BRANCHIAL ANOMALIES

David Gleinser, MD
Faculty Advisor: Harold Pine, MD
The University of Texas Medical Branch (UTMB Health)
Department of Otolaryngology
Grand Rounds Presentation
September 30, 2011
Embryology

- Branchial anomalies result from improper development of the branchial apparatus

- Branchial apparatus develops 2nd-6th week
  - Neck is shaped like a hollow tube with circumferential ridges = Arches (mesoderm)

- Ridges between arches = Clefts and Pouches
  - Clefts = outside (ectoderm)
  - Pouches = inside (endoderm)
  - “CAP”
Lateral tongue swellings

Thyroid diverticulum

Mandibular arch

Tub. impar

Second arch

Third Copula arch

Fourth arch

Entrance to larynx
Arches

- Each arch contains
  - Cartilage
  - Cranial nerve
  - Artery
  - Muscle component

- All neural crest origin

- 6 arches, only 5 form structures in humans
  - 1, 2, 3, 4, and 6
  - 5th fails to develop
1st Arch "Mandibular Arch"

- **Skeletal components**
  - Meckel’s cartilage
    - Framework for mandible
  - Malleus head and neck
  - Incus body and short process

- **Muscles**
  - Muscles of mastication
  - Anterior digastric
  - Mylohyoid
  - Tensor tympani
  - Tensor veli palatini

- **Nerve**
  - CN V (Trigeminal)

- **Artery**
  - Maxillary; external carotid
2nd Arch “Hyoid Arch”

- **Skeletal components**
  - Reichert’s cartilage
    - Stapes
    - Malleus manubrium
    - Incus long process
    - Styloid process
    - Hyoid bone (lesser horn and upper body)

- **Muscles**
  - Facial expression, buccinator, platysma, stapedius, stylohyoid, posterior digastric

- **Nerve**
  - CN VII (Facial)

- **Artery**
  - Stapedial
3rd Arch

- **Skeletal components**
  - Hyoid (greater horn and lower body)

- **Muscles**
  - Stylopharyngeus

- **Nerve**
  - CN IX (Glossopharyngeal)

- **Artery**
  - Common/Internal carotid
4th Arch

- **Skeletal components**
  - Thyroid, epiglottic, cuneiform cartilages

- **Muscles**
  - Cricothyroid, inferior constrictors

- **Nerve**
  - Superior laryngeal

- **Artery**
  - Subclavian, aortic arch
6th Arch

- **Skeletal components**
  - Cricoid, arytenoids, corniculate

- **Muscles**
  - All intrinsic muscles of larynx (except cricothyroid)

- **Nerve**
  - Recurrent laryngeal

- **Artery**
  - Pulmonary artery
Branchial Clefts and Pouches

- 4 clefts and 4 pouches
  - 5th and 6th contribute to the 4th

- Clefts provide “covering” to structures of the corresponding arch and pouch
Pouches

- **1st Pouch**
  - Eustachian tube, middle ear, mastoid, inner layer of tympanic membrane

- **2nd Pouch**
  - Tonsils, root of tongue, foramen cecum, pharynx(part)

- **3rd Pouch** – ventral and dorsal wings
  - Ventral wing – Thymus
  - Dorsal wing – inferior parathyroid glands
Pouches

- **4th Pouch**
  - Superior parathyroid glands
  - Parafollicular C-cells of thyroid gland

- **5th Pouch**
  - Contributes to Parafollicular C-cells

- **6th Pouch**
  - Contributes to laryngeal musculature and cartilage
1st Arch Anomalies

- Involves malformations of eyes, ears, palate, and mandible

- 2 main manifestations of “First Arch Syndrome”
  - Treacher Collins Syndrome
  - Pierre Robin Syndrome
Treacher Collins Syndrome

- Mandibulofacial dysostosis
- Inherited AD
- Features
  - Midface and mandibular hypoplasia
  - Ear anomalies: microtia, anotia, stenotic or atresia of EAC, malformation of malleus and incus (CHL)
  - Eye anomalies: coloboma of lower lids, down-sloping palpebral fissures
  - Cleft palate
Treacher Collins Syndrome
Pierre Robin Syndrome

