosis in the patient with a normal airway

Laryngomalacia and Comorbidities

- Incidence of **secondary lesions** varies from 8% to 58% (e.g. vocal fold paresis or paralysis, type 1 laryngeal cleft, tracheomalacia and subglottic stenosis)

- Gastroesophageal reflux disease (**GERD**) is a well-established comorbidity of laryngomalacia and many patients with laryngomalacia have symptoms of and are treated for reflux.

- Given that swallowing interrupts breathing, infants with airway compromise or respiratory distress may not be able to safely coordinate sucking, swallowing, and breathing, leading to **dysphagia**, **penetration** and **aspiration** (Symptoms present in >50% of patient with LM)

Laryngomalacia and Dysphagia

Published in final edited form as:
Laryngoscope. 2016 February ; 126(2): 478–484. doi:10.1002/lary.25440.

Laryngomalacia and Swallowing Function in Children

Jeffrey P. Simons, MD[1], **Laura L. Greenberg, MA**[1], **Deepak K. Mehta, MD**[1], **Anthony Fabio, PhD, MPH**[2], **Raymond C. Maguire, DO**[1], and **David L. Mandell, MD**[3]

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[3]Center for Pediatric ENT-Head and Neck Surgery, Boynton Beach, FL, U.S.A

- **75.7%** of patients had at least one abnormal pretreatment swallowing assessment (CSE, MBS, or FEES).

International Journal of Otolaryngology

Int J Otolaryngol. 2010; 2010: 912814.
Published online 2010 Nov 21. doi: 10.1155/2010/912814

PMCID: PMC2989454
PMID: 21113300

Aspiration before and after Supraglottoplasty regardless of Technique

Jeffrey C. Rastatter, [1, 2], James W. Schroeder, Jr., [1, 2, *] Stephen R. Hoff, [3] and Lauren D. Holinger [1, 2],

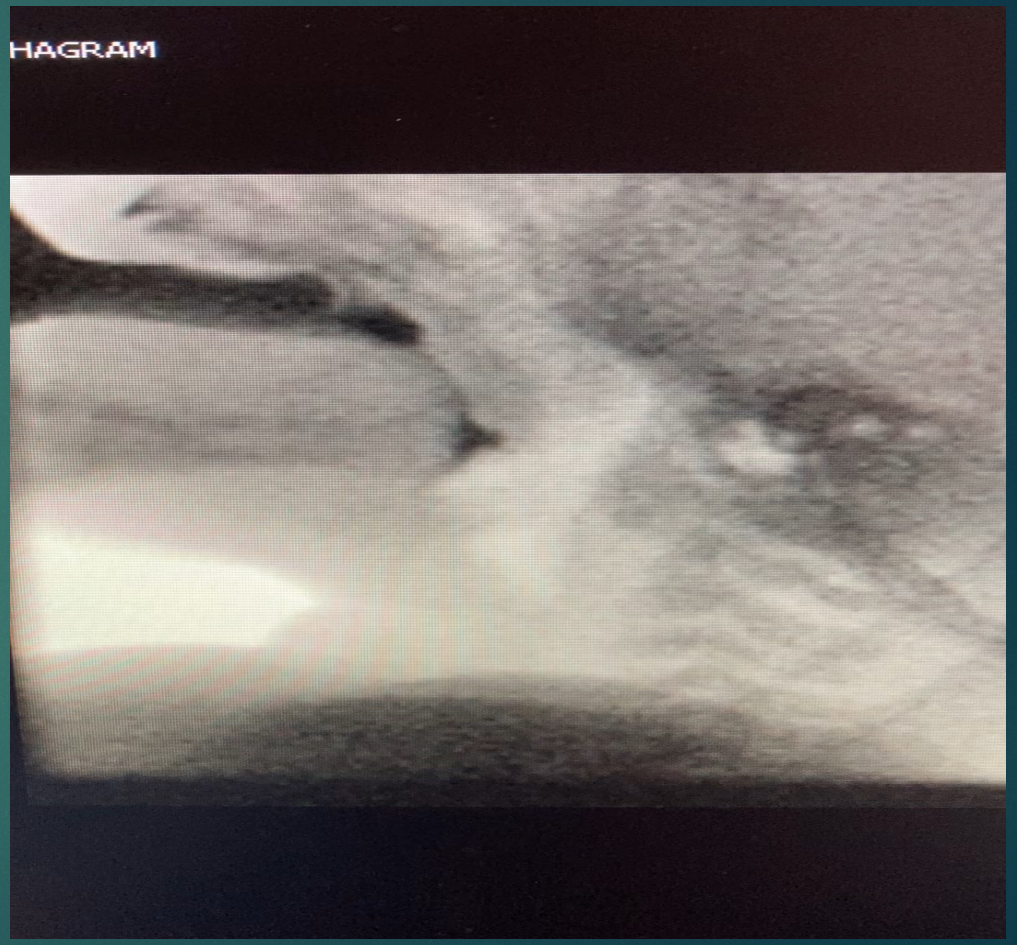
- **44.8%** developed aspiration after supraglottoplasty

Potential Therapeutic Interventions

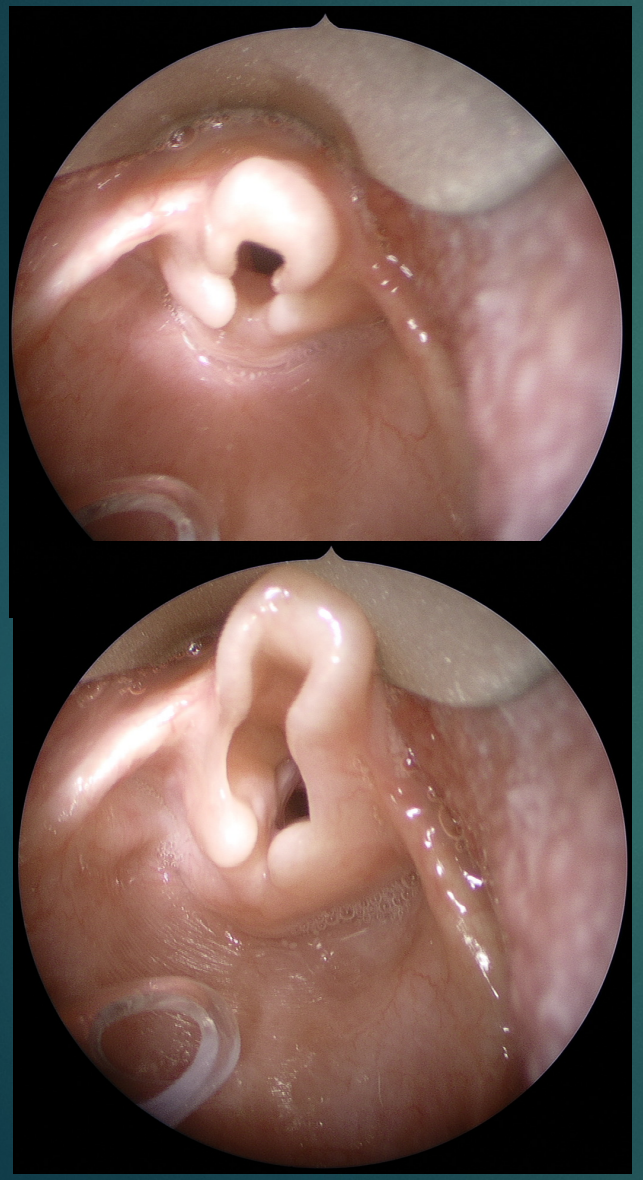
- Positioning
- Paced Techniques- Cue based feeding
- Appropriate flow rate and nipple shape
- Last resort: Thickening

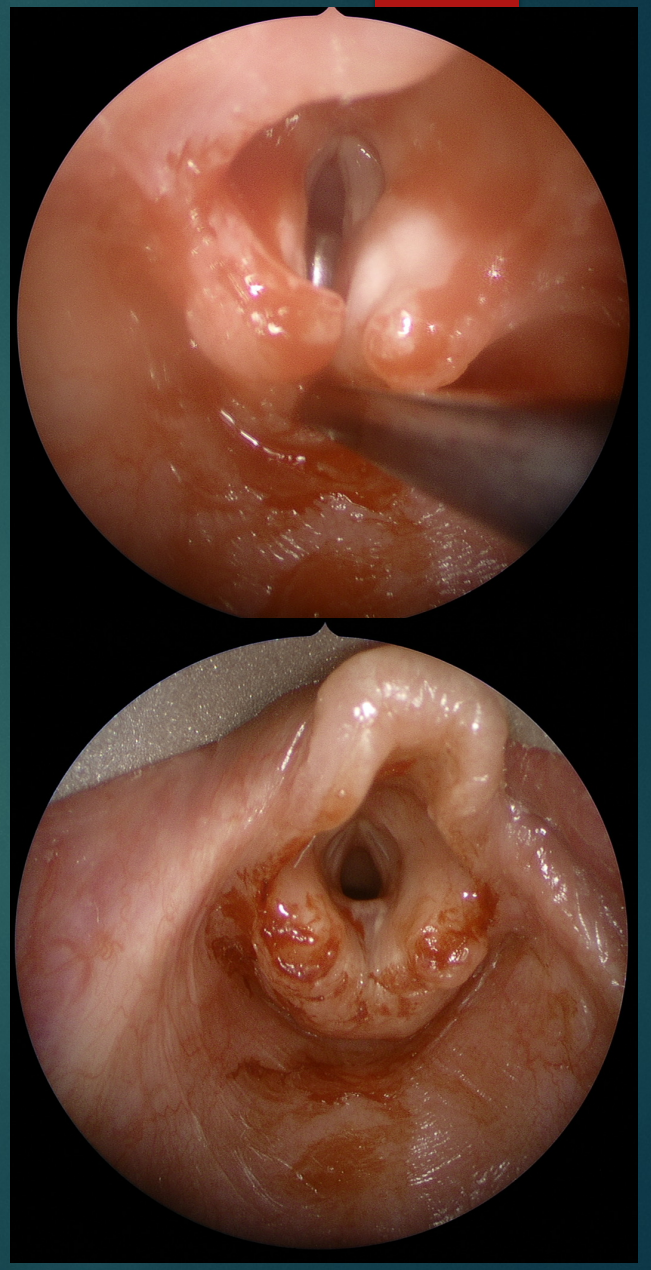
Indication for Surgical Intervention

- Severe stridor with:
  - Compromised airway
  - Feeding difficulties, FTT
  - OSA
- Dyspnea with severe suprasternal retractions, hypoxemia and hypercapnea warrant immediate surgical intervention
- Chest deformity, pulmonary hypertension and cor pulmonale are late signs of chronic obstruction



Supraglottoplasty





Complications & Aftercare

- Success rate: 69 – 94%
  - Results are influenced by severity of LM and presence of comorbidities
- Supraglottic stenosis (~4%)

Pearls in Stridor and LM

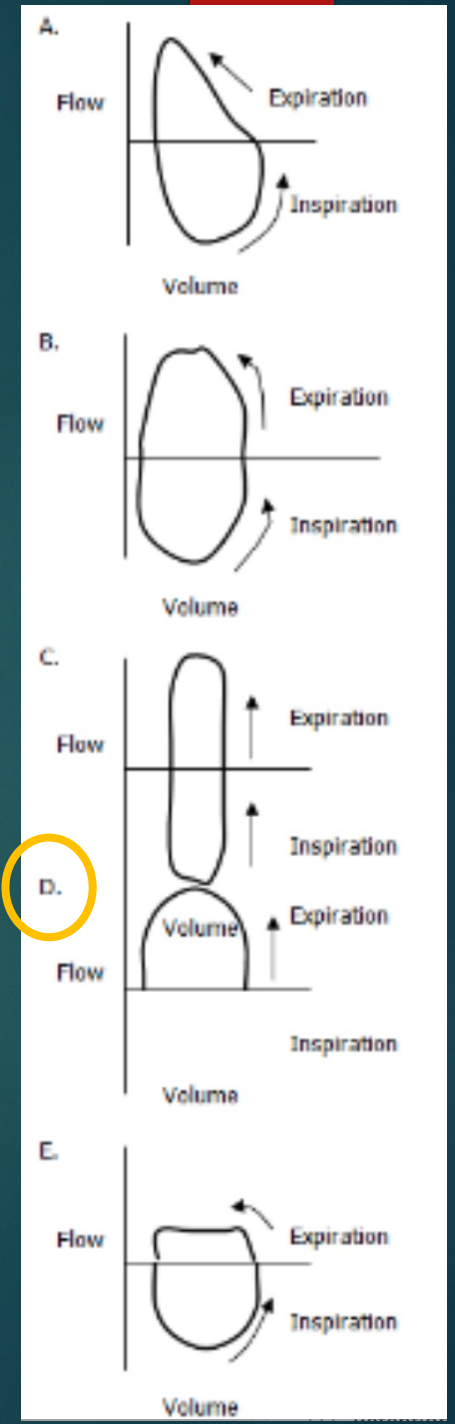
- **Take a proper history – it matters, and you have time**
- **Flexible laryngoscopy in a neonate/infant with stridor is easy to do but harder to interpret than most of us think**
- **Laryngomalacia is most common cause of stridor**
- **Up to 20% require surgical intervention (SGP)**
  - Indicated in FTT, apneas/OSA, cor pulmonale, pulmonary HTN, pectus, severe dysphagia
  - Symptom resolution in 70-100%
- **Comorbidities are important factors affecting the outcome**
- **Swallowing dysfunction is common regardless of disease severity**
  - Swallowing studies are frequently abnormal in LM patients presenting both with and without subjective dysphagia
  - Treatment of LM is associated with improvement in dysphagia
- **Multidisciplinary team management to optimize outcome**
- The stridor of mild LM often improves with crying, as tone in the pharynx is increased; conversely, in moderate to severe LM, the stridor typically will worsen with crying because of the increased airflow through the severely collapsed larynx

Goals of Laryngomalacia Treatment

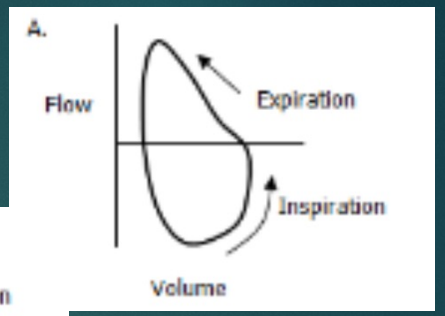
Primary Goals Laryngomalacia Management	Improve respiration
	Improve feeding
	Reduce the risk of aspiration
	Overcome OSA
Intangible Goals	Reduce parental anxiety
	Reduce hospitalization and visits (socioeconomic)
	Improve family quality of life
	Reassurance
	Quicker path to normal

Case #1

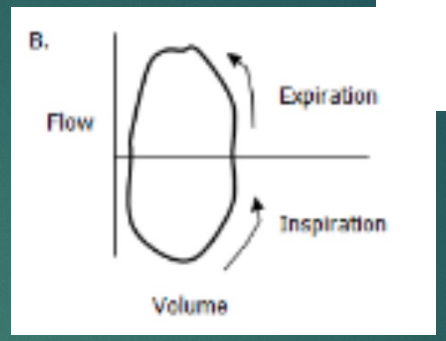
- A male infant was born at 38 weeks gestation with a birthweight of 3.1 kg. His clinical course was complicated by a diagnosis of laryngomalacia. What is the most accurate representation of the flow-volume loop representing his disease process?



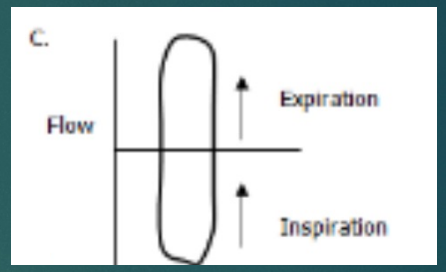
- The flow-volume loop demonstrated in Option A is consistent with obstructive lung disease.



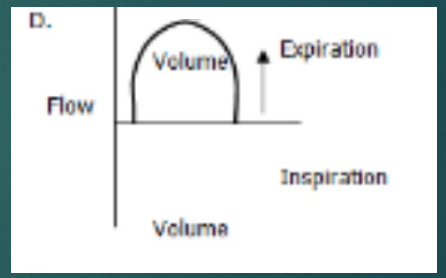
- A normal flow-volume loop is shown in Option B.



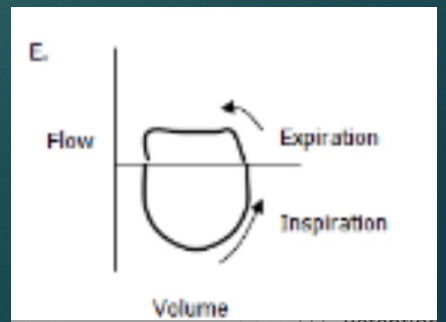
- Restrictive lung disease is found in infants with meconium aspiration and the representative flow-volume loop is represented in Option C.



- The flow-volume loop of an extrathoracic upper airway obstruction, such as vocal cord paralysis and laryngomalacia, is shown in Option D.

