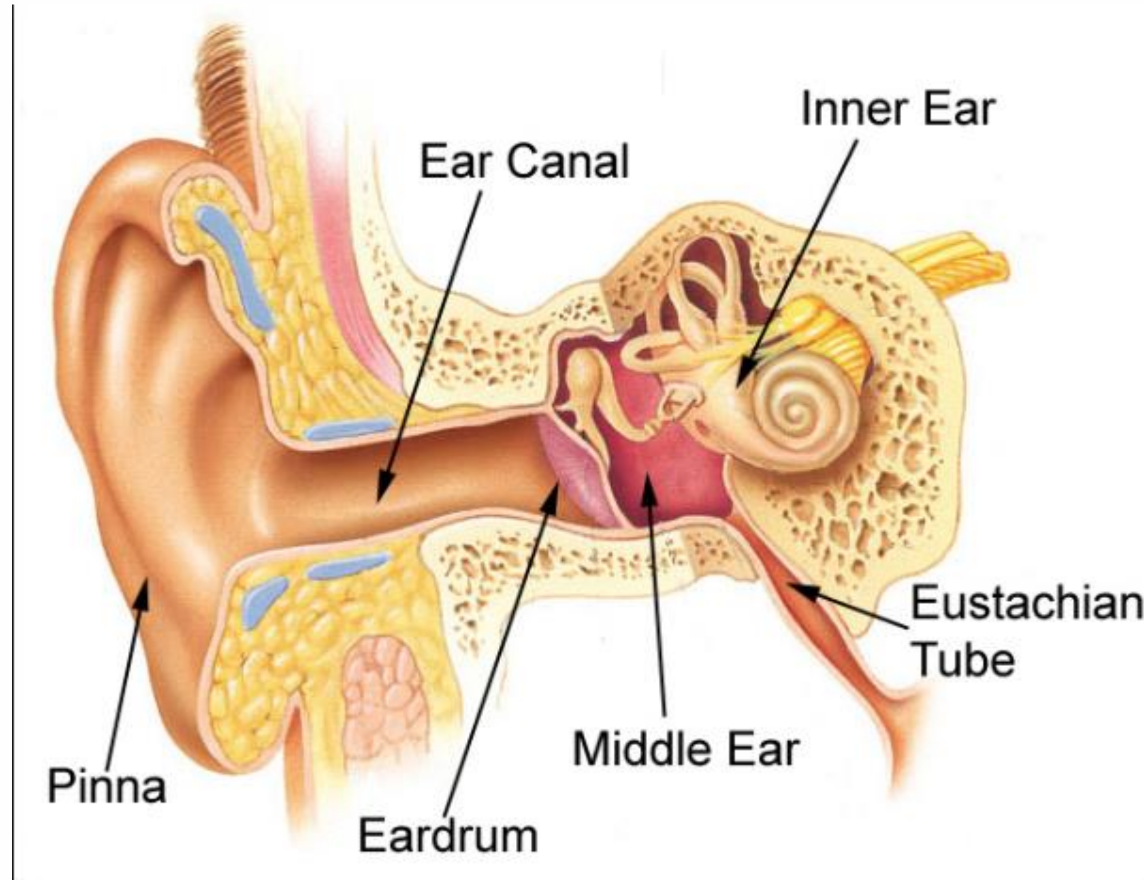
22q AND EAR, NOSE AND THROAT: WHAT YOU NEED TO DISCUSS WITH YOUR DOCTOR

Steven D. Handler, MD, MBE
Division of Otolaryngology
Children's Hospital of Philadelphia
Perelman School of Medicine at the University of Pennsylvania

EAR ANATOMY



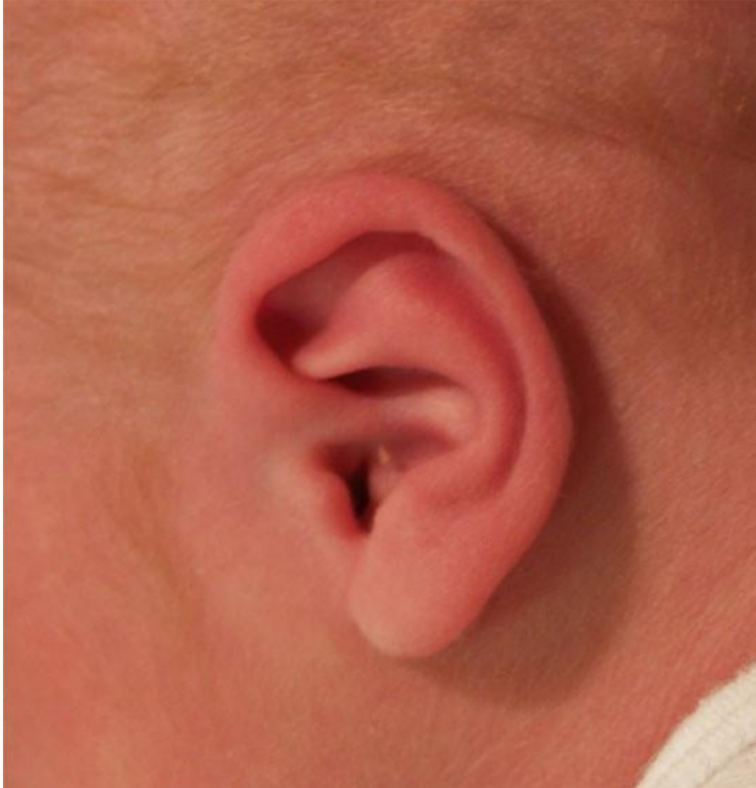
EAR (OTOLOGIC) CONCERNS

- Auricular malformations – cosmetic
- Small external ear canals
  - Visualization of Tympanic Membrane (TM)/ear drum more difficult
  - Wax may block view of TM, but rarely affects hearing
- Eustachian tube dysfunction (fluid +/or infections) related to:
  - Palate abnormalities
  - Cranial base abnormalities
  - Adenoiditis
- Ossicular (ear bones) malformations
- Inner ear malformations (cochlea, auditory nerve)

