Management of Craniofacial Anomalies

Steven Goudy M.D.
Pediatric Otolaryngology
Emory University
Epidemiology

• Syndromic Clefting
  – 30% of cases
  – Mendelian Inheritance
  – Over 300 Syndromes described

• Non-Syndromic Clefting
  – 70% of cases
  – Non-Mendelian inheritance
  – Genetic and environmental forces play a role:
    Monozygotic twins 25-40% concordance, where
dizygotic twins 3-6% concordance for CLP
Associated Anomalies

• Cleft Lip +/- Palate
  – 1-2/1000 live births
  – Other anomalies in 10-30%
  – Heart defects in 4-10%
  – Males more commonly affected

• Cleft Palate Only
  – 1/2500
  – Other anomalies in 15-60%
  – Heart defects in 7-10%
  – Females more commonly affected
Patterns of Clefting

• Multigenic/Complex  70-80%
• Mendelian (syndromic) 6%
  – Stickler Syndrome, Van der Woude Syndrome
• Chromosomal Anomalies 3%
  – Velo-cardio-facial Syndrome
• Unknown Patterns  10%
Non-Syndromic Clefting Risk

- Offspring of affected parent = 3%
- Sibling of affected person = 5%
- Sibling of 2 affected people = 10%
- Affected parent and sibling = 14%

- Estimates predict that 4-20 genes are responsible for clefting, giving genetic heterogeneity
Embryonic Development

- Facial Primordia begins during the 4th week of gestation from dorsal neural crest migration.
- Paired Branchial Arches develop at the 7th week.
- Medial and Lateral Nasal prominences fuse in the midline with the maxillary prominence, creating the lip and primary palate which is complete by the 12th week.
Palate Fusion
Abnormal Fusion = Cleft
Prenatal Diagnosis
Cleft Team

- Dental
- Speech
- Surgeon
- Audiology

Patient
Feeding

• Babies with cleft palate will NOT be able to breast feed (in general)
• Inability to separate oral and nasal cavities makes it impossible to generate necessary suction
• Specialized bottles make it possible to effectively feed babies with cleft palates
Cleft Specific Bottles
Pierre Robin Sequence

• Definition
  – Micrognathia, cleft palate, glossoptosis
• Tongue Based Airway obstruction
• Treatment Options
  – Positioning
  – Trumpet
  – Mandibular distraction
Patient Presentation

• Newborn child with severe airway obstruction and desaturations
• Unable to directly intubate patient
• Tracheotomy after LMA intubation
• Evaluation for mandibular distraction
• Decannulation after distraction
Pre-surgical Planning
Mandibular Distraction
Mandibular Distraction
Timing of Cleft Repair

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Initial Procedure</th>
<th>Second Procedure (Bilateral)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lip adhesion</td>
<td>2-4 wk</td>
<td>3 mo</td>
</tr>
<tr>
<td>Lip repair</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Primary</td>
<td>2-3 mo</td>
<td>4-5 mo</td>
</tr>
<tr>
<td>After adhesion</td>
<td>5-7 mo</td>
<td>7-10 mo</td>
</tr>
<tr>
<td>Palate repair</td>
<td></td>
<td>12-30 mo</td>
</tr>
<tr>
<td>Correction of velopharyngeal incompetence</td>
<td></td>
<td>4 yr and older</td>
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<tr>
<td>Orthodontic therapy</td>
<td></td>
<td>4 yr and older</td>
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<tr>
<td>Lip revision</td>
<td></td>
<td>4 yr and older</td>
</tr>
<tr>
<td>Premaxillary recession</td>
<td></td>
<td>5 yr</td>
</tr>
<tr>
<td>Nose</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Tip</td>
<td></td>
<td>6-10 yr</td>
</tr>
<tr>
<td>Dorsum and septum</td>
<td></td>
<td>15-17 yr</td>
</tr>
</tbody>
</table>
Lip Taping and Nasoalveolar Molding
Custom Designed Taping
Cleft Lip Repair
All Grown Up
Cleft Palate Repair

- Typically occurs at 1 year of age
- May vary according to comorbid conditions
- Trade off between maxillary growth potential vs. speech development
Dermal Matrix in Palate Repair
Adjuvant Treatments in Palate Repair
Adjuvant Treatments in Palate Repair