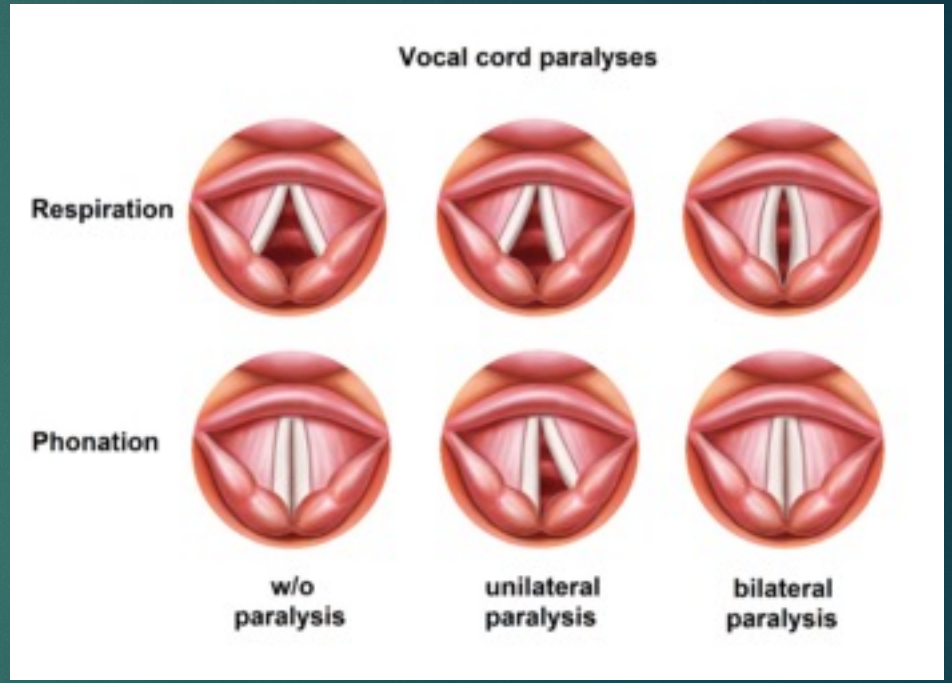


- Option E represents a flow-volume loop of an infant with intrathoracic upper airway obstruction, such as tracheomalacia and vascular rings.



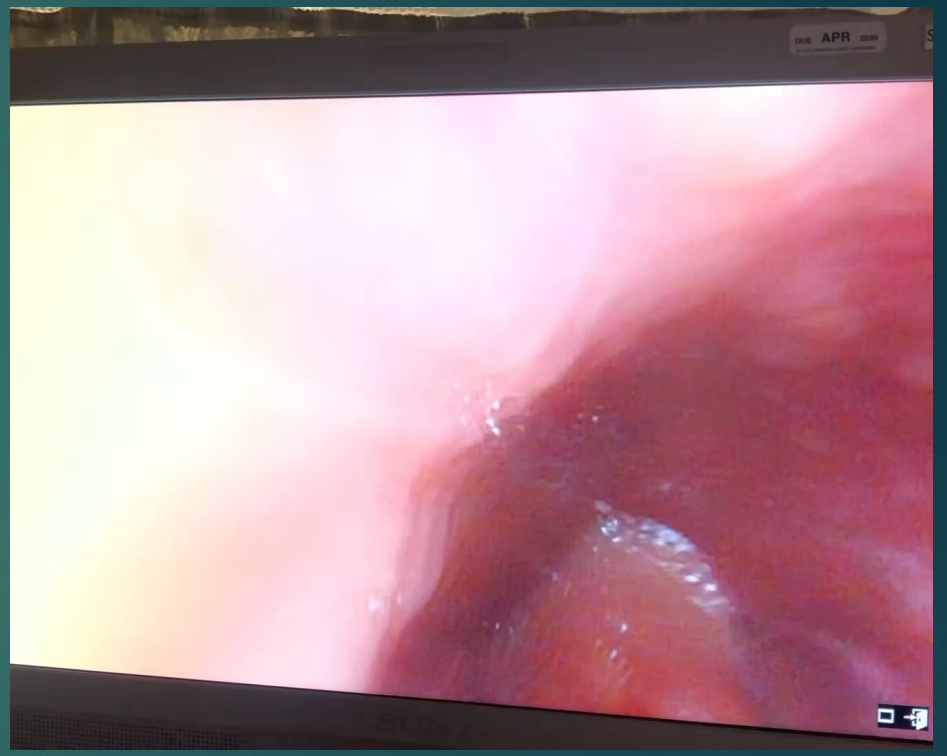
Vocal Cord Paralysis (VCP)

- Second most common (15–20%) congenital laryngeal anomaly.
- With the exception of traumatic sections of the RLN(s), VCP is caused by dysfunctional laryngeal innervations without complete muscular flaccidity or denervations.
- Associated upper airway pathological conditions are observed in 45% of the cases.



Unilateral VCP

- Prevalence: ~48% of all VCPs
- Mild stridor with hoarse, breathy cry and feeding difficulties (aspiration)
- Main etiology: **injury to the peripheral nervous system**
- Most frequent causes: cardiovascular (~50%) and esophageal surgeries for congenital mediastinal anomalies
- Tendency for natural voice improvement
- Watchful follow-up with no invasive treatment
- Tracheotomy rarely needed (~8% of the cases)

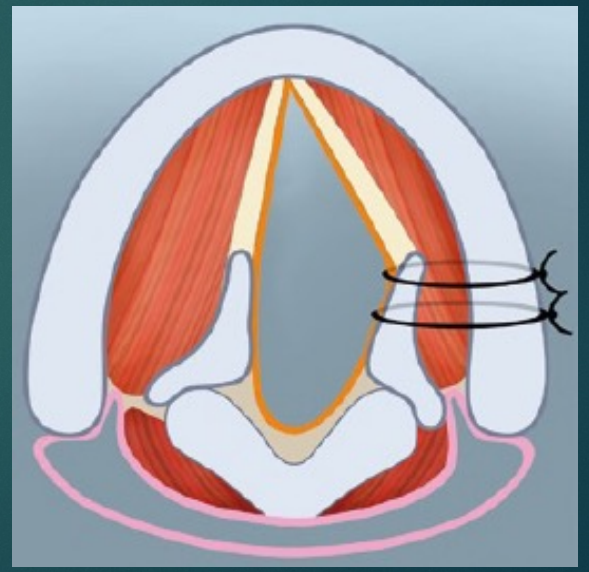


Bilateral VCP

- Prevalence: ~52% of all VCPs.
- High-pitched stridor with dyspnea, apneic spells and cyanosis but normal voice.
- Main etiologies: congenital disorders of the central nervous system, traumatic and idiopathic causes (e.g. Arnold-Chiari Malformation).
- Neurogenic BVCP must be differentiated from cicatricial posterior glottic stenosis (PGS).
- Tracheotomy is required in approximately 53% (50–65%) of the cases.
- Forty-six to sixty-four percent of all children affected by BVCP recover spontaneously during the first 6–12 months of life and up to 10% after the age of 5 years.
- Watchful follow-up until 2 years of age, before any surgical intervention is planned. The multitude of surgical options reflect an absence of consensus regarding treatment.
- The least invasive treatment is preferred.

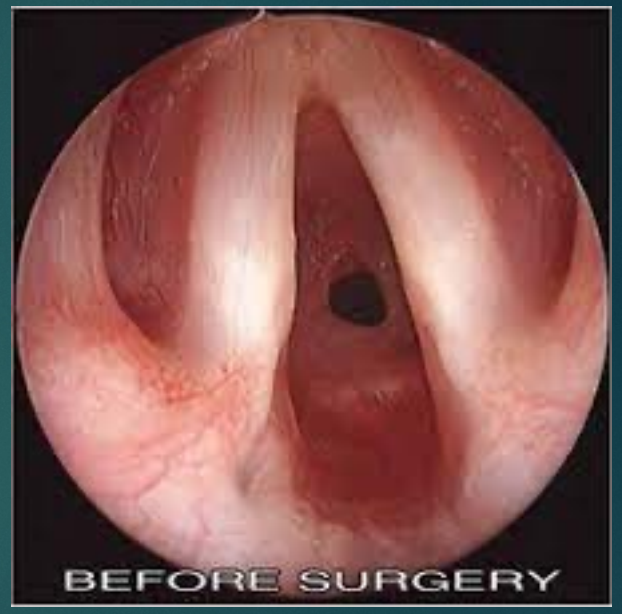
Surgical Treatment for BVCP

- **Open** surgery:
  - Arytenoidopexy using the lateral approach
  - Arytenoidectomy ± lateralisation through laryngofissure or lateral approach
  - Arytenoid separation by posterior cricoid split and cartilage grafting
- **Endoscopic** surgery:
  - laser arytenoidectomy
  - CO2 laser posterior cordotomy
  - Arytenoidopexy with the Lichtenberger needlecarrier
  - Posterior cricoid split with cartilage grafting

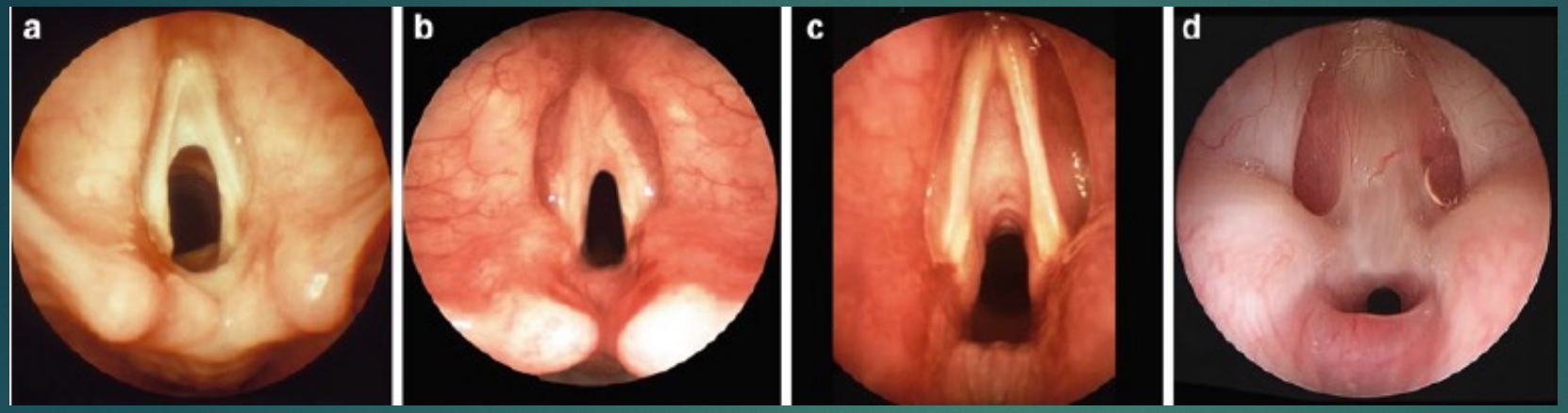


Congenital Subglottic Stenosis

- Third most common (10–15%) congenital laryngeal anomaly.
- **Most common laryngeal anomaly requiring tracheotomy in children under 1 year of age**.
- Defined as subglottic diameter less than 4.0 mm in a full-term neonate and 3.0 mm in a premature baby.
- Results from incomplete recanalisation of the laryngeal lumen during the tenth week of gestation.
- Belongs to the spectrum of laryngeal webs and atresia.
- Manifest when subglottic stenosis (SGS) shows more than a 50% luminal diameter restriction.
  - Primary biphasic stridor.
  - Recurrent or prolonged croup, barking cough.



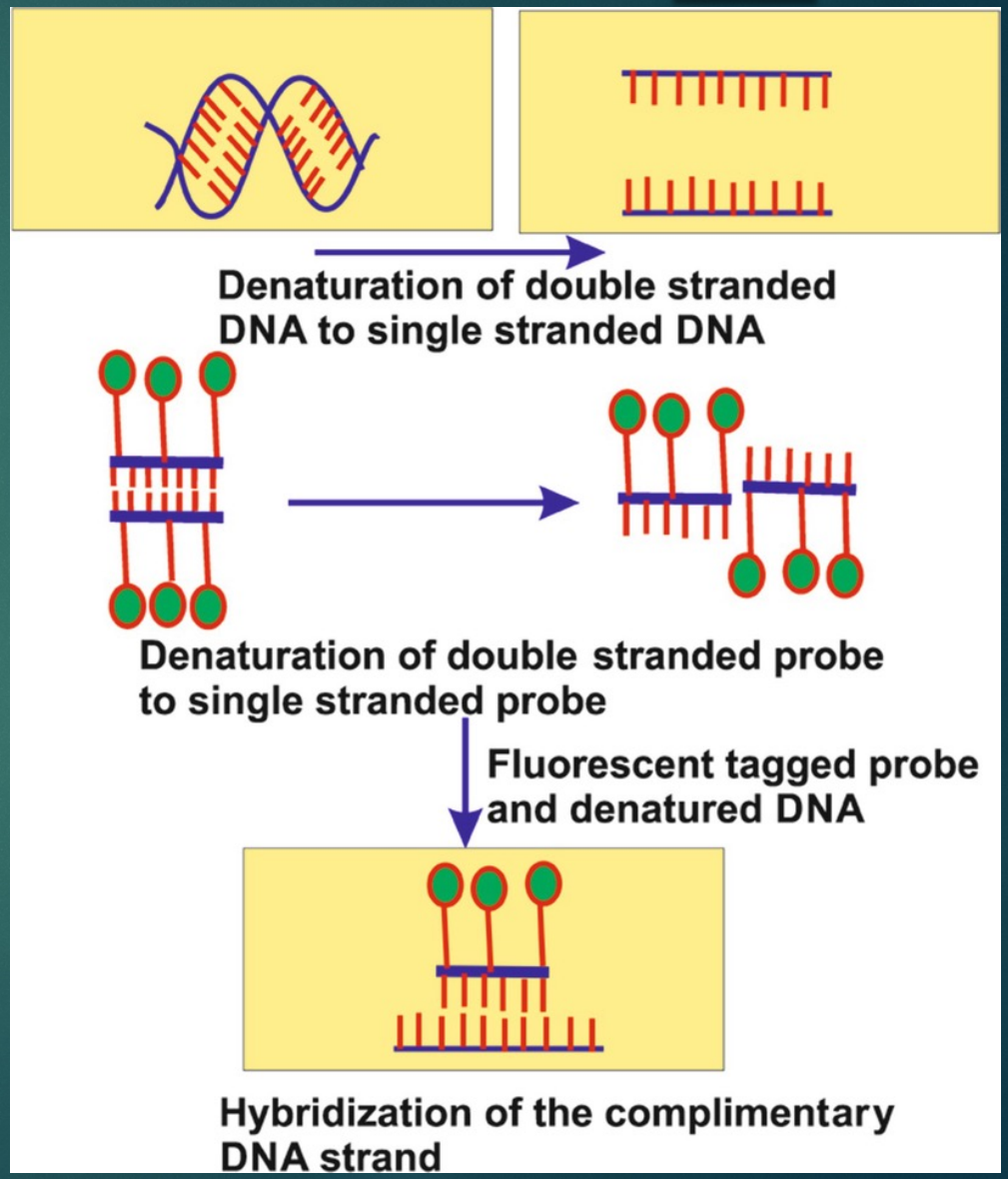
Laryngeal Web / Atresia



- Rare; typically. diagnosed in the neonatal period after an investigation for the source of aphonia or stridor
- Can extend into the subglottic area
- Association with (up to 65%): **22q11.2 (DiGeorge, Velocardiofacial Syndrome)**

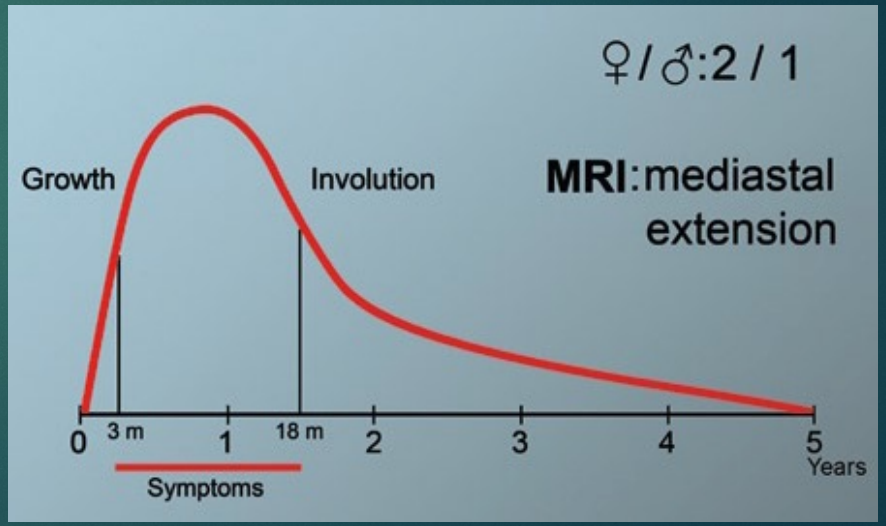
Fluorescent in situ hybridization (FISH)

- The double-stranded DNA is at first converted into single-stranded DNA, and then subsequently a fluorescent-tagged probe is used to visualize the target DNA part.