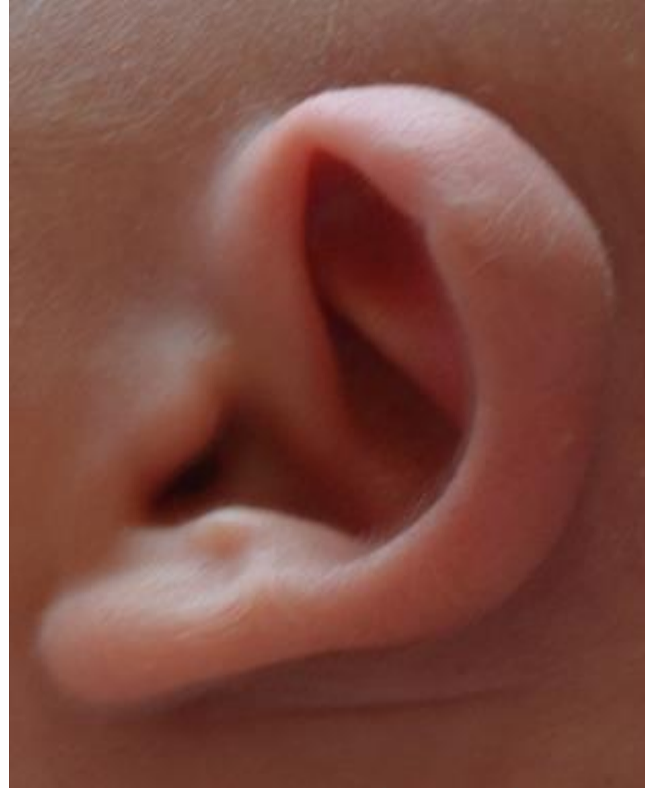
HEARING LOSS IN 22q

- Types of hearing loss
  - Conductive (mechanical)—most common—usually temporary/treatable
    - Ear canal (wax)
    - TM (perforation)
    - Middle ear (fluid, infection, middle ear ossicles)
  - Sensorineural (Nerve) --uncommon—usually permanent
    - Auditory Nerve
    - Inner Ear (cochlea)
  - Mixed (conductive and sensorineural)

AURICLE (PINNA)



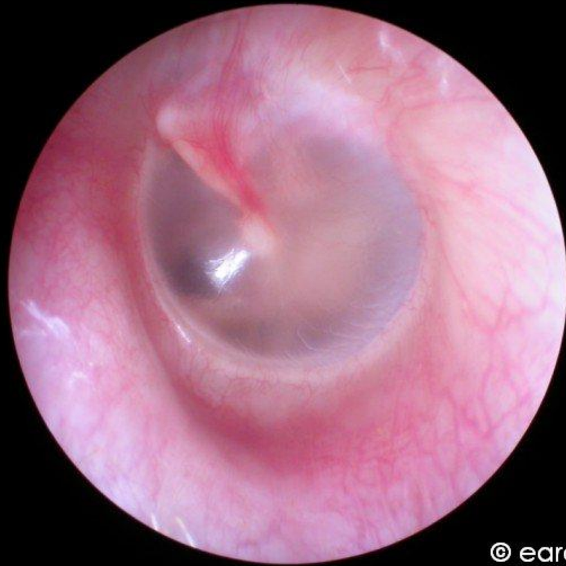
Normal



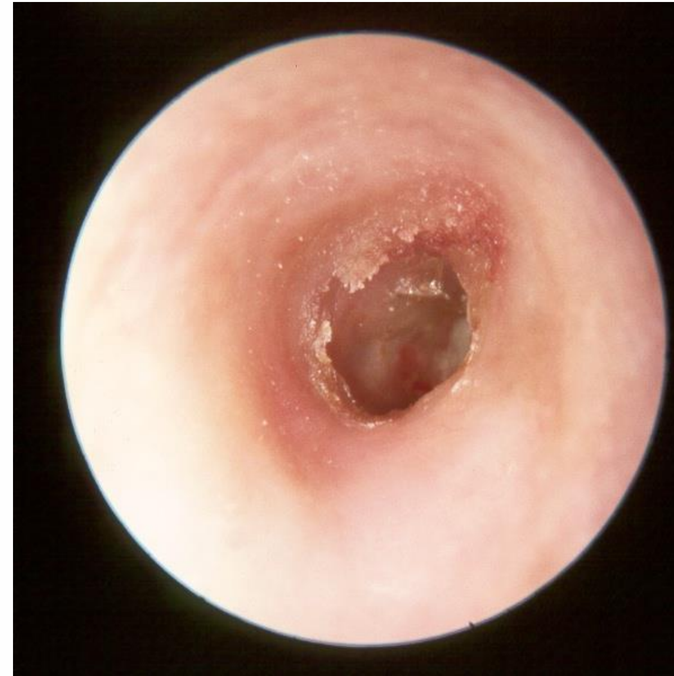
Prominotia
"Lop ear"