**Urinary Bladder Matrix (UBM)**

- Core Structure: bimodal surface
  - Epithelial basement membrane
  - Lamina propria
  - Growth factors

- Healing Response
  - Autologous progenitor stem cells
  - Chemotactic and mitogenic activity
  - Antimicrobial activity
  - Site-specific tissue remodeling
Wound Healing Complication
Craniosynostosis

- Represents *premature fusion* of bony plates of the skull
- Often associated with syndromes (Apert, Pfeiffer)
- May involve one or multiple sutures
- Skull asymmetry becomes increasingly obvious
- Surgical correction required
Craniosynostosis
Ear Function and Hearing

- Eustachian tube dysfunction is common
- Majority of patients with cleft palate have otitis media
- Many patients may fail NBHS
- Ear tubes are placed at the time of surgery
- Average frequency of ear tubes placement is 3 times
1 screening
Hearing screening on all infants before 1 month of age

3 diagnosis
Audiological diagnosis before 3 months of age if the infant does not pass the screening

6 intervention
Medical, educational, and audiologic intervention before 6 months of age if diagnosed with hearing loss
Ear Development
Ear Development

A
- Roof of rhombencephalon
- Endolymphatic duct
- Utricular portion of otic vesicle
- Saccular portion
- Mesenchymal condensation
- First pharyngeal cleft
- Tubotympanic recess

B
- Auditory ossicles embedded in loose mesenchyme
- Auditory tube
- Ext. auditory meatus
- Meatal plug
- Primitive tympanic cavity
Ear Tubes

- All children with cleft palate have chronic otitis media with effusion
- All children with cleft palate should have ear tubes placed
- They on average need 3 sets of ear tubes
- 50% of children’s ears will normalize after palate repair
- Improvement of ear function relates to reposition of levator and tensor palatini muscles
Eustachian tube dysfunction

- Eustachian tube in children is shorter, more narrow, and more horizontal in position.
Otitis Media– Risk Factors

- Age under 3 years
- Cigarette smoke exposure
- Day care
- Sibling with recurrent OM
- Craniofacial anomaly (cleft palate)
- Immunodeficiency
Otitis Media

Normal TM

Acute Otitis Media

Chronic Otitis
Tubes
Alveolar Bone Grafting

- **Primary**
  - Done at time of palate repair
  - Typically done with rib
  - If using pre-surgical repositioning may not need much or any bone

- **Secondary**
  - Done at mixed dentition age (6 or 7)
  - Usually when canine is erupting or will erupt
  - Source of bone usually iliac crest
  - Success depends on hygiene and whether bilateral or unilateral
Autogenous Bone Grafting

• Source
  – Iliac crest most common; lots of marrow present
  – Mandible/tibia less common, limited bone stock

• Disadvantages
  – Additional OR time and hospitalization
  – Risk of fracture

• Alternatives
  – Dimeneralized bone: risk of infection, poor efficacy
  – No FDA approve alternative, although BMP2 is used off label, and also use of Platelet Rich Plasma with and without stem cells
Bone Grafting
Bone Graft Market

Market Segmentation


- Machined Bone Allograft
- Standard Bone Allograft
- Demineralized Bone Matrix (DBM)
- Bone Graft Substitutes
- Bone Morphogenetic Protein (BMP)
- Cell Based Matrices
- Platelet Concentration System
Currently available BMP2 Product
Stem Cells in Clinical Trials

Mesenchymal Stem Cell

- Osteoblasts
- Adipocytes
- Chondrocytes
Velopharyngeal Insufficiency

- Normal speech pattern uses velopharyngeal port to separate oral cavity from nasopharynx.
- Velopharyngeal competence allows the creation of plosive sounds, such as P, S, and F.
- Inability to close the velopharyngeal port occurs in 20% of patients with cleft repair.
- This condition also occurs in patients with Velocardiofacial syndrome and DiGeorge Syndrome (22q11 deletion).
VPI

- Surgical correction of VPI in cleft patients typical around 4 years of age
- Tailor surgery to velopharyngeal port closure
- Pharyngeal Flap
- Sphincter pharyngoplasty
- Furlow Palatoplasty
Humanitarian Opportunities

• High rate of clefting in Asia and central and south America
• Can perform numerous surgeries in short period of time
• Timing of repair is not same as that observed here in the US