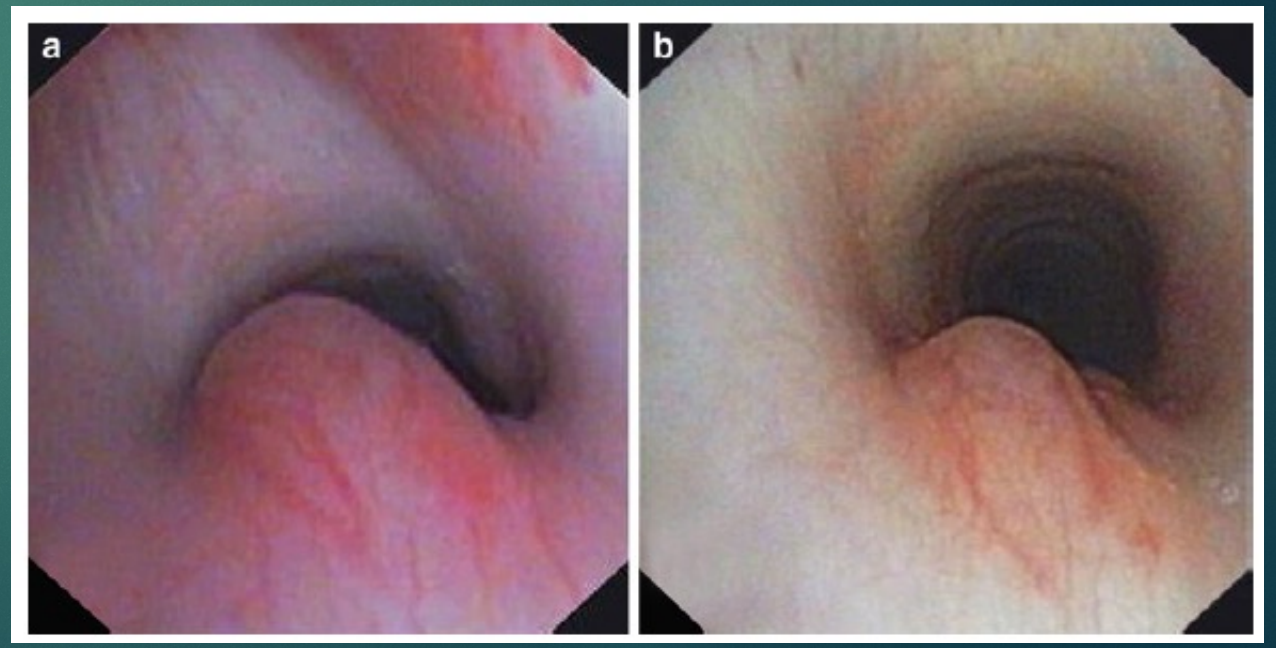
Subglottic Hemangioma

- Rare
- Benign vascular tumor characterized by hyperplasia of endothelial cells, pericytes, mast cells, fibroblasts and macrophages
- Associated in 50% of the cases with cutaneous haemangiomas, most frequently with a '**beard' distribution** (chin, lower lip, and anterior neck)
- Female predominance (2–3:1)
  - Typical evolution:
    - — Rapid proliferative phase between 4–6 weeks and 4–10 months of life
    - — Stabilisation as of 10–12 months of life
    - — Slow involution phase between 10–12 months and 5–10 years of age



Subglottic Hemangioma

- Clinical course:
  - No symptoms during the first weeks of life
  - Beginning of symptoms between 2 and 4 months of age
  - All infants are symptomatic by 6 months of age
  - Progressive resolution between 12 and 18 months of age
  - Complete resolution by 5–12 years of age
- Symptoms:
  - Biphasic, predominantly inspiratory stridor
  - Barking cough
  - Recurrent or prolonged ‘croup’
  - Voice altered to varying degrees
- Treatment:
  - Propranolol, 2mg/kg/day



Case #2

You are called to see a 3-hour old term male infant born vaginally with Apgar scores of 8 at 1 minute and 9 at 5 minutes. The infant has episodes of respiratory distress and cyanosis at rest. Both of these symptoms resolve during crying. The infant does not have a murmur and the rest of his exam is normal. All of the following are true about the infant's likely diagnosis **EXCEPT**:

A. 2/3 of cases are unilateral

B. An oral airway may provide temporary improvement

C. Females are affected more than males at a 2:1 ratio

D. Surgical repair can be delayed depending on severity

E. The infant's diagnosis is not typically associated with other congenital abnormalities

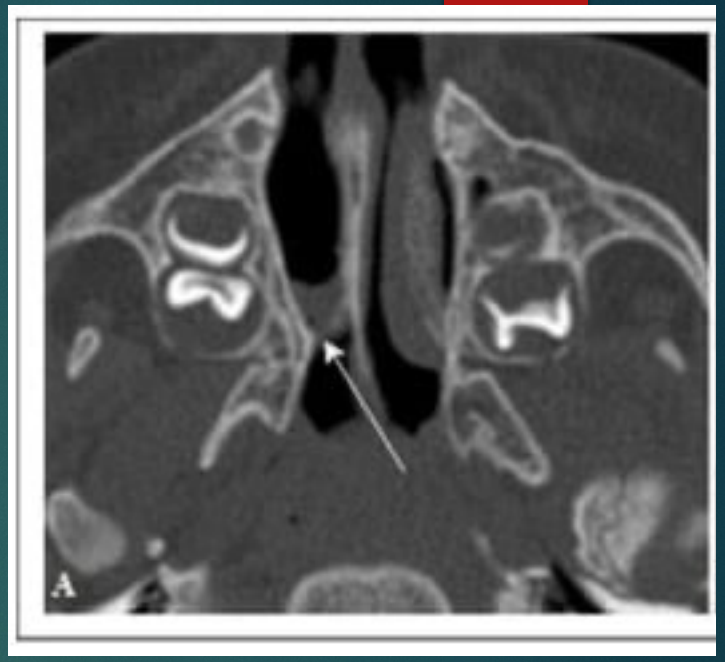
Case #2

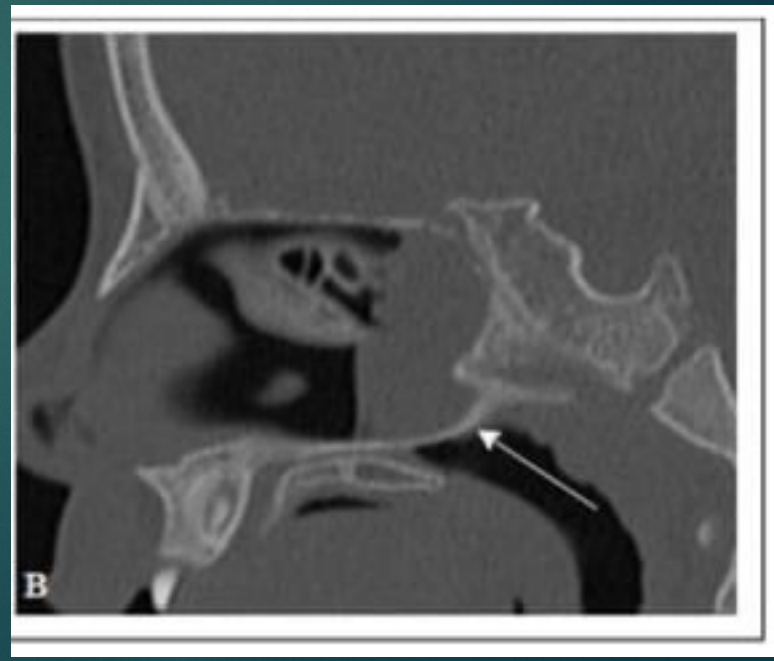
- Answer E.
- The infant's diagnosis **is** typically associated with other congenital abnormalities.

  The infant described in this vignette most likely has choanal atresia.

Choanal Atresia

- Choanal atresia is a rare congenital disorder caused by a failure to develop the posterior nasal cavity (choana), resulting in a missing opening between the nasopharynx and the nasal cavities.
- The incidence of this malformation reported in the literature is approximately 1/5000–7000 live births.
- Generally, choanal atresia may affect one or both choanae
- Unilateral form is more common
- Incidence is higher in females than in males (2:1)
- The unilateral form is frequently on the right side
- Bilateral atresia is more commonly associated with syndromes such as CHARGE, Treacher-Collins, and Crouzon's disease.
- Bilateral atresia is a congenital disease that represents a surgical emergency. It is severe because the infant, during the first month of life, breathes preferentially through the nose.





Case / Question #3

During fetal development, a **cleft lip** results when the maxillary process fails to fuse normally with which of the following processes?

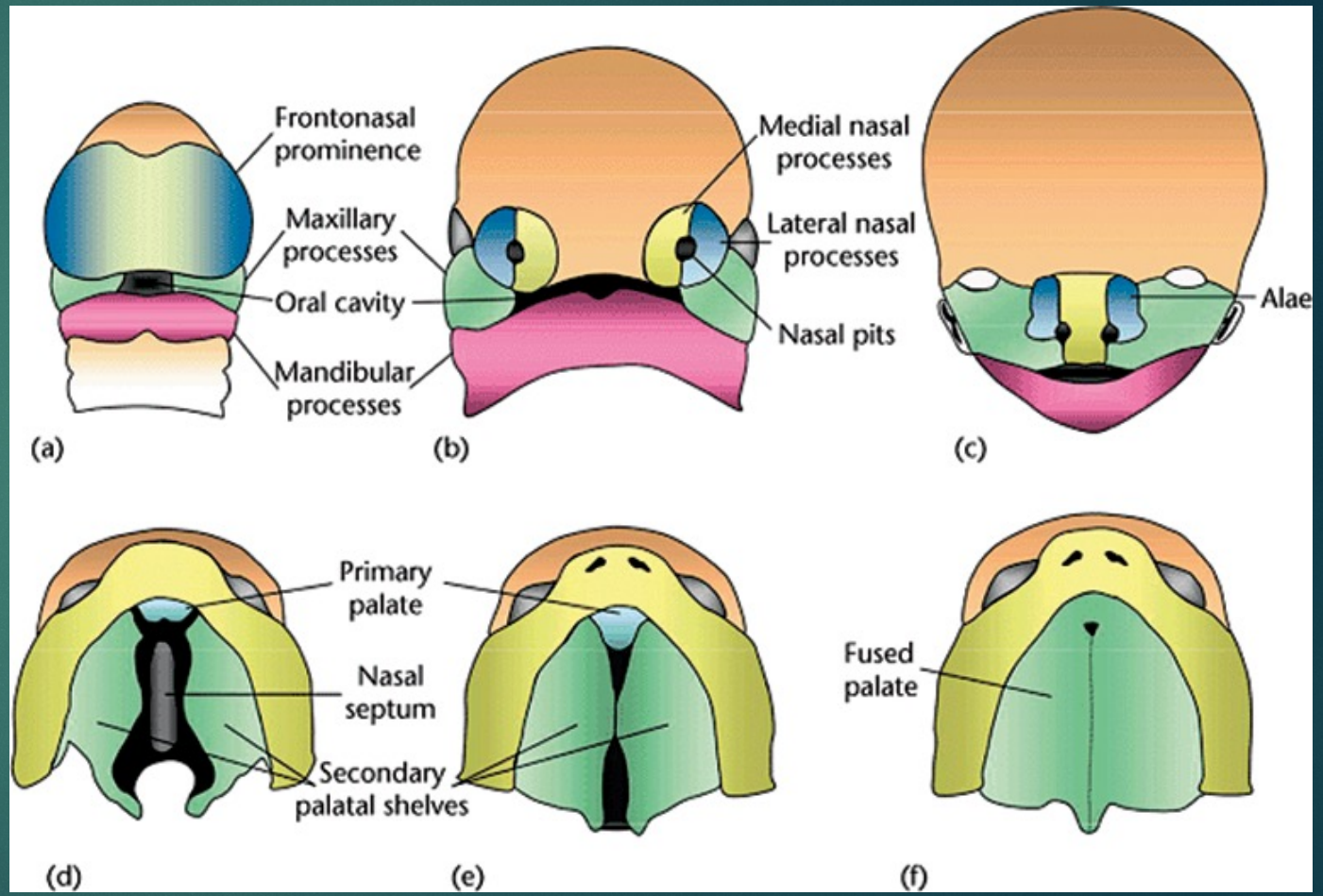
A. Lateral nasal

B. Frontonasal

C. Medial nasal

D. Mandibular

- **Answer C.** Medial nasal.
- The developing fetus has five facial prominences that are populated by neural crest cells: frontonasal, paired maxillary, and paired mandibular.
- The frontonasal prominence gives rise to the nasal pit or placode, around which develops the medial and lateral nasal processes. A failure of fusion between the maxillary prominence and the medial nasal process results in a common cleft of the lip.



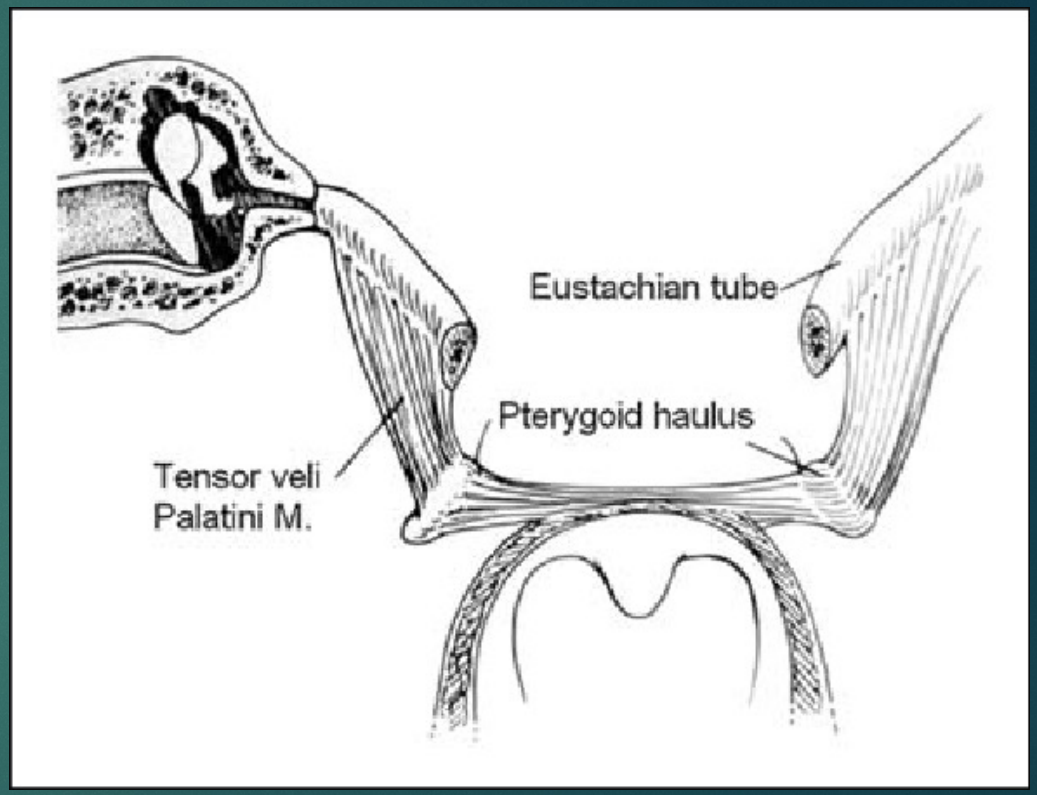
Cleft Lip / Palate

- Both genetic and environmental factors have been shown to influence the risk of CL/P and CPO (isolated CP).
- Approximately 70% of all cases of CL/P and 50% of cases of CPO are designated as nonsyndromic.