Overfolded helix

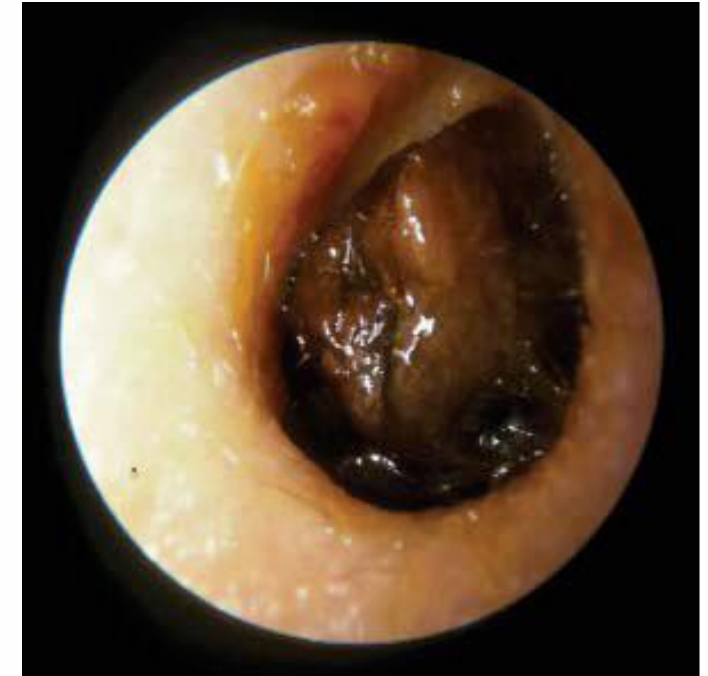
EAR CANAL



Normal Ear Canal



Stenotic Ear Canal

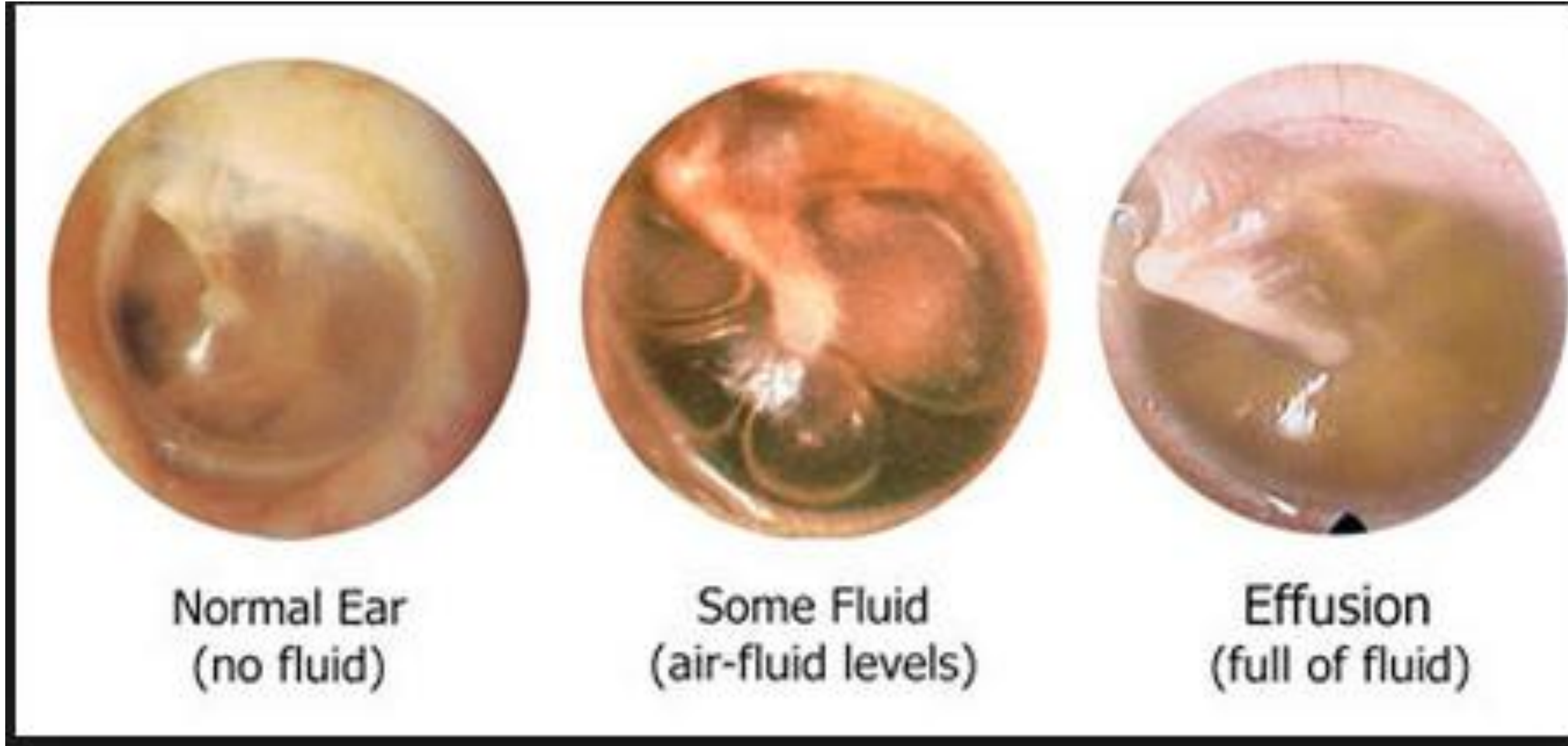


Wax Impaction

TREATMENT OF WAX

- Wax comes out by itself—often needs no intervention
- Periodic use of peroxide or debrox
- Irrigations
- Clean out by ENT
- NO Q-tips

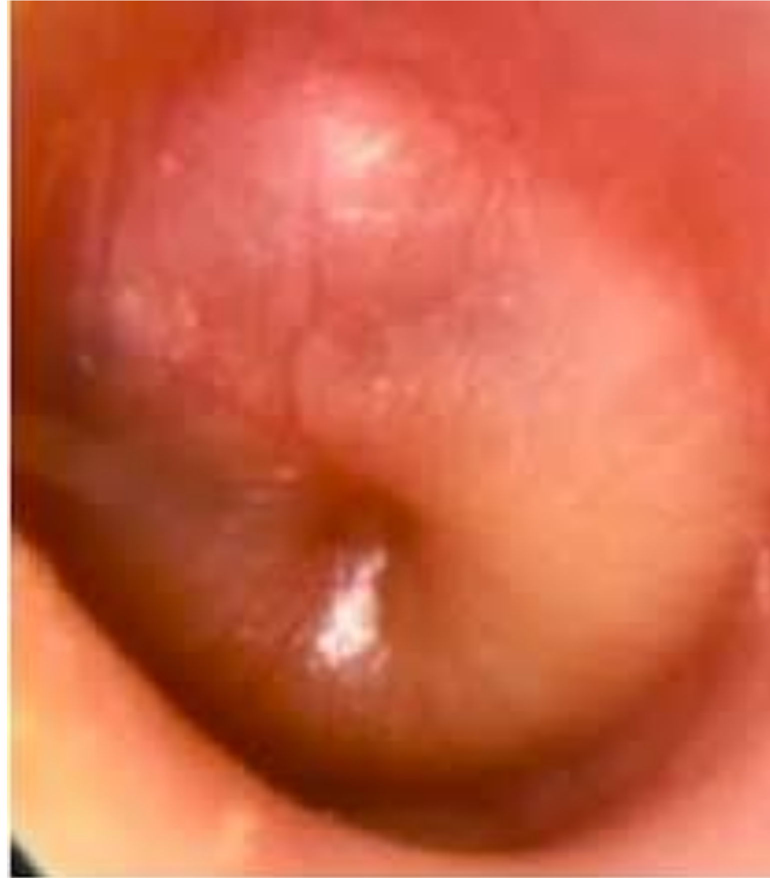
MIDDLE EAR FLUID



TREATMENT OF MIDDLE EAR FLUID

- Observation
- Antihistamines/decongestants do not help
- **No** antibiotics
- Ear tubes for chronic fluid