3 Main features

- Micrognathia (small mandible)
- Glossoptosis (posterior displacement/retraction of tongue)
- Cleft palate (U-shaped)
2nd Arch Anomalies

- Malformed auricle
  - Microtia
- Ossicular malformation
  - Stapes, malleus, incus
  - CHL
- Muscular asymmetry of face
- Hyoid malformation
  - lesser horn and upper body
3rd Arch Anomalies

- Hyoid anomalies
  - Lower body
  - Greater horn

- Aneurysm of carotid artery
4th Arch Anomalies

- Laryngeal stenosis
- Laryngoptosis (low position of larynx)
- Chondromalacia
- Double aortic arch
- Pulmonary artery sling
  - Left pulmonary artery originates from right pulmonary artery
  - Slings around right main-stem bronchus
Double Aortic Arch
Pulmonary Artery Sling
1st Pouch Anomalies

- Atretic eustachian tube -> recurrent OM
- ET diverticuli
- Absence
  - Tympanic cavity
  - Mastoid antrum
- Perforated TM
- Bifid tongue
- Branchiogenic nasopharyngeal cysts (very rare)
2nd Pouch Anomalies

- Thyroglossal duct cyst
  - 7% of population
  - Failure of ablation of TGD
  - Anywhere from base of tongue to upper mediastinum

- Typical finding
  - Cystic lesion just below hyoid in midline that moves with deglutination and tongue protrusion
TGD Cyst

- May contain thyroid tissue
  - Potentially the only functioning thyroid
- Perform U/S or CT to look for thyroid and to assess lesion
- Treatment – surgical
- May contain cancer
  - 1%
  - Papillary carcinoma
TGD Cyst
2nd Pouch Anomalies

- Lingual Thyroid
  - Failure of decent of thyroid -> atopic
    - 90% of cases at the base of tongue (lingual thyroid)
  - 4:1 female:male
  - Usually not noted until teenage or young adult
  - Asymptomatic (most cases); dysphagia, airway compromise
  - Reddish mass (well vascularized) at base of tongue
Lingual Thyroid

- Hypothyroidism – 70% of cases
- 2/3 cases – only functioning thyroid tissue
  - Thyroid function study prior to treatment
- Treatment
  - Asymptomatic – Monitor
  - Symptomatic
    - Excise +/- transplant tissue into muscles of neck
    - Radioiodine therapy (destroys all thyroid tissue)
  - Usually require lifelong thyroid replacement
Lingual Thyroid
Lingual Thyroid
Lingual Thyroid
3rd and 4th Pouch Anomalies

- DiGeorge Syndrome
  - Congenital absence of thymus and parathyroids
  - Partial deletion of chromosome 22
  - CATCH-22
    - Cardiac anomalies
    - Abnormal facies
    - Thymic aplasia
    - Cleft palate
    - Hypocalcemia
  - Tetany and impaired cellular immunity (T-cells)
3rd and 4th Pouch Anomalies

- Accessory or undecended parathyroid glands
- Thymic cysts
Branchial Cleft Anomalies

1\textsuperscript{st} Cleft

- Cysts, EAC atresia or stenosis, pits of lower lips, preauricular sinuses or tags
Branchial Cleft Anomalies

- **2nd Cleft**
  - Cysts
  - Cervical sinuses

- **3rd Cleft**
  - Cysts (rare)
  - Thymic cysts

- **4th Cleft**
  - Cysts (extremely rare)
  - Cysts on the Vagus nerve -> cough
Branchial Cleft Cysts

- Results from failed obliteration of branchial clefts
- 2-3% are bilateral
- 2\textsuperscript{nd} cleft cyst is the most common type
  - ~95% of cases
1st Branchial Cleft Cyst

- **Work Classification**
  - **Type I**
    - Preauricular mass or sinus
    - Ectoderm
    - Sinus tract is anterior and medial to the EAC
      - Preauricular region → Lateral to CN VII → Parallels EAC
        → Ends in EAC or middle ear
  - **Type II**
    - More common than Type I
    - Presents at the angle of mandible or submandibular region
      - Angle of mandible -> Lateral or medial to CN VII -> Ends in concha or bony-cartilaginous junction of EAC.
1st Brachial Cleft Cyst Type I
2nd Branchial Cleft Cyst