Cleft type	Syndro				Gene
Cleft lip +/– cleft palate	Autosom	Cleft palate only	Oculofaciocardiodental	*BCOR*	*ACTB*
	Familial		CHARGE	*CHD7*	*CDH1*
	Craniofr		Lethal and Escobar multiple pterygium	*CHRNG*	*EFNB1*
	Roberts		Stickler type 1	*COL2A1*	*ESCO2*
	Holopros		Stickler type 2	*COL11A1*	*GLI2*
	"Oro-fac		Stickler type 3	*COL11A2*	*GLI3*
	Hydrolet		Desmosterolosis	*DHCR24*	*HYLS1*
	Van der V		Smith-Lemli-Opitz	*DHCR7*	*IRF6*
	X-linked		Miller	*DHODH*	*PHF8*
	Gorlin		Craniofrontonasal	*EFNB1*	*PTCH1*
	CLP—ec		Kallmann	*FGFR1*	*PVRL1*
	Holopros		Crouzon	*FGFR2*	*SHH*
	Holopros		Apert	*FGFR2*	*SIX3*
	Branchio		Otopalatodigital types 1 and 2	*FLNA*	*TFAP2A*
	Holopros		Larsen syndrome; atelosteogenesis	*FLNB*	*TGIF*
	Ectrodac		Hereditary lymphedema-distichiasis	*FOXC2*	*TP73L*
	Ankylob		Bamforth-Lazarus	*FOXE1*	*TP73L*
	Tetra-amelia with CLP		"Oro-facial-digital"	*GLI3*	*WNT3*
			Van der Woude/popliteal pterygium	*IRF6*	
			Andersen	*KCNJ2*	
			Kabuki	*MLL2*	
			Cornelia de Lange	*NIPBL*	
			X-linked mental retardation	*PQBP1*	
			Isolated cleft palate	*SATB2*	
			Diastrophic dysplasia	*SLC26A2*	
			Campomelic dysplasia	*SOX9*	
			Pierre Robin	*SOX9*	
			DiGeorge	*TBX1*	
			X-linked cleft palate and ankyloglossia	*TBX22*	
			Treacher Collins	*TCOF1*	
			Loeys-Dietz	*TGFBR1*	
			Loeys-Dietz	*TGFBR2*	
			Saethre-Chotzen	*TWIST1*	

Cleft Lip/Palate and ENT

- Children with cleft palate are at increased risk for eustachian tube dysfunction.
- Up to 90% of infants born with CLP suffer from OME before their first birthday
- Children with cleft palate receive on average of approximately 3 sets of tympanostomy tubes.
- The majority requires another otologic or upper airway surgery (e.g. T&A).



> Cleft Palate Craniofac J. 2018 May;55(5):743-746. doi: 10.1177/1055665617752210.
Epub 2018 Jan 23.

Otolaryngology Service Usage in Children With Cleft Palate

Kenneth R Whittemore Jr [1 2], Jenna M Dargie [1], Briana K Dornan [1], Brian Boudreau [1]

Affiliations + expand

PMID: 29360407 DOI: 10.1177/1055665617752210

Inherited Craniofacial Disorders

A 3-year-old boy presents with delayed milestones of development. He has not yet started running, and his speech output is also not normal compared to his peers. On physical examination, the child is brachycephalic, has an increased distance between his eyes, has a tongue that he struggles to keep in his mouth, and his nose appears small and set back from the plane of his face. Extremity examination reveals flat, wide thumbs and mild webbing of the toes. There is a history of tracheostomy performed in the neonatal period. Which of the following conditions might have necessitated the tracheostomy?

- A. Congenital tracheal cartilaginous sleeve
- B. Respiratory distress syndrome
- C. Cystic fibrosis
- D. Vocal cord paralysis

Inherited Craniofacial Disorders

Acrocephalosyndactyly syndromes involving premature closing of the cranial sutures are:

- Type I: Apert syndrome
- Type II: Vogt/Crouzon syndrome
- Type III: Saethre–Chotzen syndrome
- Type IV: Waardenburg syndrome
- Type V: Pfeiffer syndrome

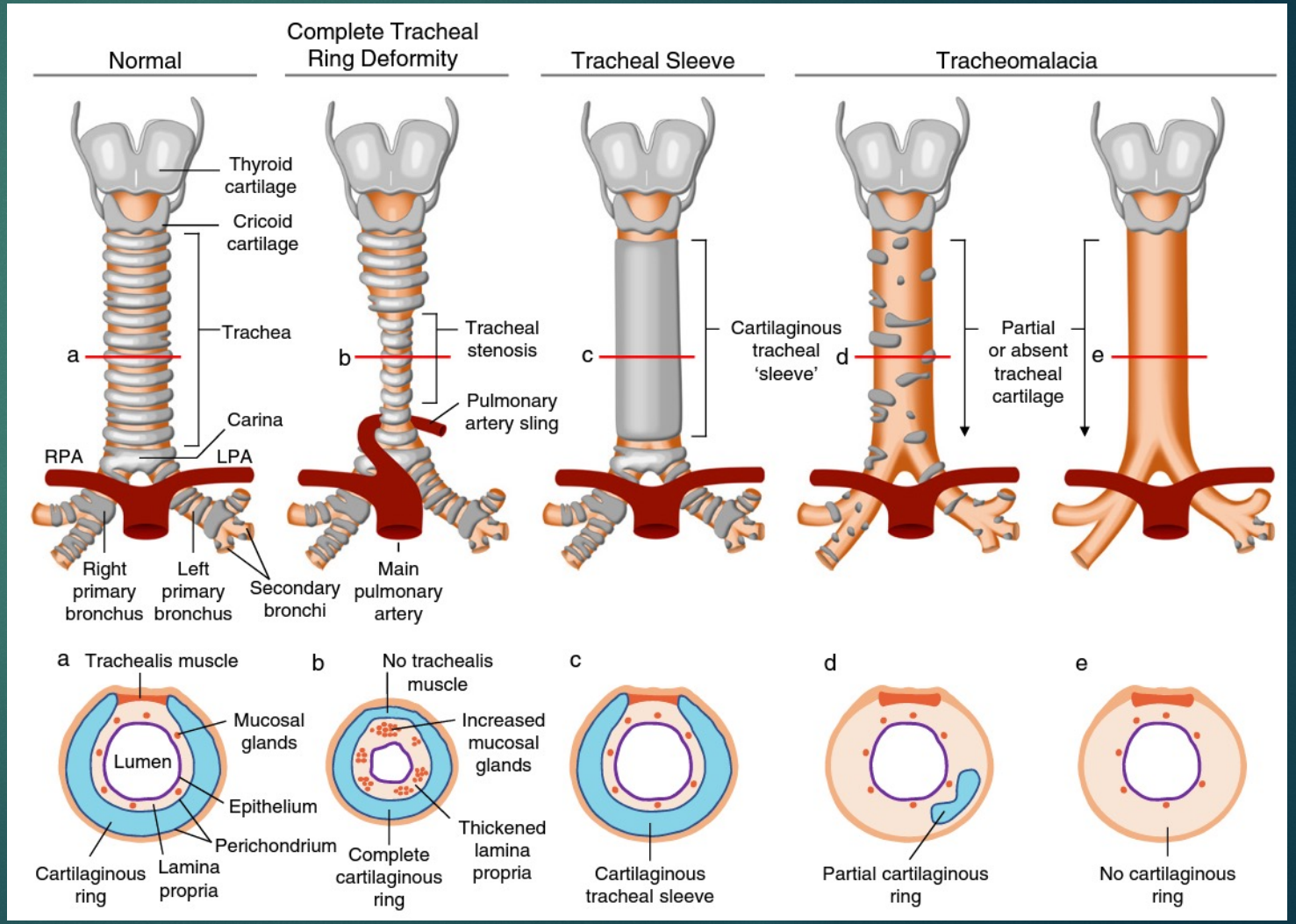
- Caused by mutation in fibroblast growth factor receptor (FGFR) genes
- Most common is Apert Syndrome, followed by Pfeiffer Syndrome

Pfeiffer Syndrome

- The incidence of Pfeiffer syndrome is approximately 1 in 100 000 worldwide
- Dominant genetic disorder, which is also known to be a part of a condition caused by mutation in fibroblast growth factor receptor (FGFR) genes.
- Clinical features: craniosynostosis, midface hypoplasia, and limb anomalies. Broad thumbs and great toes are often associated with partial syndactyly of the second and third fingers, as well as second and third toes in Pfeiffer syndrome.

Congenital Tracheal Cartilaginous Sleeve

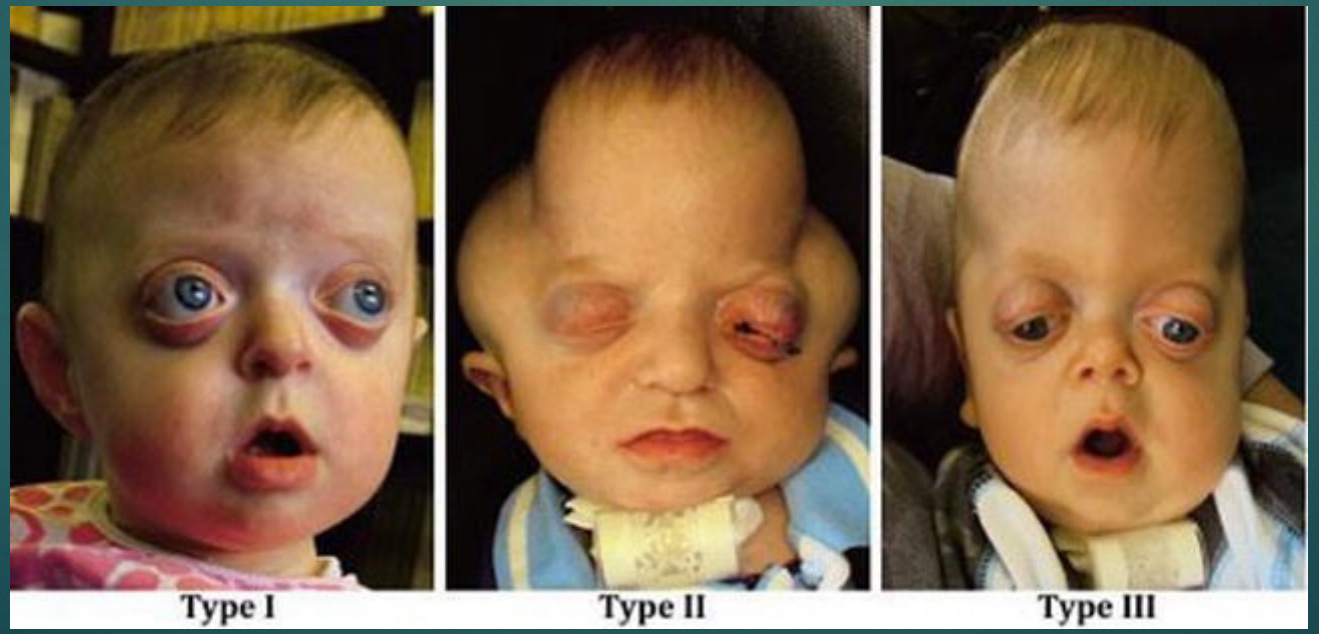
- = Rare congenital airway malformation in which there is a continuous tracheal cartilaginous cylinder instead of discrete cartilaginous rings.
- TCS is almost always diagnosed postmortem. It is usually discovered incidentally via endoscopic examination while trying to establish causes of persistent respiratory difficulties, direct visualization of the trachea during tracheostomy or at autopsy



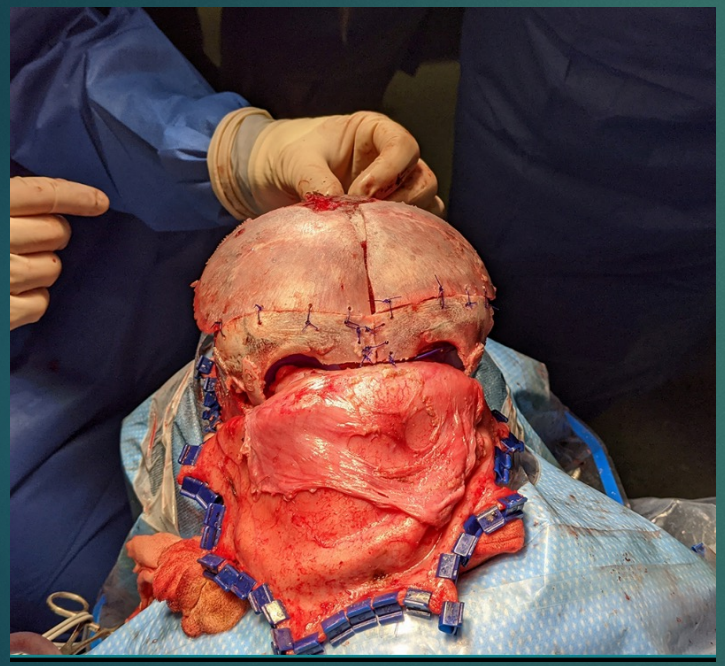
Pfeiffer Syndrome

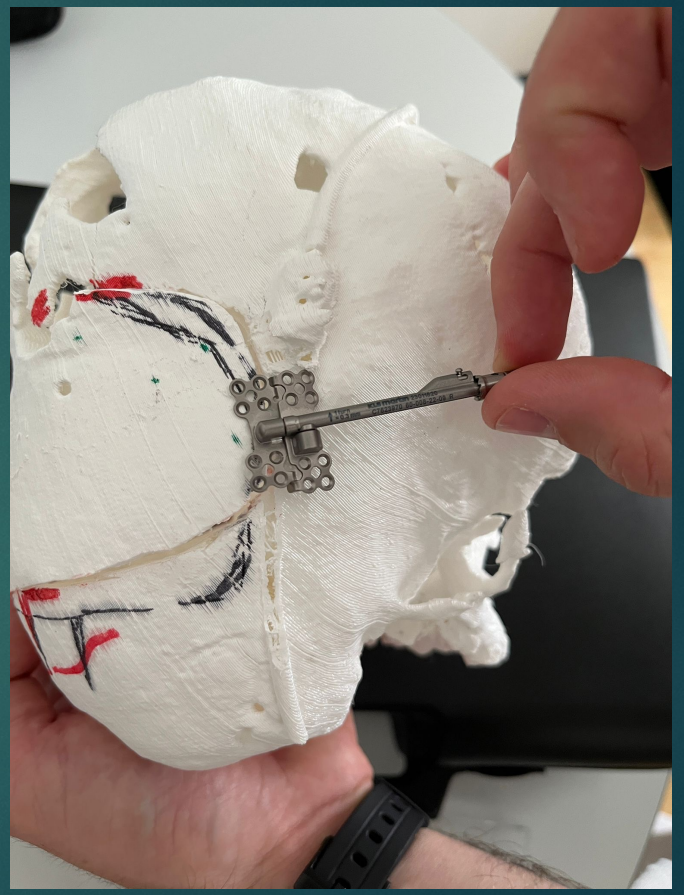
Based on a classification developed by Dr Cohen in 1993, the three types of Pfeiffer syndrome are defined as:

- Type 1: mild expression of the classic findings
- Type 2 and type 3 are more severely affected, characterized by severe ocular proptosis and midfacial hypoplasia, with type 2 including the cloverleaf skull deformity, which is not present in type 3.
- In type 2 and type 3 infants, it is often associated with hydrocephalus, abnormal fixation and ankylosis of elbow joints as well as visceral anomalies.
- Type 2 and type 3 Pfeiffer syndrome are usually associated with neurodevelopmental abnormalities and have shortened life expectancy