MIDDLE EAR INFECTION



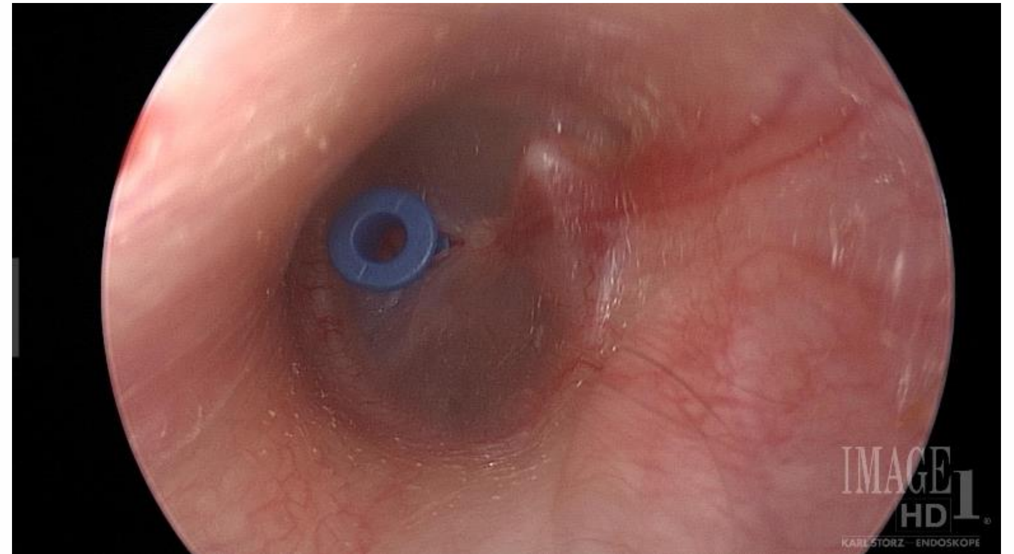
TREATMENT OF MIDDLE EAR INFECTIONS

- Most are viral (≈80%)
- AAP Guidelines
  - Antibiotics
    - Severe infection (pain, fever >39°) in child of any age, unilateral or bilateral
    - Non-severe bilateral infection in child <23 months
  - Watchful waiting
    - Non-severe unilateral infection in child < 23 months
    - Non-severe bilateral infection in child > 23 months
  - Ear tubes

CRITERIA FOR TUBE INSERTION

- Middle ear fluid
  - >3 months of fluid in a child with normal speech development
  - As soon as possible in a child with speech delay
- Recurrent ear infections
  - 3 infections/6 months
  - 4 infections/12 months
  - Earlier if confounding variables such as antibiotic allergy/resistance, immune deficiency
  - Complications of ear infection (abscess, facial nerve paralysis, mastoiditis)

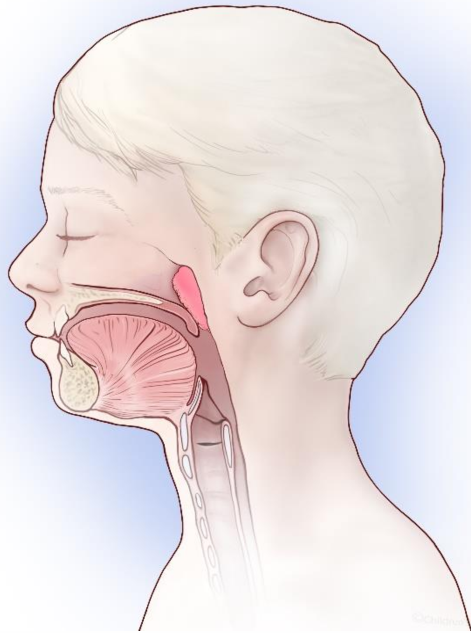
EAR TUBES/GROMMETS

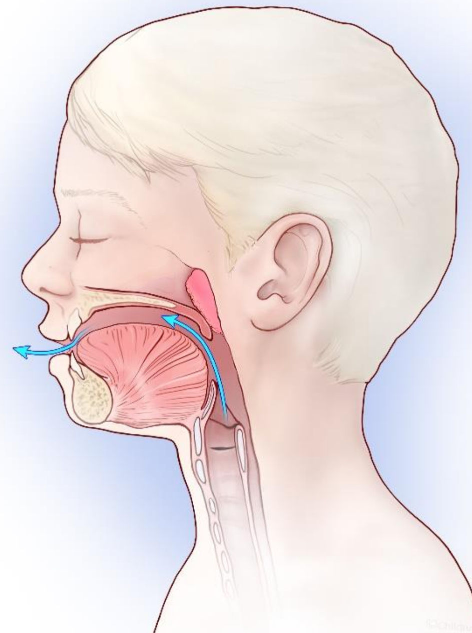


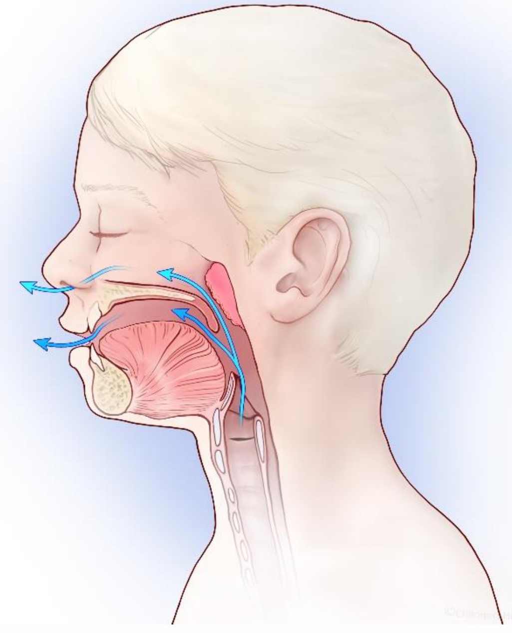
EARS -YOUR JOB

- Talk to your doctor about how easy it is to visualize your child's TM
  - ear wax?
  - periodic drops?
- Get a hearing test at regular intervals-repeat earlier if you think things have changed
- Discuss pros and cons of tubes vs medical management
- Consider immunological work up for recurrent infections
- Hearing aids may be needed in some cases of sensorineural hearing loss