- Most common branchial cyst
- Presents as a mass just anterior and medial to the SCM in the neck

- Tract
  - Anterior neck -> Along carotid sheath -> Between external and internal carotid arteries -> **superficial** to CN IX and XII -> Opens into tonsillar fossa
2nd Branchial Cleft Cyst
3\textsuperscript{rd} Branchial Cleft Cyst

- Closely associated with the thyroid gland
  - If patient with recurrent thyroid abscesses, consider diagnosis

- Usually on the left

- Tract:
  - Lateral neck (similar or lower location than 2\textsuperscript{nd}) -> Deep to carotids -> Deep CN IX, superficial to CN XII, Superficial to superior laryngeal nerve -> Pierces thyrohyoid membrane -> Opens into apex of pyriform sinus
3rd Branchial Cleft Cyst
4th Branchial Cleft Cyst

- Very rare
  - ~ 200 cases reported in the literature
- Also associated with recurrent thyroid abscesses
- Theoretical path of tract:
  - Low in neck (anterior to SCM) -> Deep to common carotid -> Loops around aortic arch on the left (subclavian on the right) -> Deep to superior laryngeal nerve -> Superficial to recurrent laryngeal nerve -> Opens into pyriform sinus
Work-up

- **Ultrasound**
  - Round mass with uniform low echogenicity and lack of internal septations
  - Advantages: No radiation, no sedation for children, low cost
  - Not typically ordered alone

- **CT**
  - Homogeneous lesion with low attenuation centrally and a smooth enhancing rim
  - Often part of the work-up
  - More radiation, higher cost, may require sedation (children)
Work-up

- **MRI**
  - Hypointense on T1 and hyperintense on T2
  - Advantages: No radiation
  - Disadvantages: Sedation for children, very expensive

- **Fluroscopic fistulography or CT fistulography**
  - Inject radiopaque dye into the fistula or sinus to delineate course

- **Barium swallow esophagography**
  - Help locate fistula tract in type 3 and 4 anomalies

- **FNA**
  - Usually only done if suspect cancer
  - May cause cyst to collapse -> much harder to remove at time of surgery
Treatment – Infected Cyst

- **Antibiotics**
  - Should cover respiratory flora and Staph aureus (broad spectrum)
  - Cover 2-4 weeks

- **Abscess**
  - Consider needle aspiration to drain
    - May work without causing as much scaring as I&D
  - I&D if needle aspiration doesn’t work

- Once infection cleared, operate
Treatment - Surgical

- Complete surgical excision of tract and cyst is treatment of choice in most cases

- 1st cysts
  - Must identify facial nerve as tract is usually associated with it
  - If possible, wait till 2 years of age
    - Mastoid tip defined
    - Facial nerve larger and deeper
    - Controversy: waiting can lead to more infections → more scar → more difficult surgery
  - Lacrimal probes can help locate tract
Treatment - Surgical

- 3\textsuperscript{rd} and 4\textsuperscript{th} cysts
  - Must identify the recurrent laryngeal nerve as closely associated (will be deep to tract)
  - Removal of ipsilateral thyroid lobe is advocated to ensure complete removal of tract
  - Perform DL to examine pyriform sinus
    - Fogarty vascular catheter can be placed through the sinus tract
Endoscopic Cauterization of Pyriform Sinus Opening

- Literature describes this for treatment of 4th sinus tracts, but has been performed with 3rd cleft anomalies

- Recommendation
  - Performed alone
  - Performed with surgical resection of cyst and tract
Endoscopic Cauterization of Pyriform Sinus Opening

- Verret et al
  - Performed endoscopic cauterization of sinus in 10 children with 4th branchial cleft anomalies (no surgical excision!)
    - Dilated sinus opening with balloon catheter → cautery with electrocautery ball coagulator
    - 7 showed no recurrent disease after 3 years
    - 3 lost to F/U
Sources