Pfeiffer Syndrome





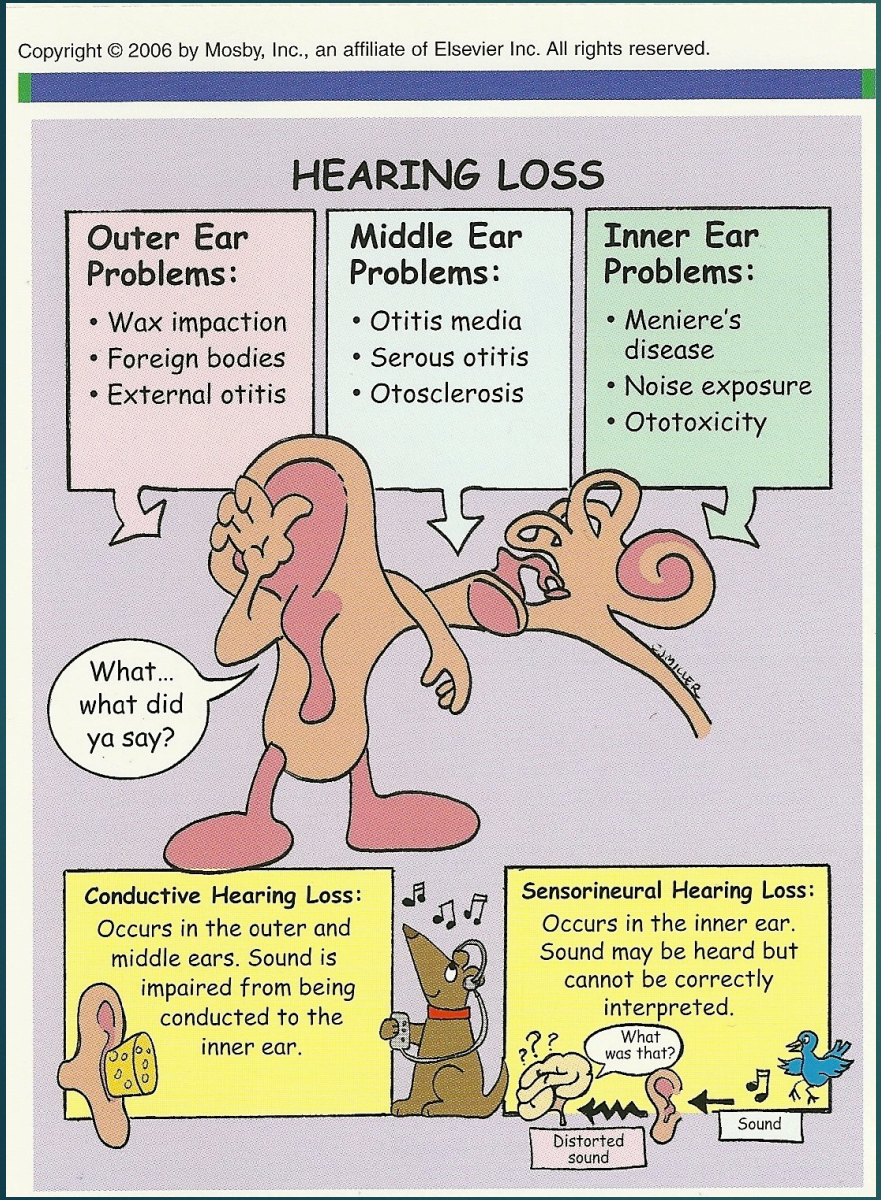
Pfeiffer Syndrome

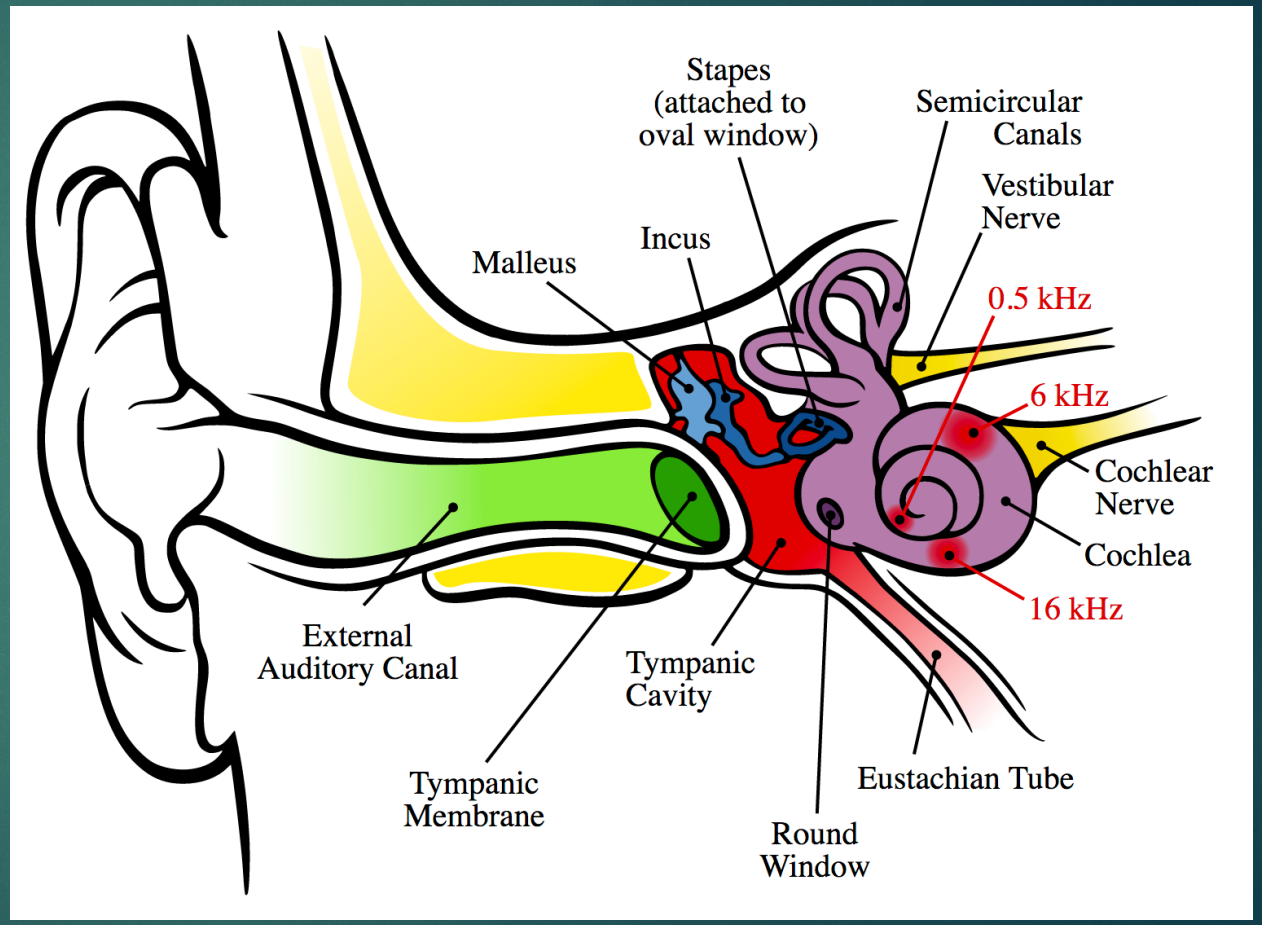
Which of the following fingers are most likely to be joined by syndactyly in this condition?

A. Thumb and index finger
B. Index and middle fingers
C. Middle and ring fingers
D. Ring and little fingers

Originally described in 1964 by Rudolf Pfeiffer, characterised by premature fusion of certain cranial bones (craniosynostosis), midface hypoplasia with beaked nasal tip, ocular proptosis, abnormally broad and medially deviated thumbs and great toes, and conductive hearing loss

Hearing Loss

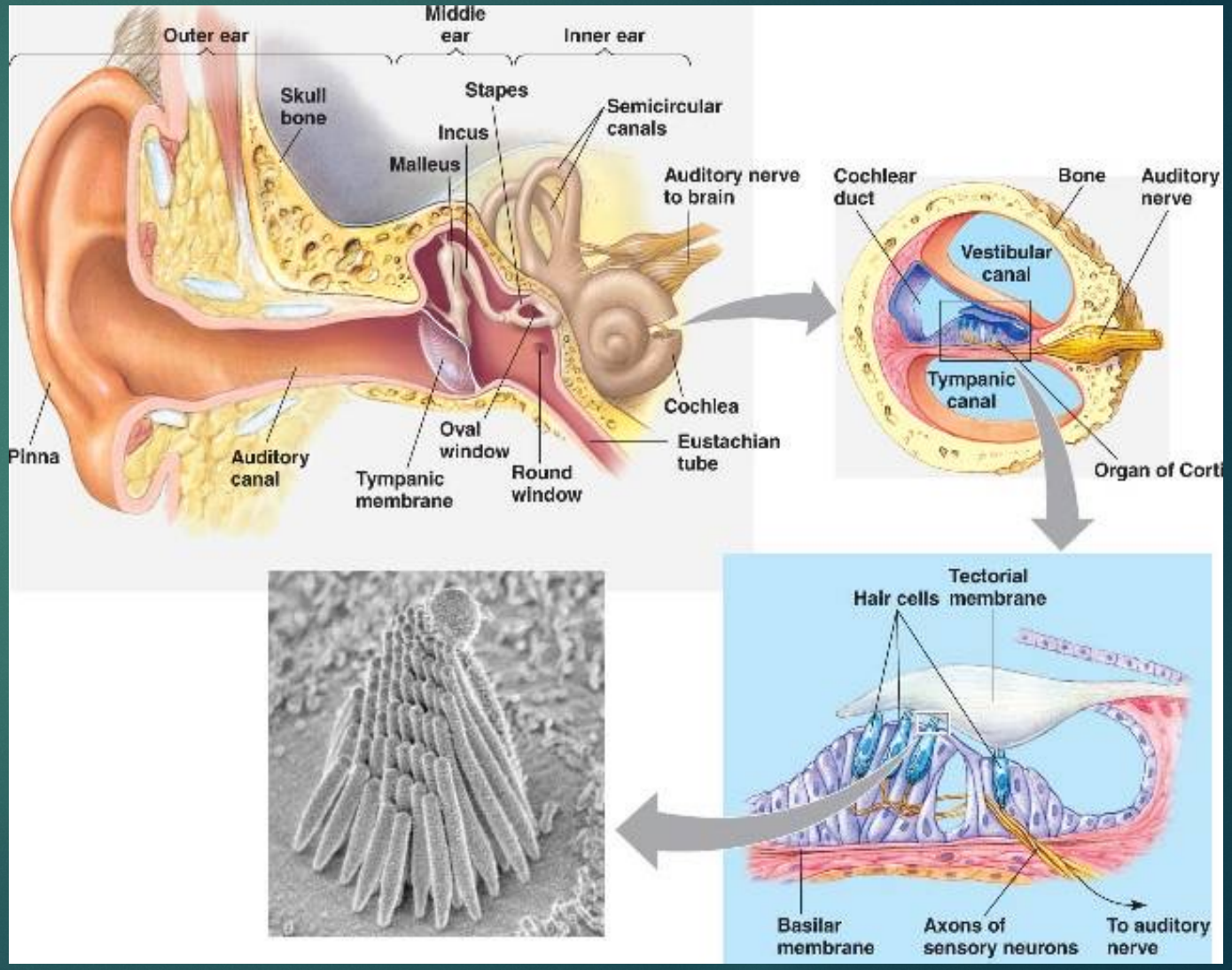




http://tcc-yr9-physics.wikispaces.com/9-3+Hearing+Sounds

Hearing – Structure of the EAR

- Sound waves enter the outer ear via the **pinna** and cause the **tympanic membrane** (eardrum) to vibrate.
- The bones of the middle ear (**malleus**, **incus**, **stapes**) transmit the vibrations to the **oval window** on the **cochlea** ("snail") of the inner ear.
- Vibrations of the **stapes** against the **oval window** produce pressure waves in the **perilymph** of the **cochlea**.
- The waves transmit the sound energy to the **Organ of Corti**.

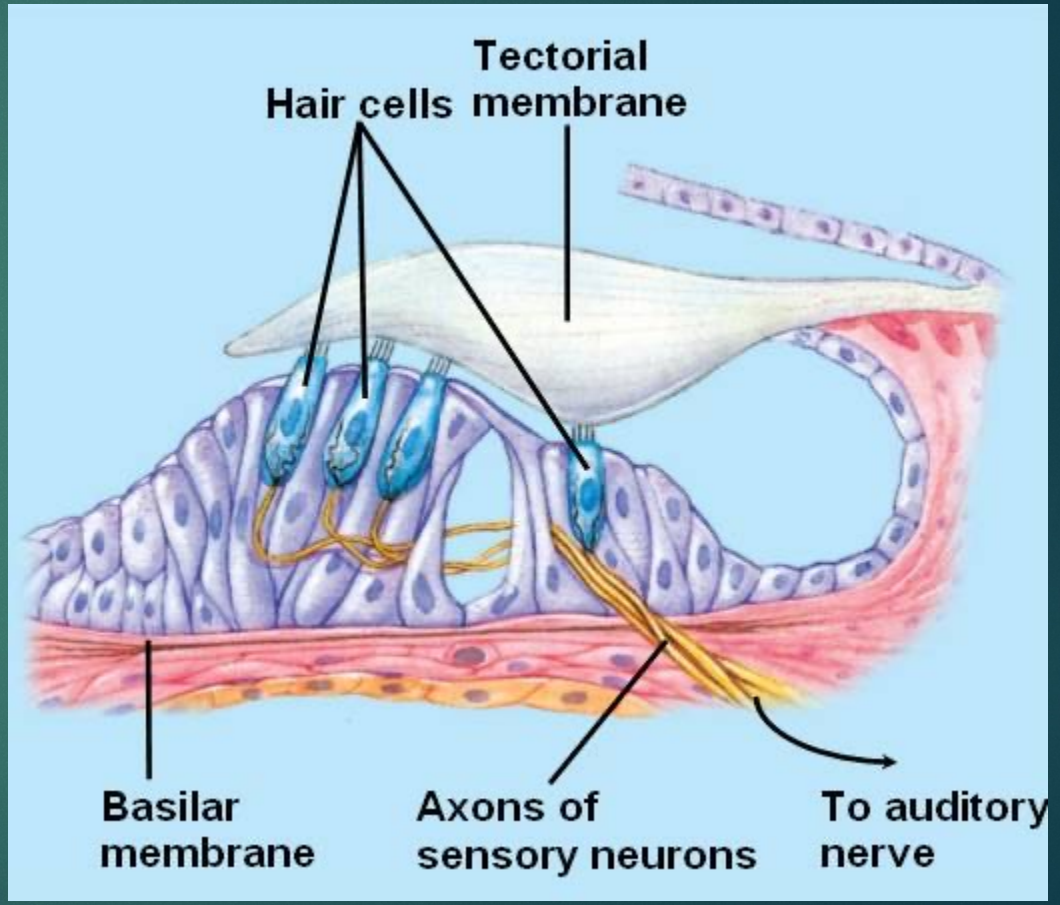


http://bio1152.nicerweb.com/Locked/media/ch50/hearing.html

Hearing – Organ of Corti

- **Organ of Corti:**
  Wave energy from the cochlea causes the basilar membrane to vibrate, bending **hair cells**.

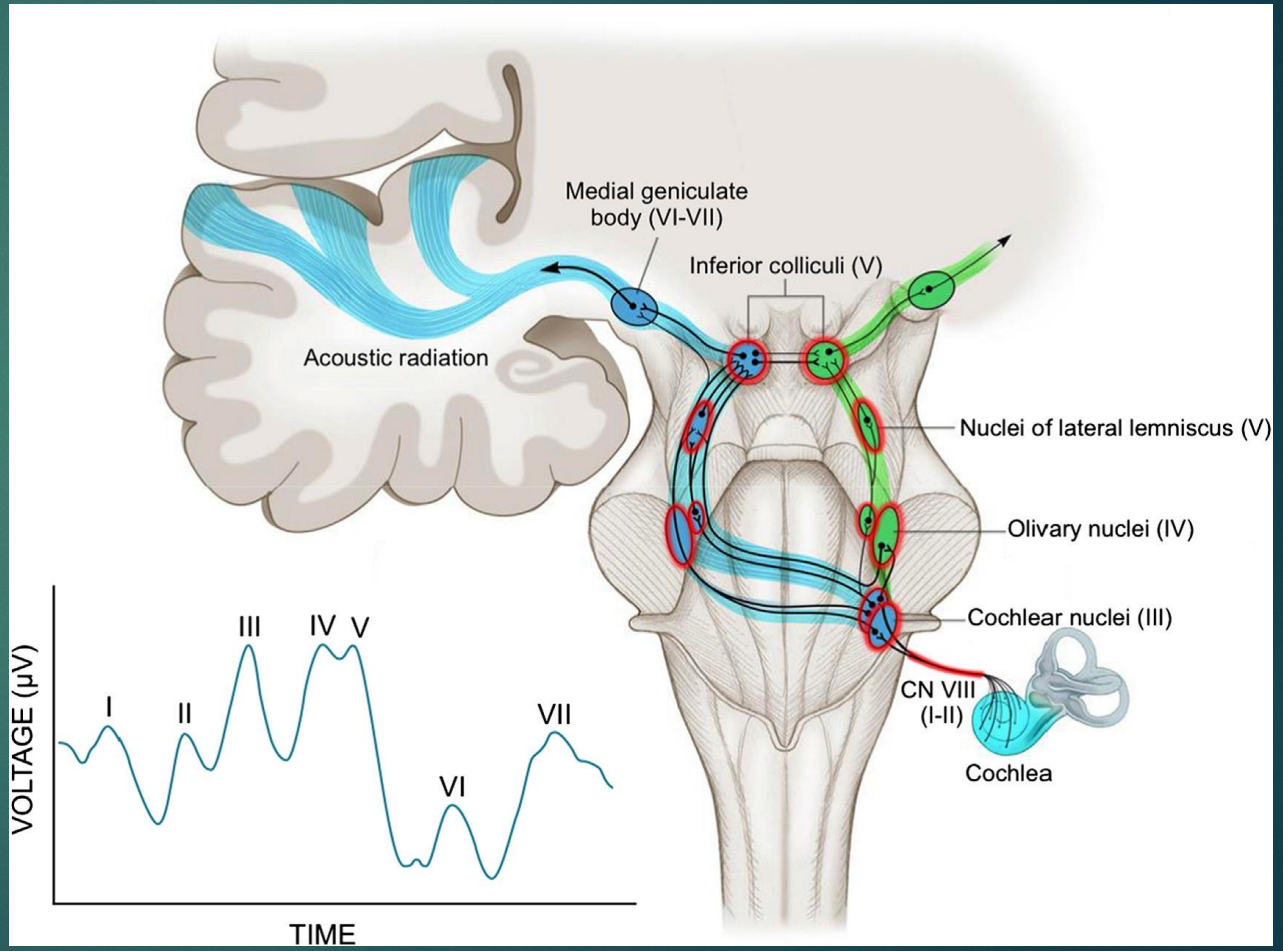
- The **hair cells** depolarize their membranes; **action potentials** travel down the **auditory nerve** to the **temporal lobe** of the cerebrum.



http://bio1152.nicerweb.com/Locked/media/ch50/hearing.html

Auditory Pathway and ABR

- ABR generator sites:
  - I Distal auditory nerve
  - III Caudal brainstem
  - V Lateral lemniscus
- Waves reflect axonal activity



https://pharmaceuticalintelligence.com/2015/02/22/neonatal-pathophysiology/

Case / Question #4

Which of the following is NOT a cause of conductive hearing loss?