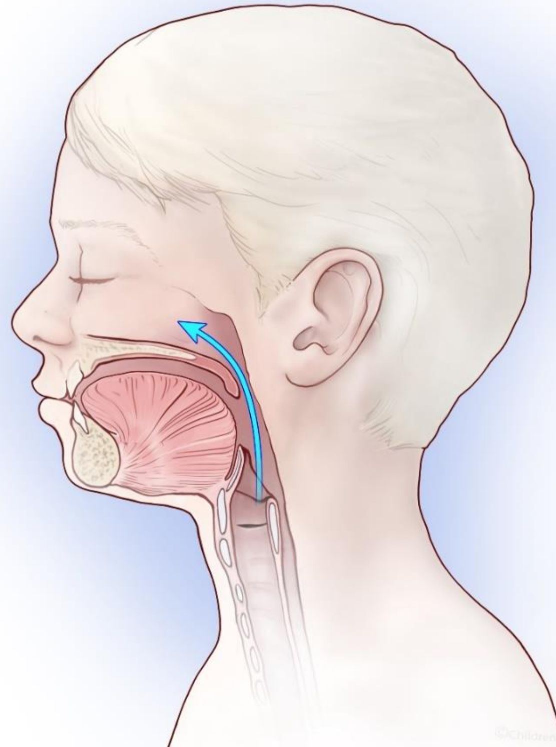
ADENOIDS AND 22Q
NORMAL SPEECH



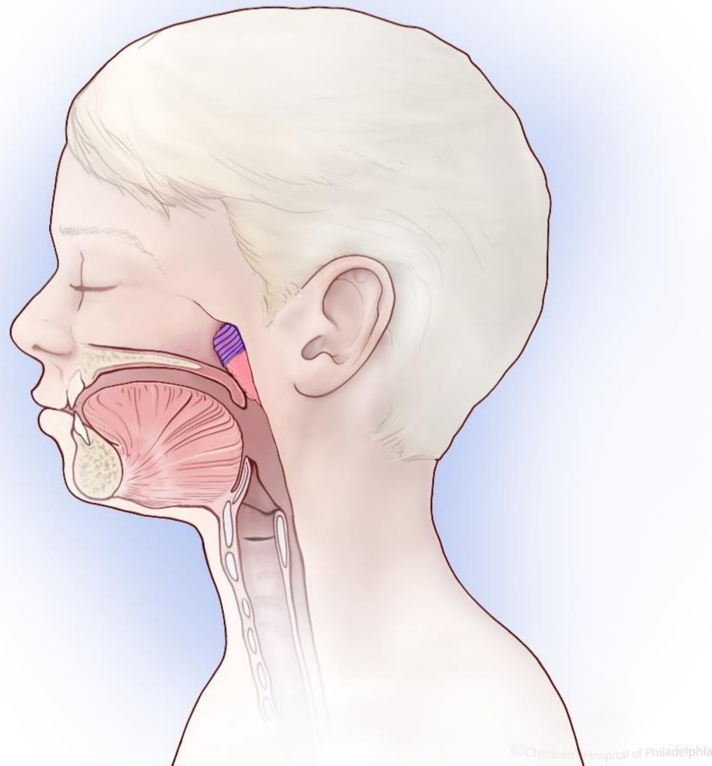


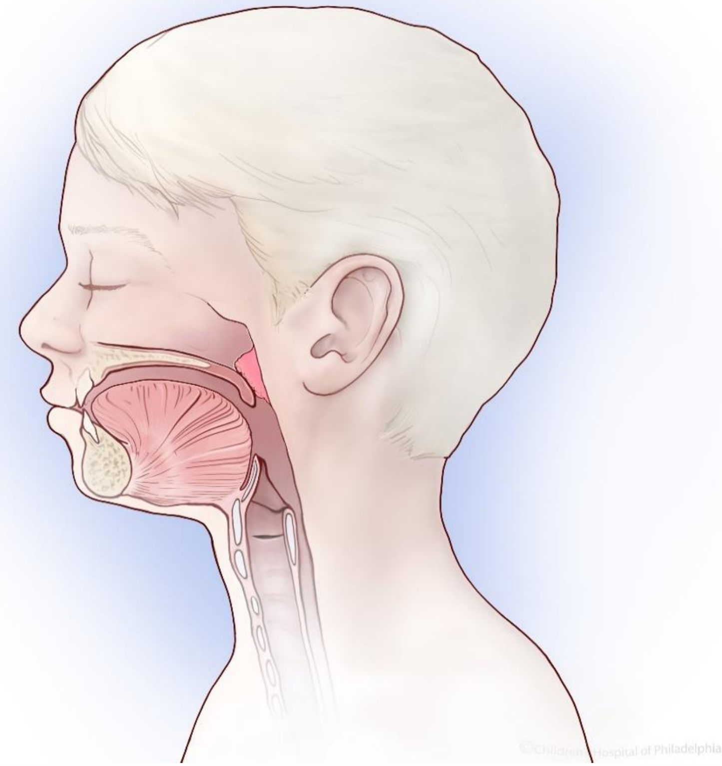


COMPLETE ADENOID REMOVAL



PARTIAL (SUPERIOR) ADENOID REMOVAL





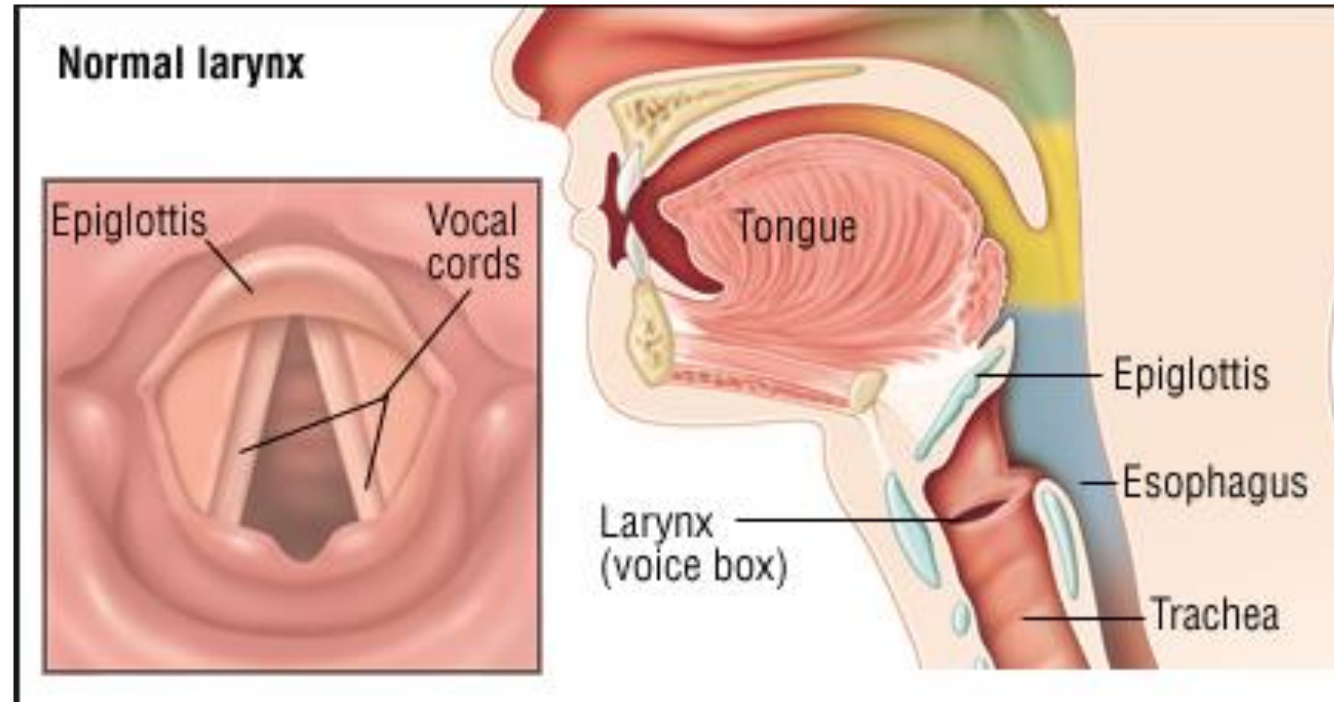
ADENOIDS - YOUR JOB

- Question need for adenoidectomy for ear or sinus disease
- Insist on superior (partial) adenoidectomy, if possible
- Total adenoidectomy, however, is usually necessary prior to pharyngeal flap surgery
  - to permit proper placement of flap
  - to prevent post-op Obstructive Sleep Apnea (OSA)
  - Child may be severely hypernasal during interval between adenoidectomy and flap surgery

WHAT ABOUT TONSILS?

- Tonsils do not play a role in ear or sinus disease and should not be removed just because adenoids are to be removed.
- However, they should be removed if:
  - they are contributing to sleep disturbance/apnea
  - they are involved with recurrent tonsil infections
  - they could interfere with placement of a planned pharyngeal flap
  - they could contribute to post-op OSA

AIRWAY -- LARYNX AND TRACHEA



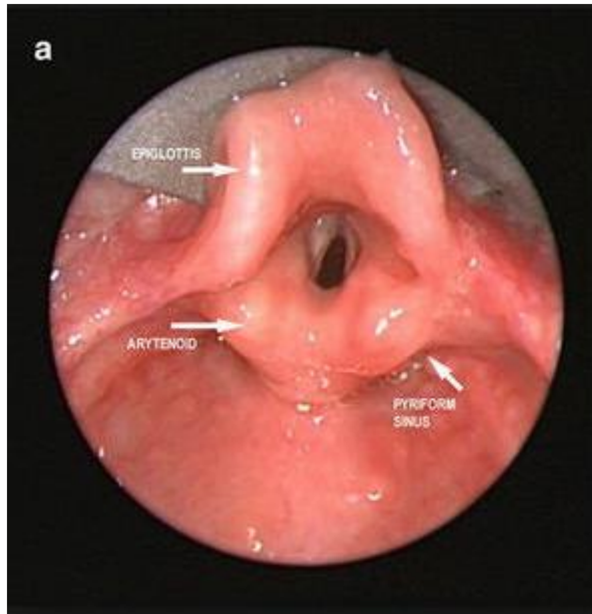
AIRWAY SYMPTOMS

- Noisy breathing (squeaky, high pitched stridor) is NOT normal
- Chronic hoarseness may be sign of vocal strain
  - Can be habit
  - Can be related to VPD
- Choking or difficulty swallowing
- These conditions require ENT evaluation and probable endoscopic exam of upper airway.

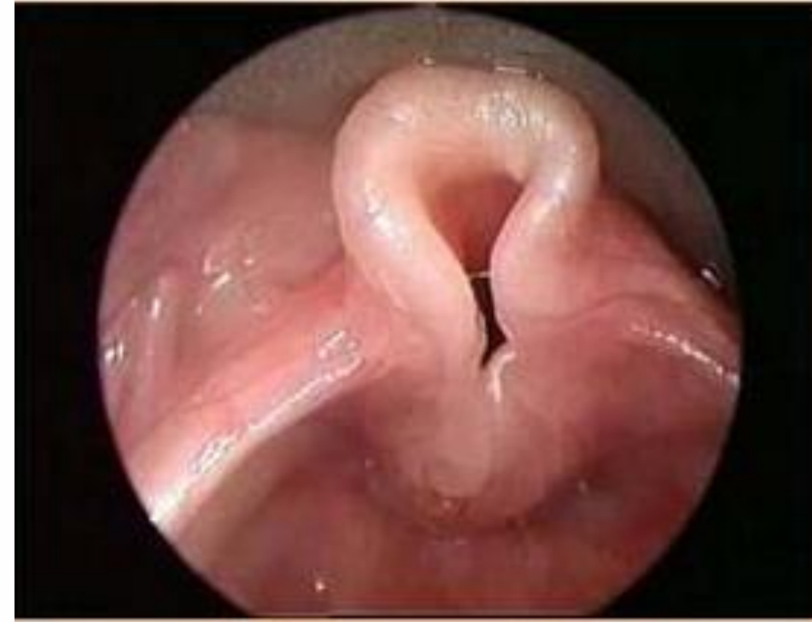
AIRWAY CONCERNS

- Airway concerns include breathing, voice and swallowing
- Larynx
  - Laryngomalacia
  - Laryngeal web
  - Vocal cord nodules
  - Vocal cord paralysis
  - Laryngeal cleft
  - Subglottic stenosis
- Trachea
  - Tracheomalacia
  - Tracheo-Esophageal Fistula (TEF)
  - Vascular ring