A. Abnormal development of the cochlear hair cells

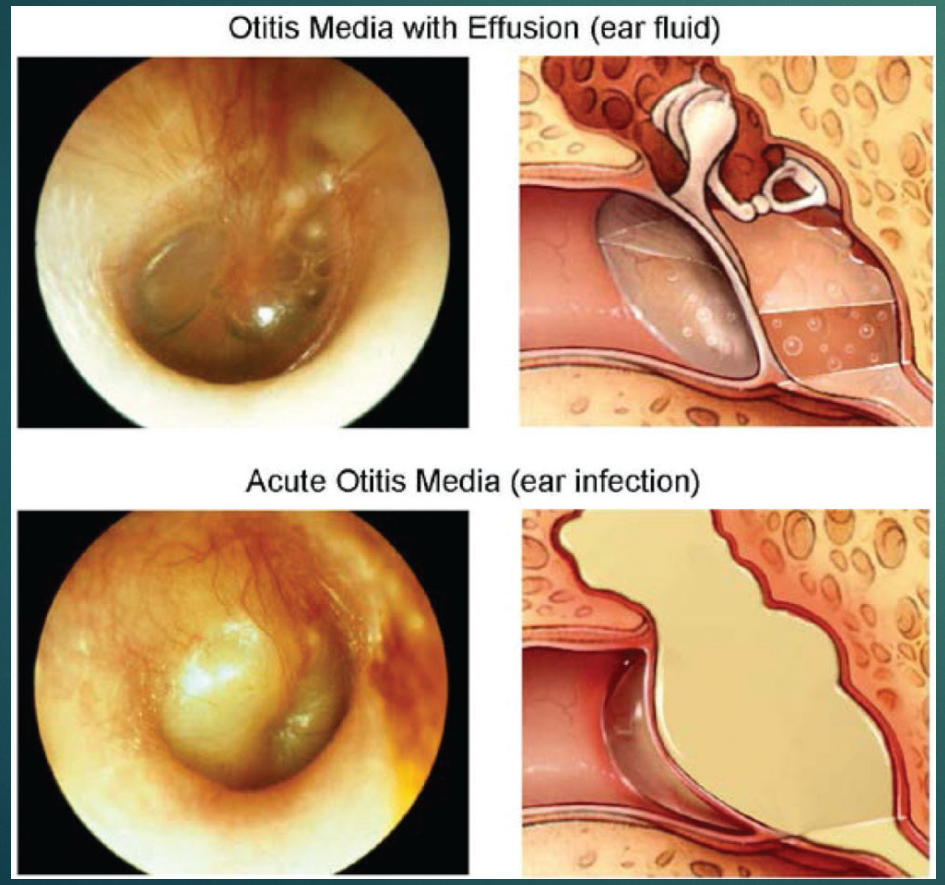
B. Canal stenosis

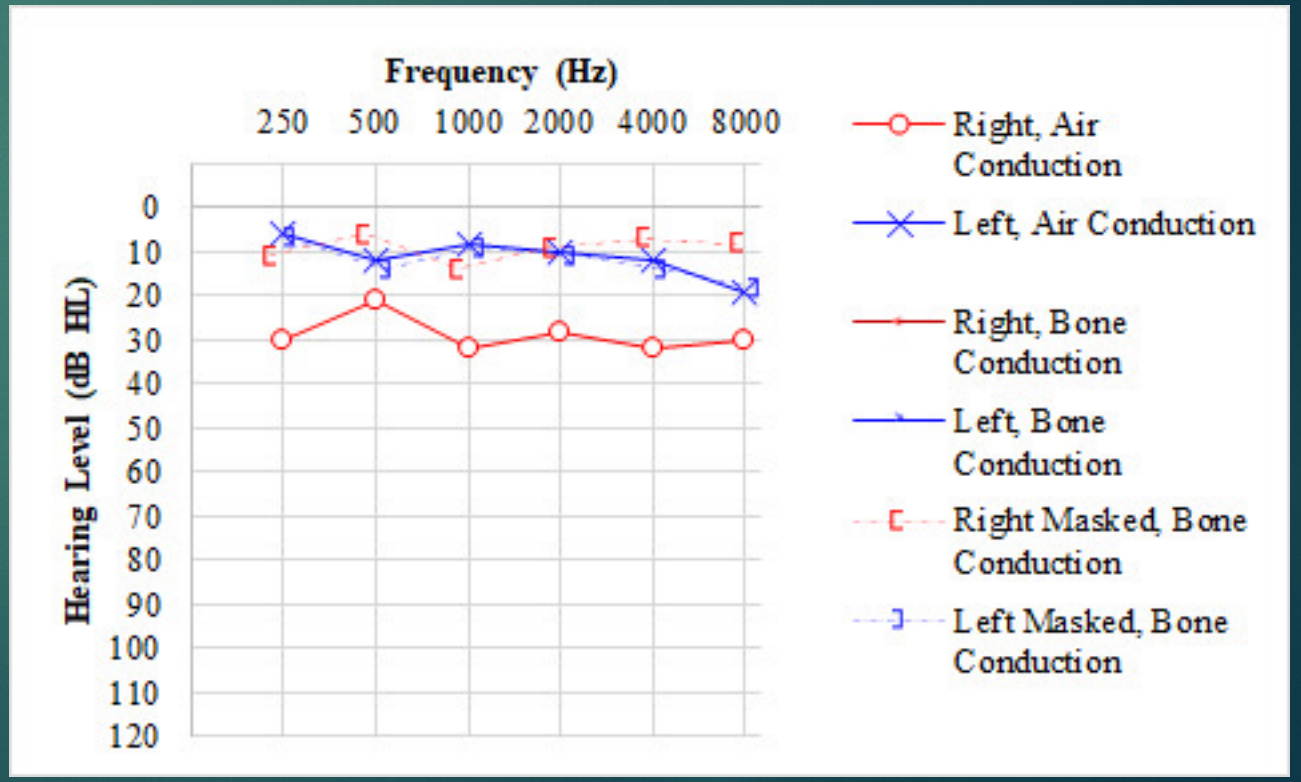
C. Fluid in the middle ear

D. Microtia

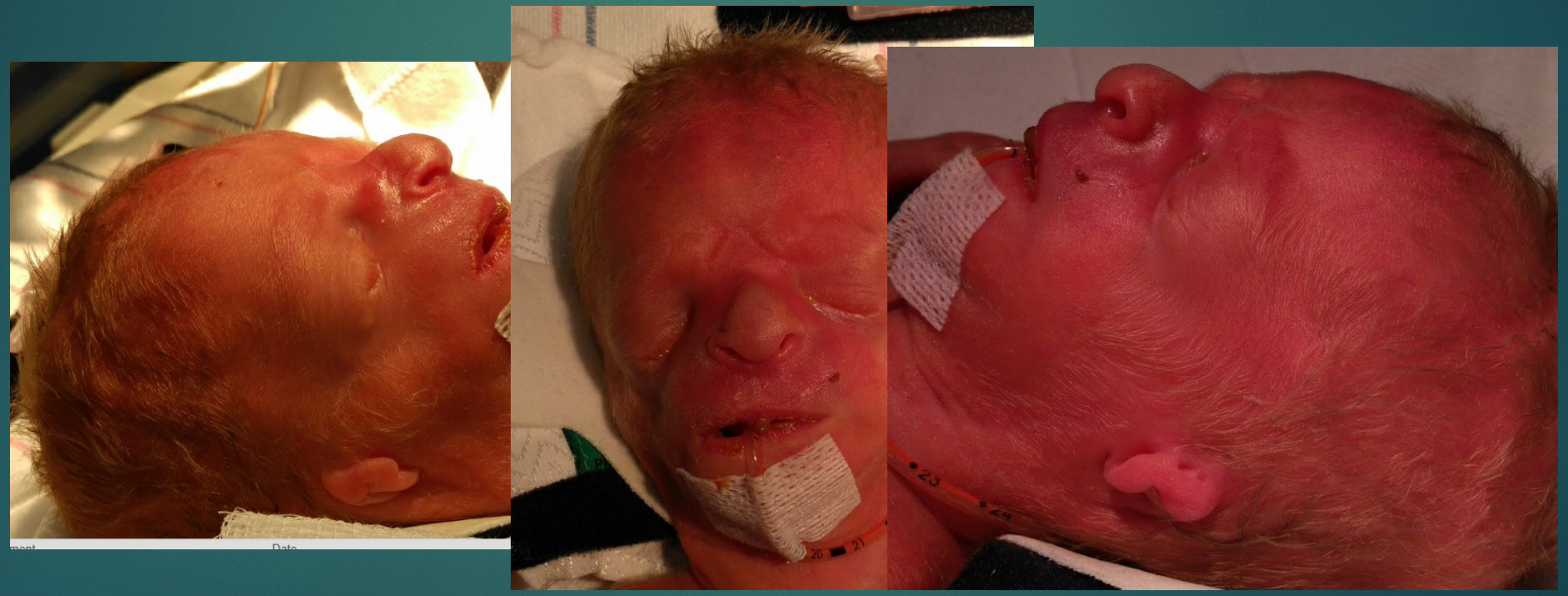
E. Stapes fixation

- Answer A. Abnormal development of the cochlear hair cells. Conductive hearing loss results from interference in the transmission of sound from the external auditory canal to a normal inner ear. In this type of hearing loss, bone conduction is good and air conduction is poor. It can result from fluid in the middle ear, microtia, canal stenosis, and stapes fixation. In sensorineural hearing loss, there is decreased bone and air conduction due to abnormal development or damage to the cochlear hair cells or auditory nerve.

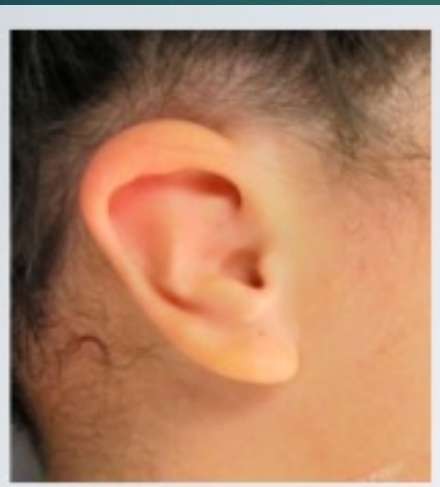




Newborn / Infant

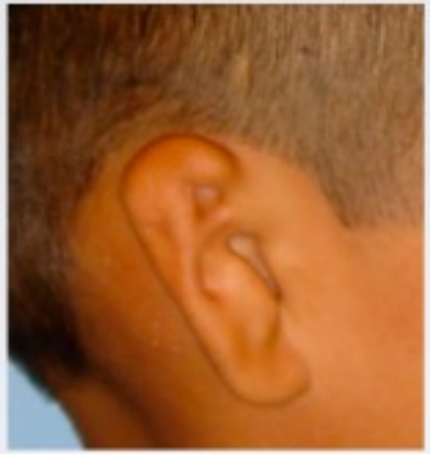


Microtia Grading



Grade 1

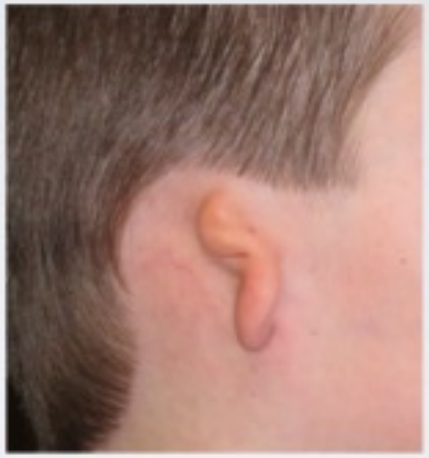
Smaller than normal, but the ear has mostly normal anatomy



Grade 2

Part of the ear looks normal, usually the lower half

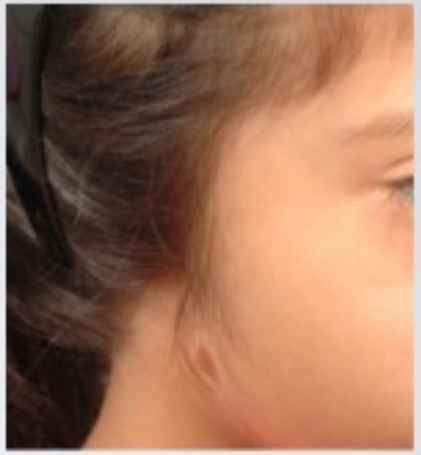
The canal may be normal, small or completely closed



Grade 3

Just a small remnant of "peanut-shaped" skin and cartilage

There is no canal, which is called aural atresia



Grade 4

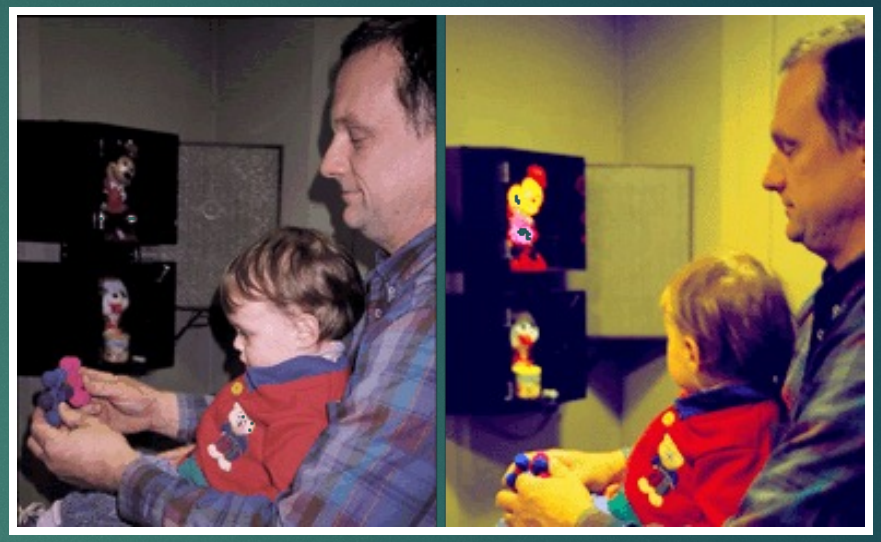
Complete absence of both the external ear and the ear canal, also called "anotia"

Audiologic Testing

- Clinical testing
  - Weber test
  - Rinne test
- Audiometry
  - Pure tone audiometry
  - Speech audiometry
  - Impedance testing

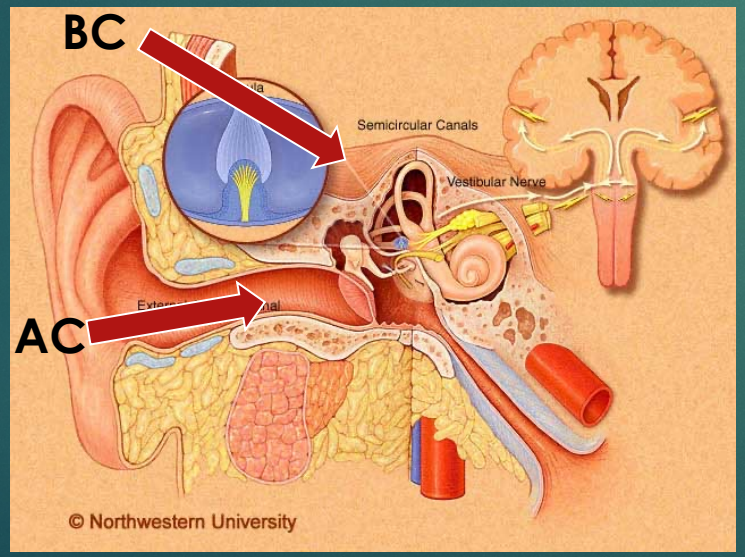
Audiologic Testing in Children

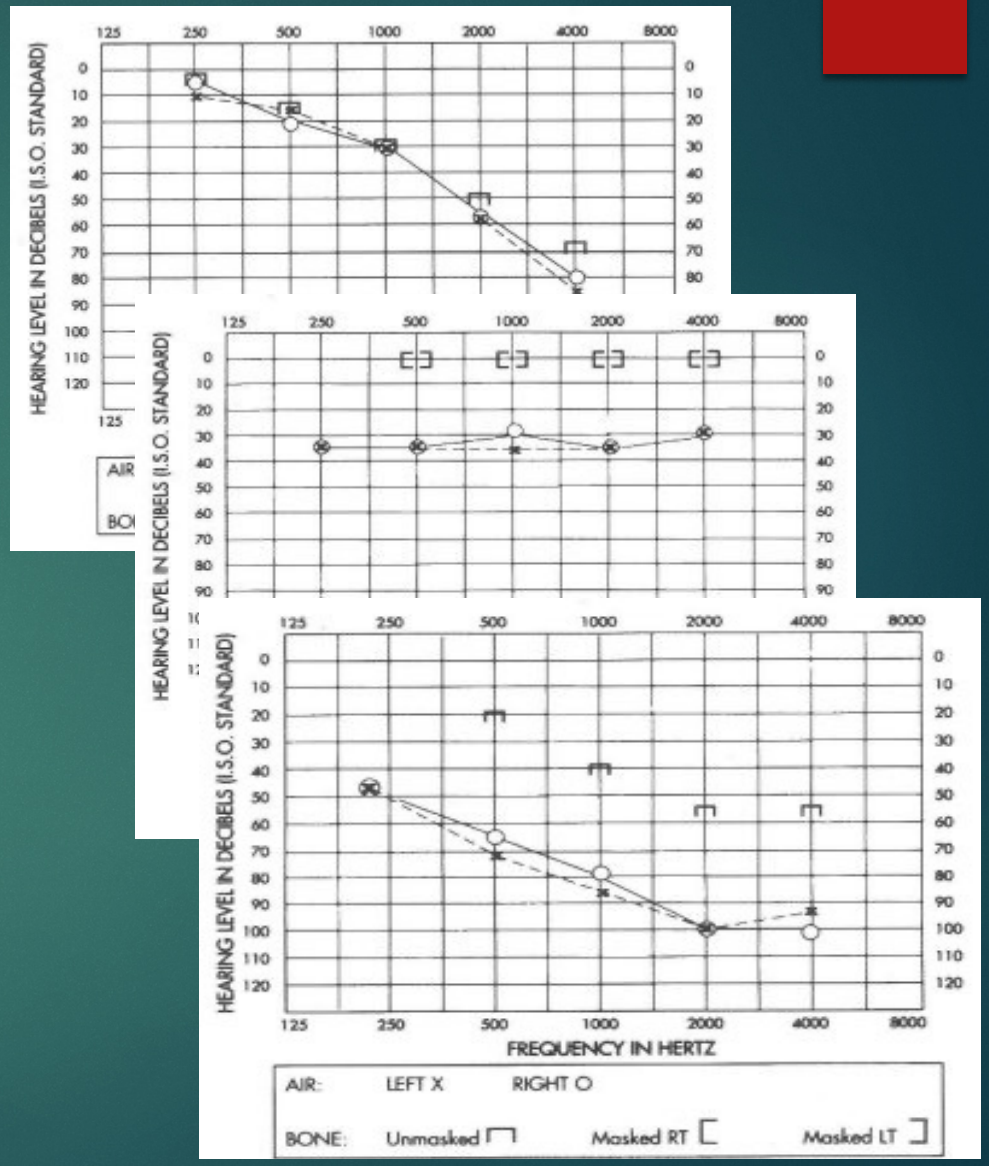
- Behavioral observation audiometry
- Visual reinforcement audiometry
- Play audiometry
- Speech audiometry
- Immittance testing
- Auditory brain stem response testing (ABR)
- Otoacoustic emissions



Types of Hearing Loss

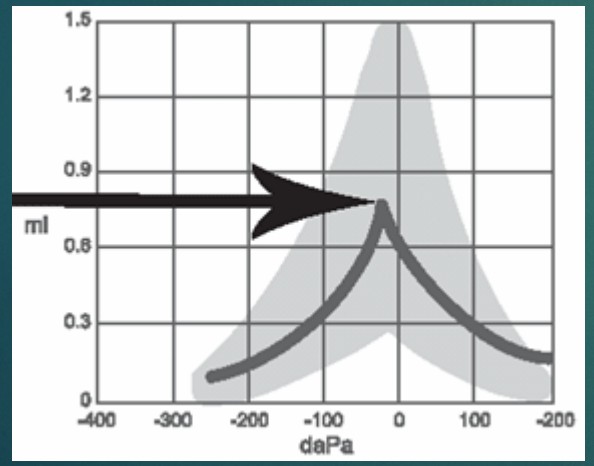
- **Sensorineural HL**
- **Conductive HL**
- **Mixed HL**

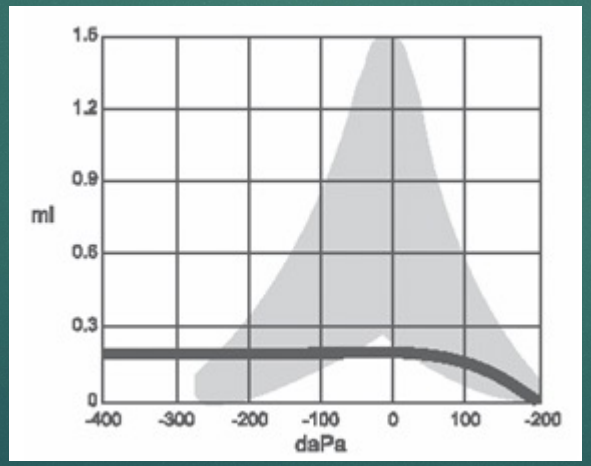


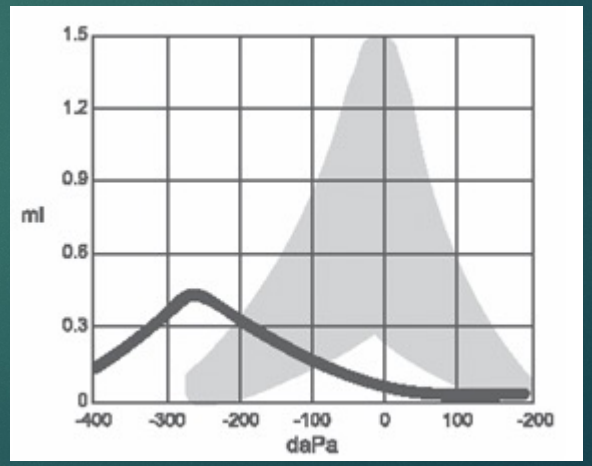


Tympanograms

- **Type A: normal**
- **Type B: OME or perforation**
- **Type C: retracted**

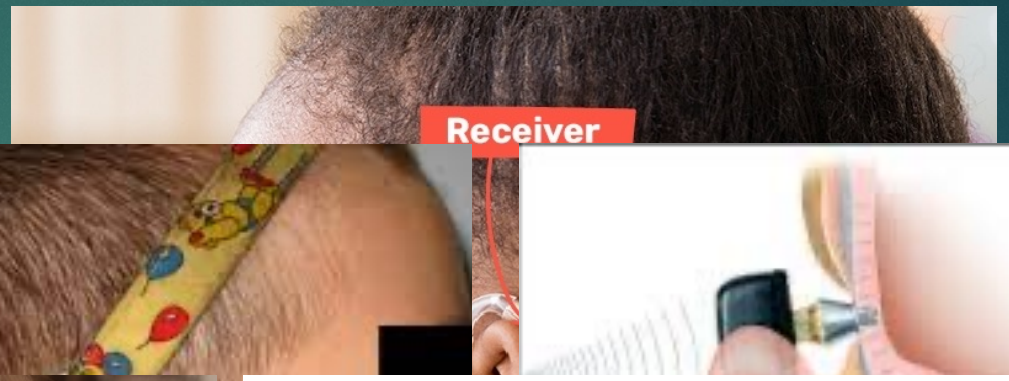


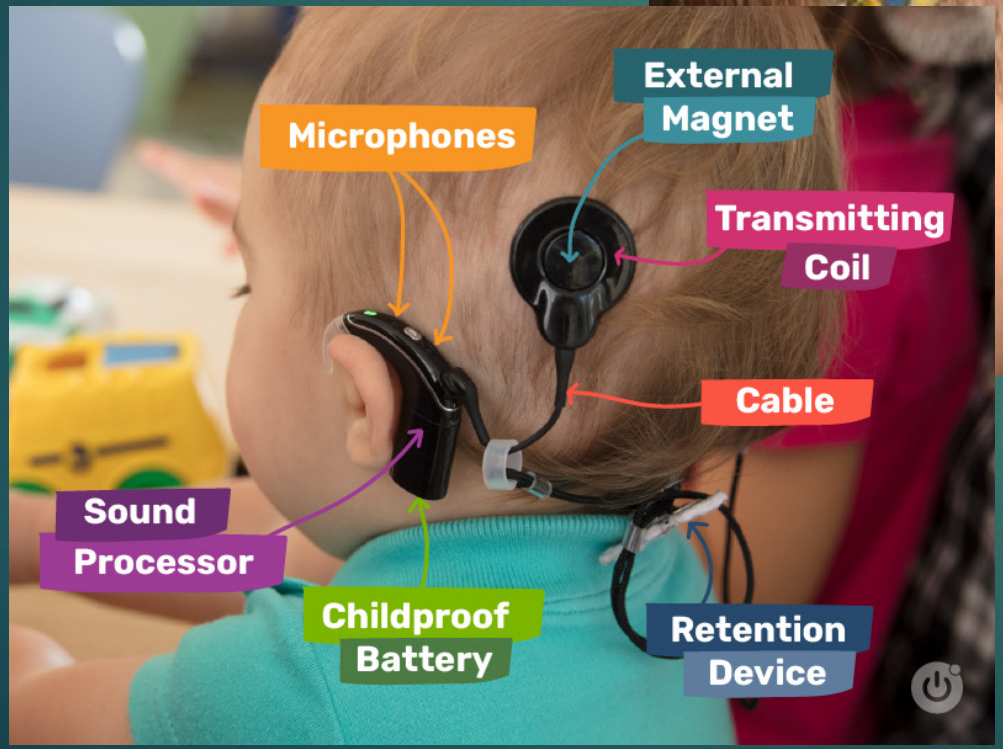


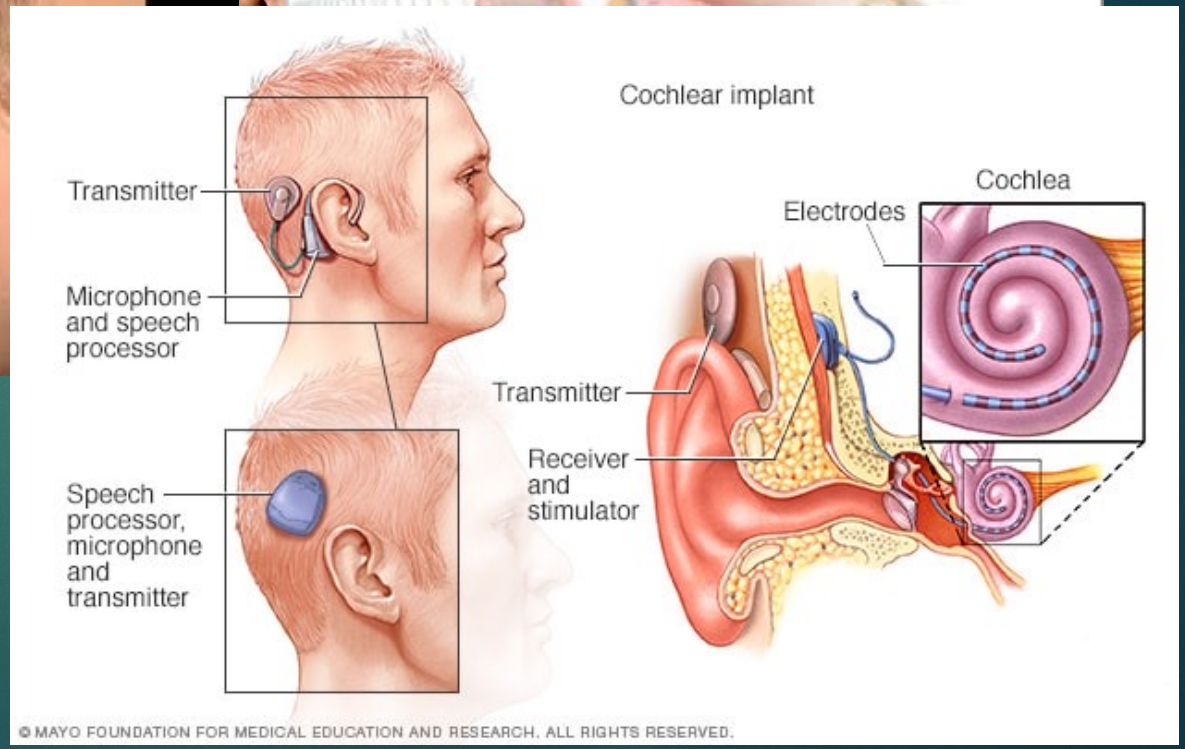


Hearing Rehabilitation

- Conventional hearing aids
- Bone conduction
- Cochlear Implant







My (personal) Goals and Future/Outlook

- Strengthen collaboration with NICU
- Work together on airway management
- Create an annual sim lab / airway course
- Research projects (together with SLP):
  - Lip and Tongue Tie Assessment and Management
  - Palatal Groove in Intubated Children
  - Increase ENT involvement in CMF abnormalities (e.g. oral appliances for PRS babies)
  - Non-invasive 3D scanning (for oral cavity)

An Oral Appliance With Velar Extension for Treatment of Obstructive Sleep Apnea in Infants With Pierre Robin Sequence

Margit Bacher, M.D., Judit Sautermeister, M.D., Michael S. Urschitz, M.D., M.Sc., Wolfgang Buchenau, M.D., Joerg Arand, M.D., Christian F. Poets, M.D.

References:

- An Approach to the Airway Management in Children with Craniofacial Anomalies; Brahim Boudjenah, Al Moataz Billah Adham, Surendra M. Chinnappa, Omed Assaf and Mashael Al-Khelaifi; DOI: 10.5772/intechopen.93426
- https://radiopaedia.org/articles/fibromatosis-colli
- https://healthjade.net/fibromatosis-colli/
- Quintanilla-Dieck L, Penn EB, Jr.: Congenital Neck Masses. Clin Perinatol 2018, 45(4):769-785.
- https://www.ncbi.nlm.nih.gov/books/NBK519057/#:~:text=Less%20than%201%25%20of%20thyroglossal,an%20asymptomatic%20midline%20neck%20mass.
- Kim HJ, Lee YS, Chang HS. Thyroid Cancer Arising from Thyroglossal Duct Cyst. J Endocr Surg. 2020 Dec;20(4):97-102. https://doi.org/10.16956/jes.2020.20.4.97
- Peyton J, Park R, Staffa SJ, Sabato S, Templeton TW, Stein ML, Garcia-Marcinkiewicz AG, Kiss E, Fiadjoe JE, von Ungern-Sternberg B et al: A comparison of videolaryngoscopy using standard blades or non-standard blades in children in the Paediatric Difficult Intubation Registry. Br J Anaesth 2021, 126(1):331-339.
- https://www.ncbi.nlm.nih.gov/books/NBK532882/
- Mahmud N, Abdul Latif H, Mohd Zaki F, Goh BS: Tracheal cartilaginous sleeve in Pfeiffer syndrome: lesson learnt from its rarity. BMJ Case Rep 2021, 14(4).
- Sinner DI, Carey B, Zgherea D, Kaufman KM, Leesman L, Wood RE, Rutter MJ, de Alarcon A, Elluru RG, Harley JB et al: Complete Tracheal Ring Deformity. A Translational Genomics Approach to Pathogenesis. Am J Respir Crit Care Med 2019, 200(10):1267-1281.
- Epidemiology of Cleft Lip and Palate. Mairaj K. Ahmed, Anthony H. Bui and Emanuela Taioli. DOI: 10.5772/67165
- Whittemore KR, Jr., Dargie JM, Dornan BK, Boudreau B: Otolaryngology Service Usage in Children With Cleft Palate. Cleft Palate Craniofac J 2018, 55(5):743-746.
- Kuo CL, Lien CF, Chu CH, Shiao AS: **Otitis media with effusion in children with cleft lip and palate: a narrative review**. *Int J Pediatr Otorhinolaryngol* 2013, **77**(9):1403-1409.
- Messineo D, Chernikava M, Pasquali V, Bertin S, Ciotti M, de Soccio G, Savastano V, Catalano C: **Radiological Parameters Review for Choanal Atresia**. *Pediatr Rep* 2021, **13**(2):302-311.
- Brzecka D, Garbacz M, Mical M, Zych B, Lewandowski B: **Diagnosis, classification and management of ankyloglossia including its influence on breastfeeding**. *Dev Period Med* 2019, **23**(1):79-87.
- https://www.hearingfirst.org/hearing-evaluation-and-technology/hearing-devices

Questions?