LARYNX

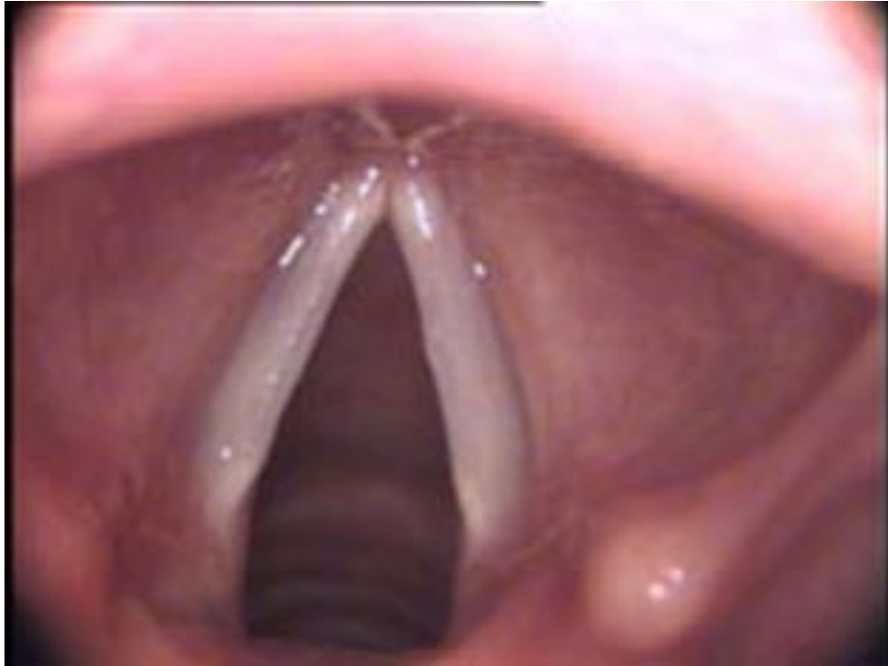


Normal

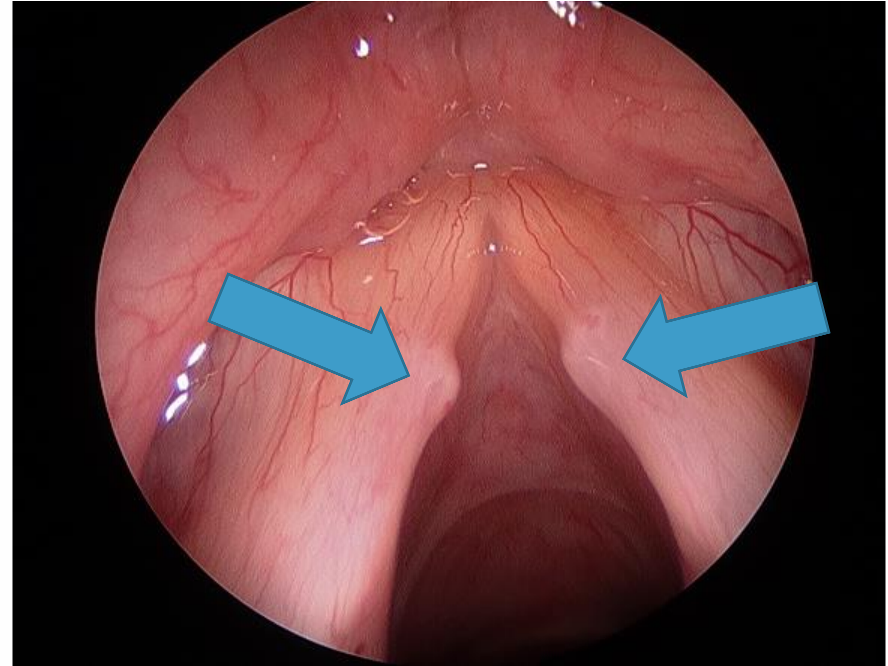


Laryngomalacia

LARYNX

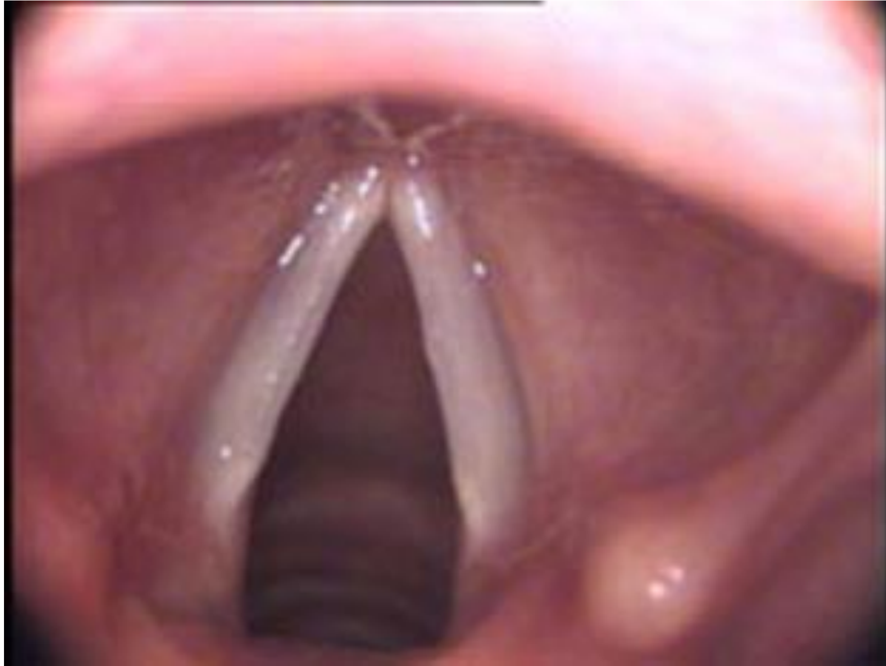


Normal

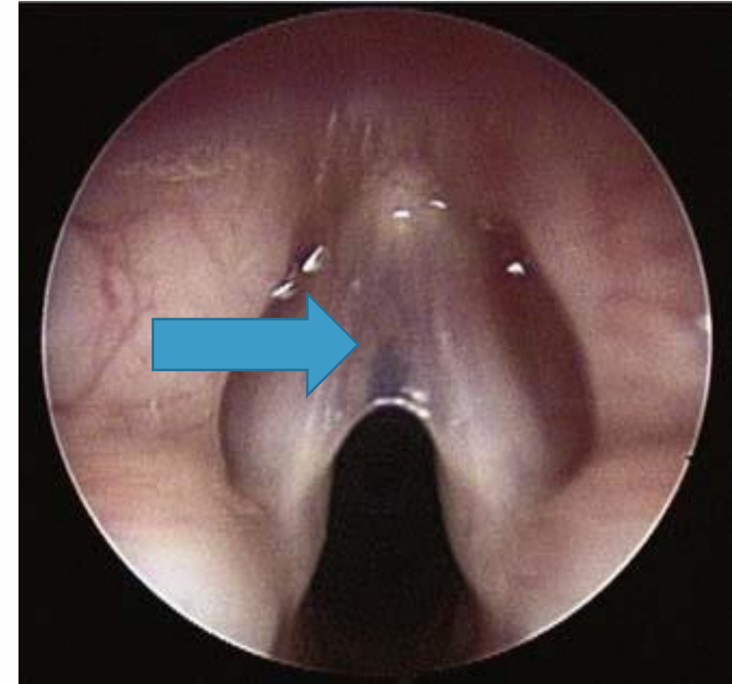


Vocal Cord Nodules

LARYNX

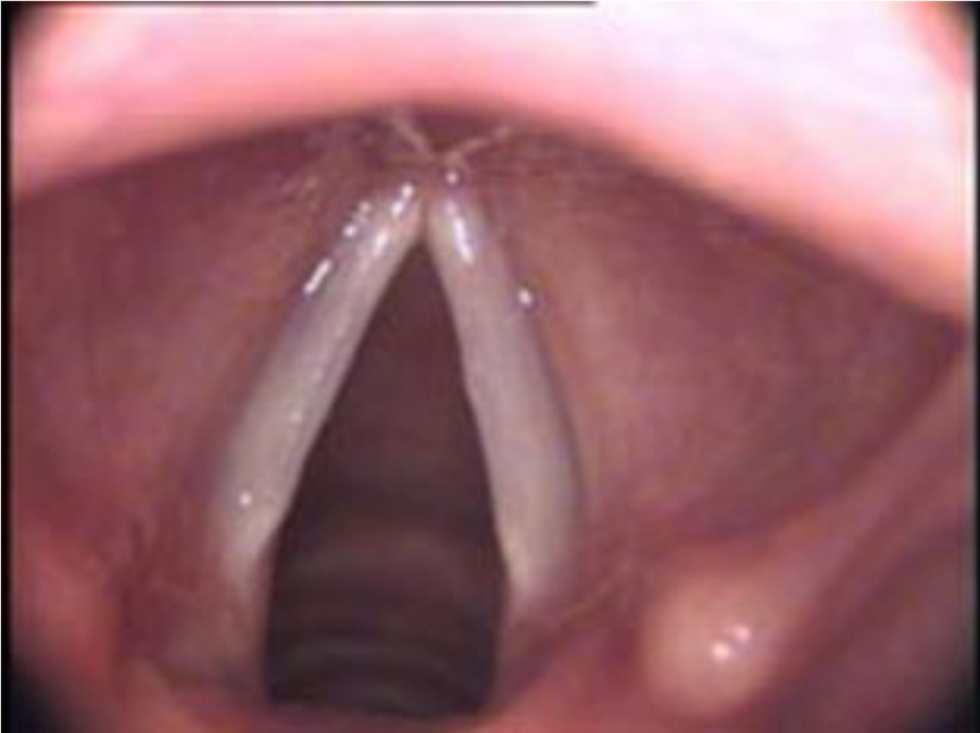


Normal

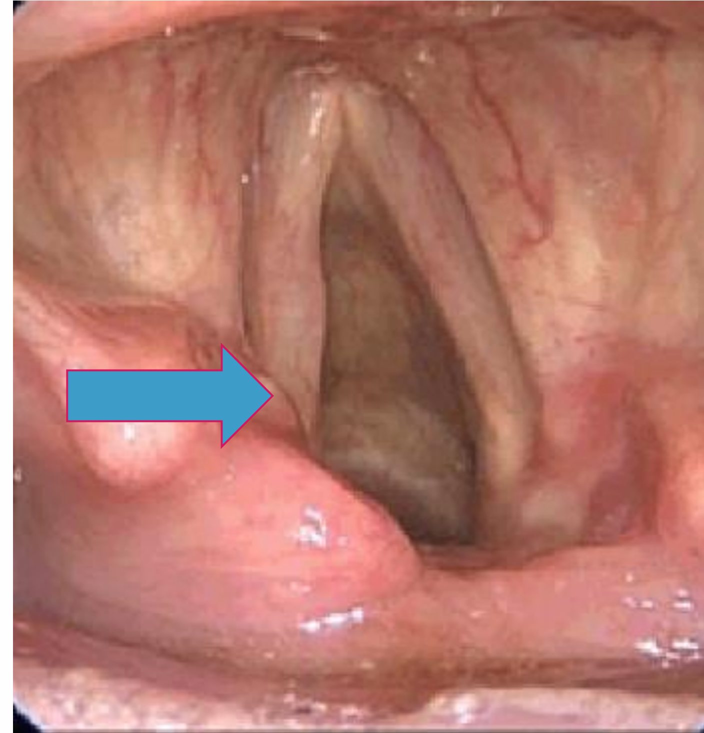


Laryngeal Web

VOCAL CORD MOVEMENT

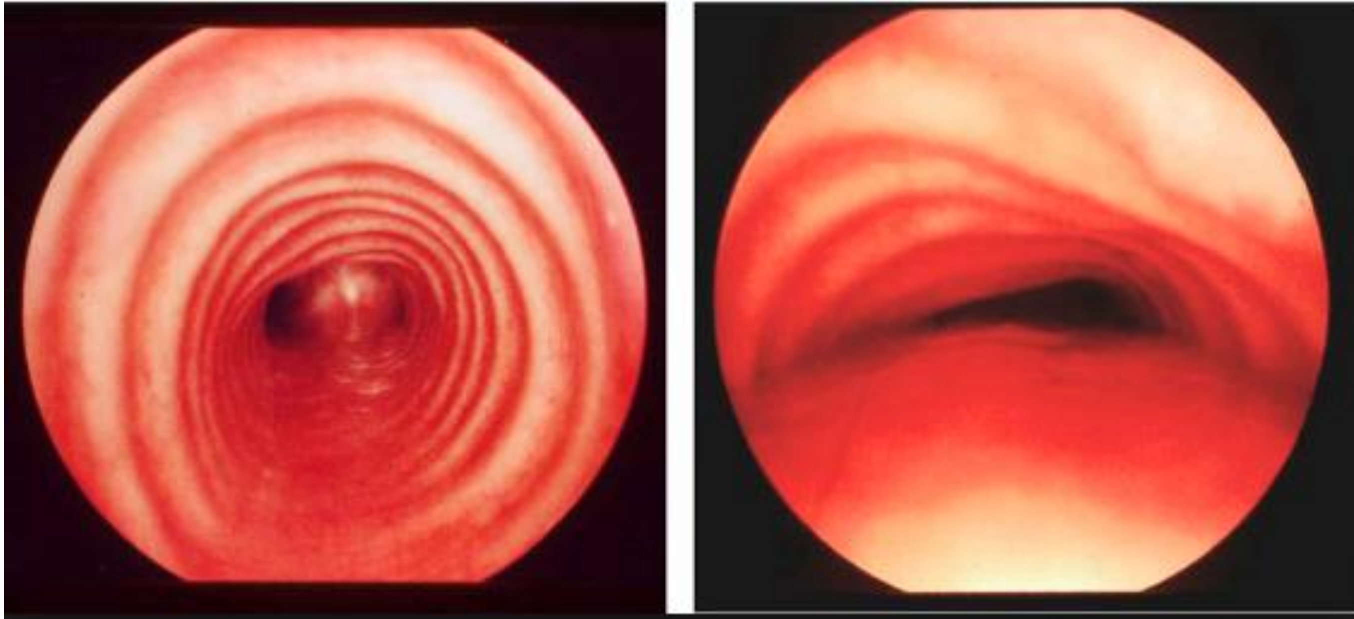


Normal opening



L Vocal Cord Paralysis

TRACHEA



Normal Tracheomalacia

AIRWAY MANAGEMENT

- Laryngomalacia and Tracheomalacia most often resolve over time
- Vocal nodules usually respond to voice therapy—surgery rarely needed
- Laryngeal web/cleft may require surgical correction
- Vocal cord paralysis
  - May resolve over time if nerve is still intact
  - Contralateral cord compensates
  - Injections?
  - Bilateral paralysis may require tracheostomy, re-innervation surgery
- Subglottic stenosis
  - Child may outgrow
  - May require tracheostomy

AIRWAY – YOUR JOB

- Report stridor (noisy breathing) to your physician
- Report **chronic** hoarseness to your physician
- Advocate for an evaluation by ENT (Otolaryngology)

EAR NOSE &THROAT AND 22q

- Frequently affected in children with 22q
- Can have serious consequences for health
  - Breathing
  - Hearing
  - Speech
- Bring up these issues with your physician
- Advocate for your child
- Educate your clinical team

THANK YOU