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Neck Masses

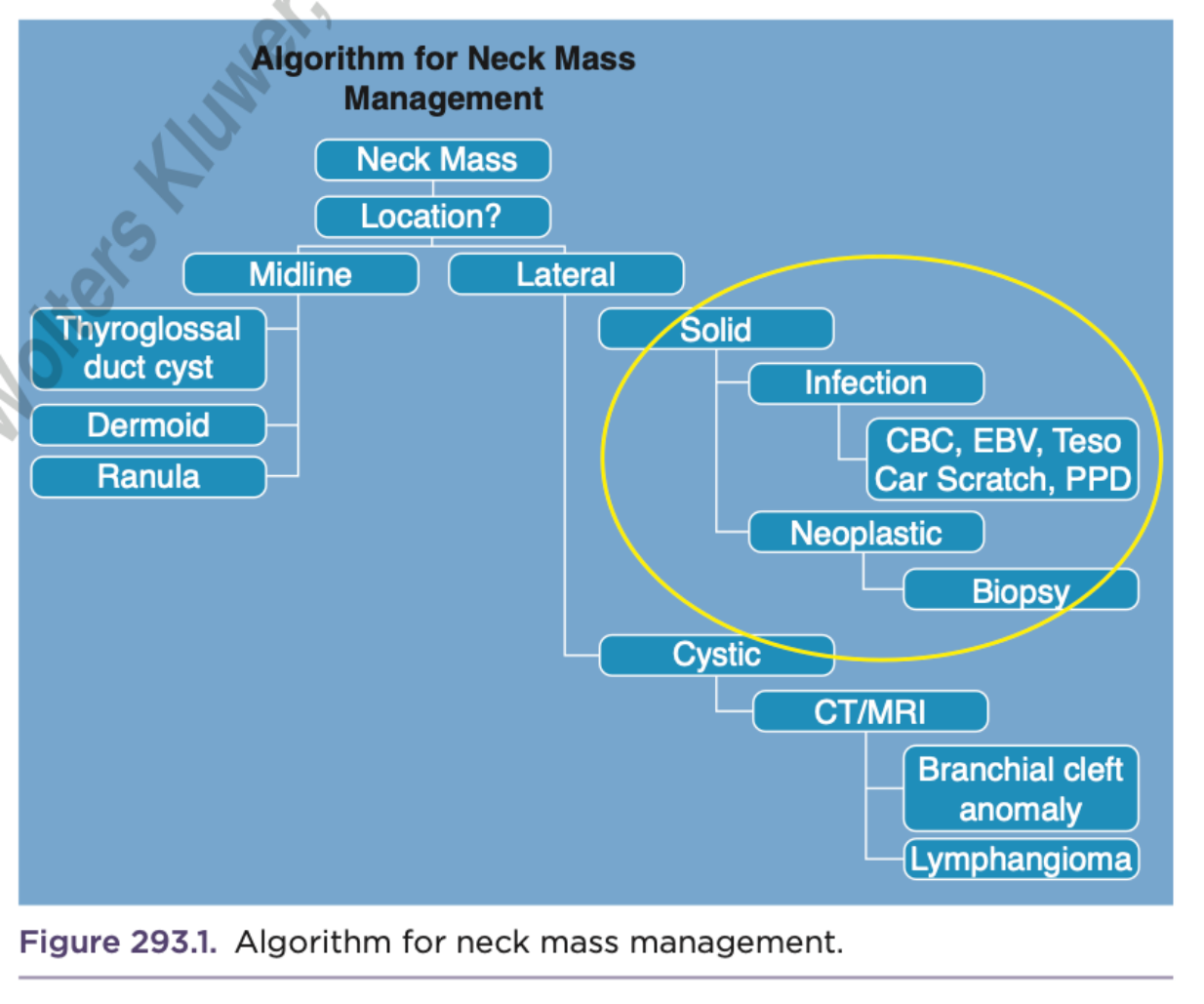
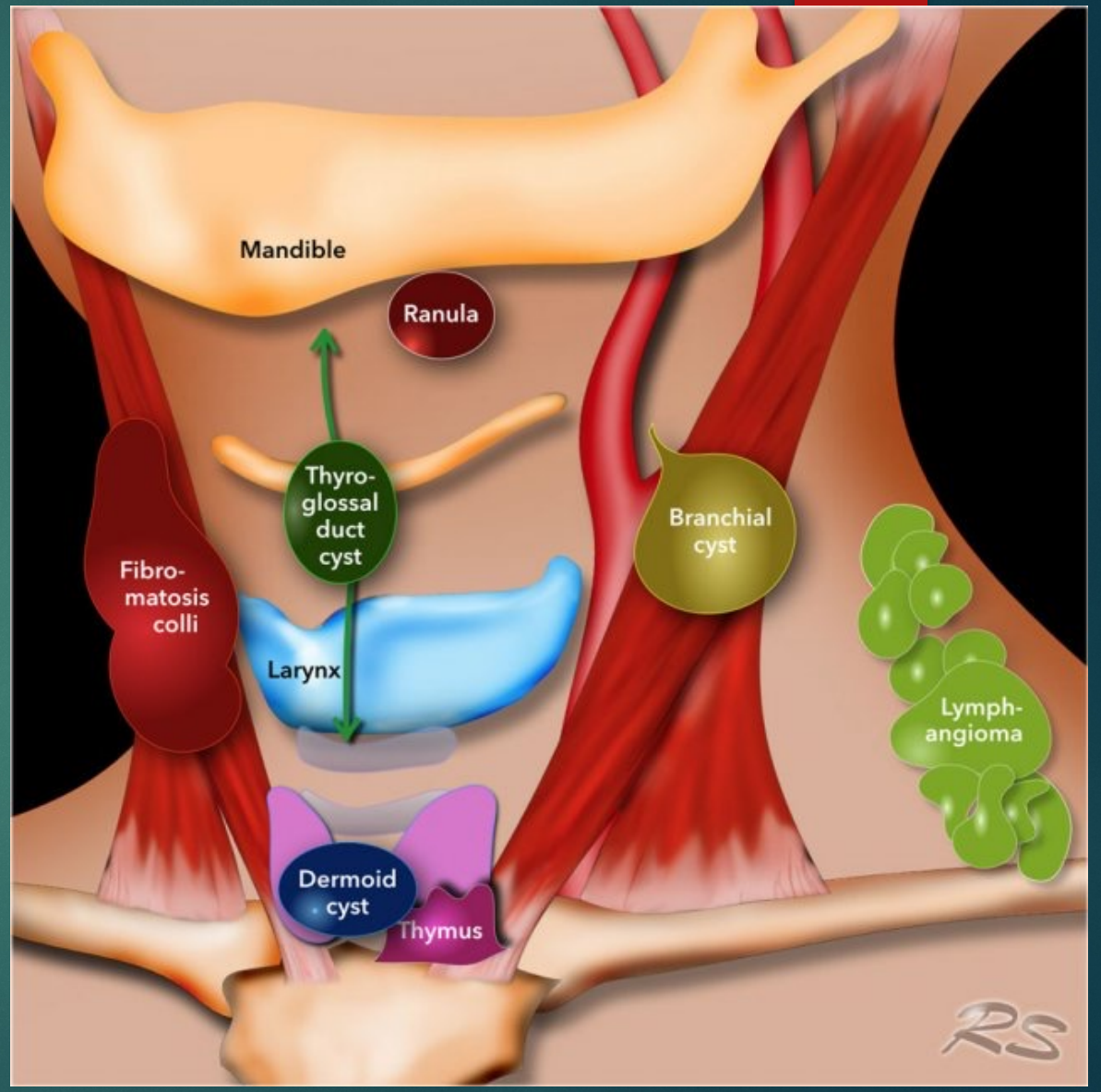


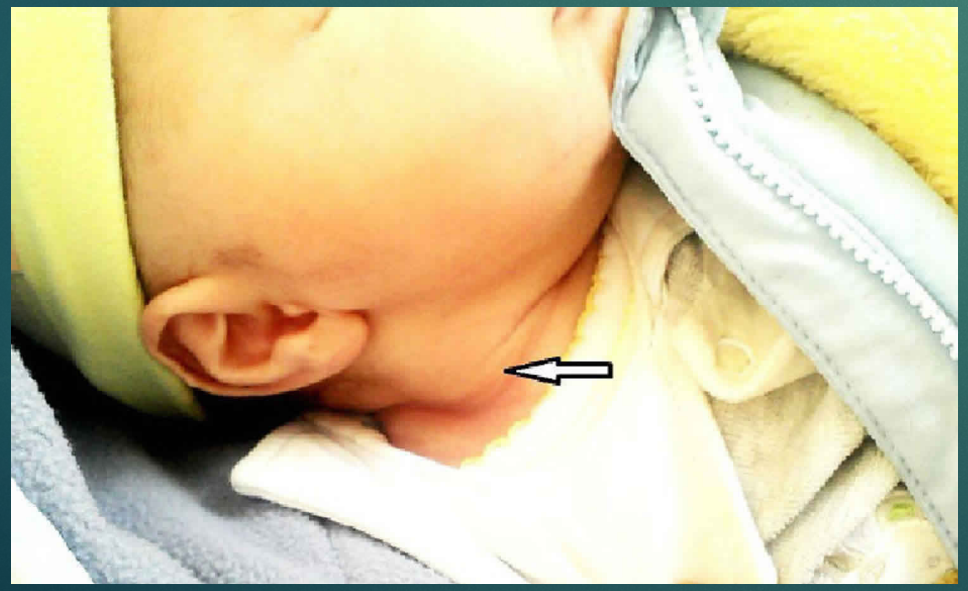
Figure 293.1. Algorithm for neck mass management.

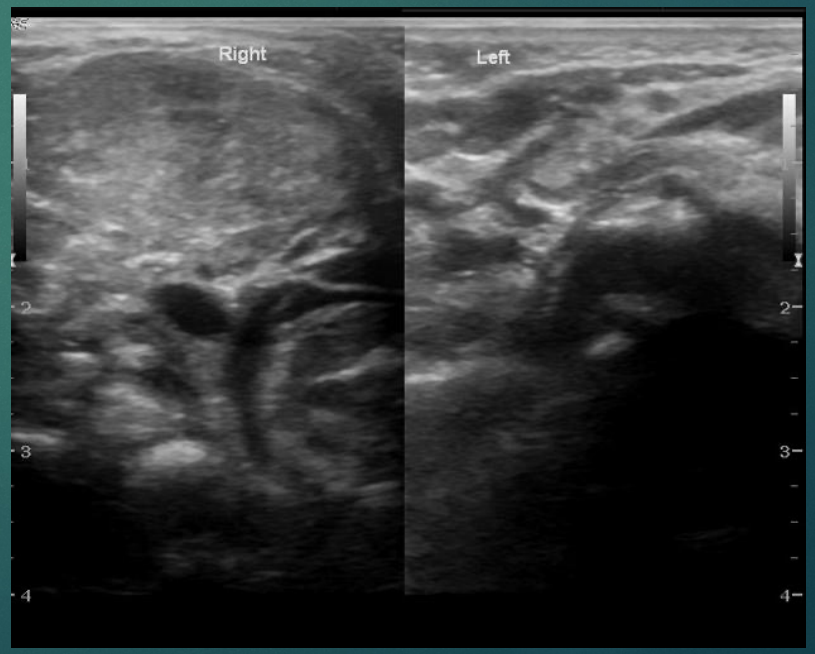


Fibromatosis Colli

= rare form of infantile fibromatosis that occurs within the SCM

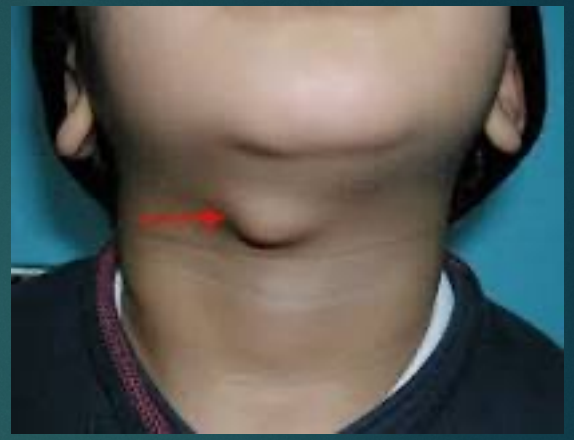
- Thought to be related to birth trauma
- Swelling in the neck, usually unilateral, with torticollis
- Ultrasound is the imaging modality of choice. The sternocleidomastoid muscle is diffusely enlarged (but mostly involves the muscle belly)
- s a self-limiting condition and usually resolves within 4-8 months and mostly requires nothing more than physiotherapy in form of active and passive stretching exercises.





Thyroglossal Duct Cysts

- Thyroglossal duct cysts are the most frequently occurring congenital cervical anomalies, with a 7% population prevalence
- Often presents as infected neck mass
- The exact location of the TGDC mirrors the embryologic pathway of the TGDC. Most TGDCs are in the thyro-hyoidal region (66%), followed by the suprahyoidal region (26%). Suprasternal (5%) and intralingual (2%) locations are less common.
- (!) ~1% chance of malignant transformation (papillary thyroid carcinoma)
- Studies showed a female-to-male ratio of 7:1, with an average age at the time of surgery 42.25 years (range, 26–68 years)



To clip or not to clip…

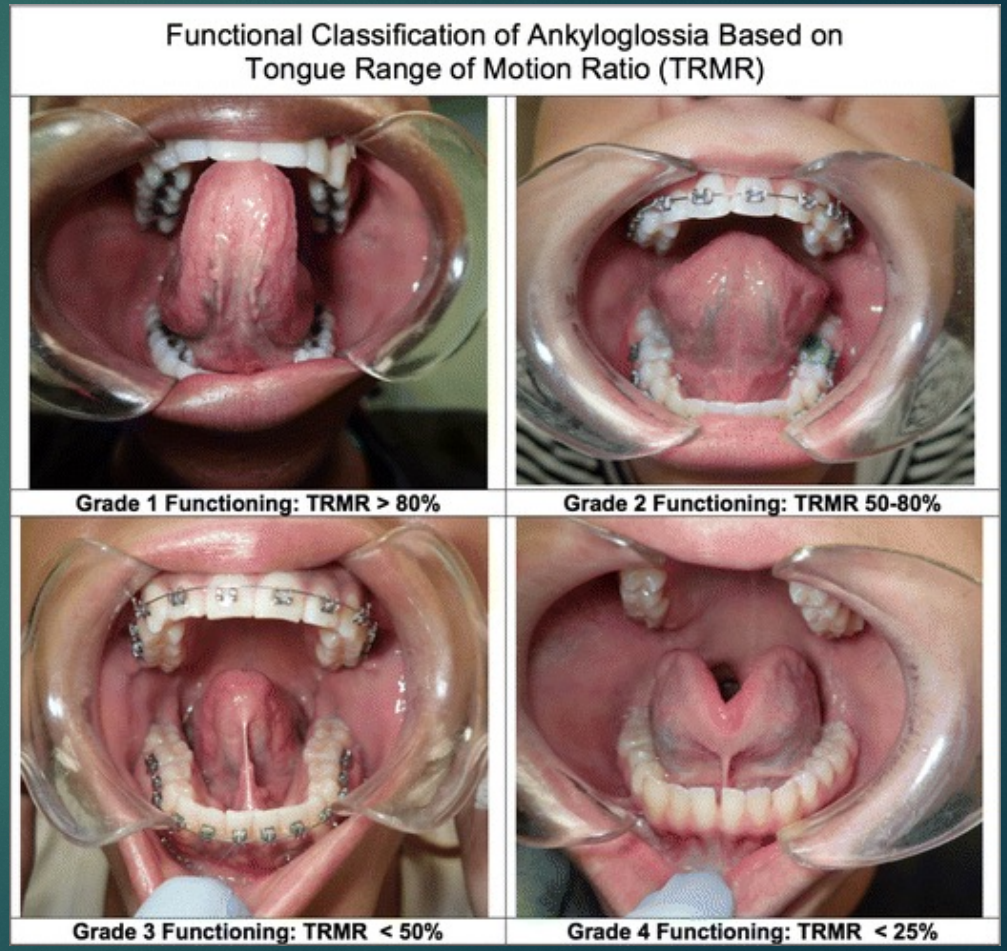
- **Ankyloglossia**
  - = short lingual frenulum (ankyloglossia, tongue-tie) is defined as a congenital malformation that limits lingual mobility, thus leading to impaired tongue function.
  - Its prevalence in newborns is estimated in the literature at between 4.2% and 10.7%
  - Male to female ratio of 3:1
  - The discrepancies in tongue-tie frequency assessment are most probably caused by the lack of unambiguous and objective diagnostic criteria, as well as significant differences in study groups

Ankyloglossia

- **Classification**:

Classification of ankyloglossia according to Kotlow (based on the "free tongue" length).

Normal, clinically acceptable range of "free tongue" >16mm	
Class 1: mild ankyloglossia	12-16 mm
Class II: moderate ankyloglossia	8-11 mm
Class III: severe ankyloglossia	3-7 mm
Class IV: complete ankyloglossia	<3 mm



Ankyloglossia

- **Treatment**:
  - (in my opinion):
  - Frenulectomy is rarely indicated for speech reasons (unless it is very severe or there are concomitant oral-motor problems).
    - It may, however, be warranted for problems with early feeding, bolus manipulation, dentition, or aesthetics.
  - No definitive proof that it's affecting breastfeeding
  - Ask SLP eval before clipping
  - Don't just clip it, RELEASE it
  - Highly recommend post-intervention myofascial release therapy
  - Little effect of intervention if no neuromuscular retraining of appropriate tongue resting posture and suckle