Cleft Lip & Cleft Palate

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Objectives

1. Describe different types of cleft lip and cleft palate variations
2. Understand the embryological development of cleft lip and cleft palate
3. Understand current modalities of diagnosing cleft lip and/or cleft palate prenatally
There is no financial interest with any organization that could be perceived as a real or apparent conflict of interest in the context of the subject of this activity.
What is a Cleft Lip?
• Separation of the two sides of the lip
• Often includes bones of the maxilla and/or alveolus
• Ranges from a slight notch in the pink vermilion of the lip to complete separation in one or both sides of the lip, extending into the nose

What is a Cleft Palate?
• Opening in the roof of the mouth where the two sides of the palate did not fuse as the baby was developing
Embryology

- **Processes** = programmed to grow, move, and join with each other to form a part of the embryo
- Normally processes grow into an open space by cellular migration and multiplication, touch each other, and fuse together.

- Clefts result when processes do not reach each other in time and an open space (cleft) between them persists

Embryology

**4th wk:** frontonasal prominence, **maxillary processes** & **mandibular processes** surround primitive oral cavity

**5th wk:** nasal pits have formed, which leads to formation of paired **medial** and **lateral nasal processes**

**6th wk:** **medial nasal processes** merge with **maxillary processes** to form upper lip & primary palate

**Lateral nasal processes** form nasal alae; **mandibular processes** fuse to form mandible

**6th wk:** secondary palate develops as bilateral outgrowths from **maxillary processes**, which grow vertically down the side of the tongue

**8-12th wk:** Palatal shelves elevate to horizontal position above tongue, contact one another & begin to fuse

**8-12th wk:** Fusion of the palatal shelves divides the oronasal space into separate oral and nasal cavities

Embryology

- Unilateral cleft lip
  - Lack of fusion of maxillary process & medial nasal processes
- Primary cleft palate
  - Unsuccessful fusion of median palatine process
- Secondary cleft palate
  - Unsuccessful fusion of lateral palatine processes to each other & with nasal septum
- Macrostomia
  - Unsuccessful fusion of maxillary & mandibular processes

In general, any factor that can prevent the processes from reaching each other by slowing down migration and/or multiplication of neural crest cells, by stopping tissue growth and development or by killing established cells can cause a cleft.
Epidemiology

• Cleft lip:
  • Native American 3.7 in 1000 live births
  • Asian 2.1 in 1000 live births
  • White 1 in 1000 live births
  • Black 0.41 in 1000 live births

• Increasing incidence with both parents > 30yo

• Cleft lip & palate 1/1000 births 45%
• Isolated cleft palate 1/2500 births 35%
• Isolated cleft lip 20%
• Submucous cleft palate 1/1200 births
Epidemiology

- Most common type of cleft palate is a bifid uvula (2% population)
  - May be associated with submucous cleft, levator dehiscence, VPI
- 2nd most common cleft palate is left unilateral complete
- Cleft palate: 0.5 in 1000, consistent among races
- Submucous cleft palate
  - Weatherly-White studied 10,836 children
    - 9 had submucous cleft
    - 1 in 1200
Predisposing Factors

• Environmental factors
  • Teratogens: alcohol, anticonvulsants, retinoic acid, thalidomide
  • Smoking
  • Altitude
  • Viral infection (e.g., rubella)
  • Folic acid deficiency at conception or during early pregnancy

• Genetic factors
  • Inherited from one or both parents
  • >30 potential candidate loci & genes identified as strong susceptibility genes:
    • MSX1, TGFA, TGFB1, TGFB2, TGFB3, RARA, & MTHFR are among the strongest

• Syndromic Clefts
  • Syndrome = abnormality in genes on chromosomes that result in malformations or deformities that form a recognizable pattern
  • Cleft lip ± cleft palate is part of >400 syndromes
  • ~30% of all clefts are syndromic

• Non-syndromic Clefts
  • Only 1 defect or multiple anomalies that are the result of a single initiating event or primary malformation

Genetic Factors

- > 40% of isolated cleft palates are part of malformation syndromes
- Most common syndrome associated with cleft lip and palate together = van der Woude syndrome (think lip pits)
- Most common syndrome linked with cleft palate = velocardiofacial syndrome
### Genetic Counseling

#### Affected Relatives

<table>
<thead>
<tr>
<th>Condition</th>
<th>Predicted Outcomes*</th>
</tr>
</thead>
<tbody>
<tr>
<td>CL±CP</td>
<td></td>
</tr>
<tr>
<td>One sibling</td>
<td>≈ 4%</td>
</tr>
<tr>
<td>One Parent</td>
<td>≈ 4%</td>
</tr>
<tr>
<td>Sibling and a Parent</td>
<td>≈ 16%</td>
</tr>
<tr>
<td>CP</td>
<td></td>
</tr>
<tr>
<td>One Sibling</td>
<td>≈ 2.4%</td>
</tr>
<tr>
<td>One Parent</td>
<td>≈ 2.4%</td>
</tr>
<tr>
<td>Sibling and a Parent</td>
<td>≈ 15%</td>
</tr>
</tbody>
</table>

#### Counseling Risks for Cleft Lip with or without Cleft Palate (CL ± CP and Cleft Palate (CP)) for Various Situations

<table>
<thead>
<tr>
<th>Situation</th>
<th>Proband Has CL ± CP</th>
<th>Proband Has CP</th>
</tr>
</thead>
<tbody>
<tr>
<td>Frequency of defect in the general population</td>
<td>0.1%</td>
<td>0.04%</td>
</tr>
<tr>
<td>My spouse and I are unaffected</td>
<td></td>
<td></td>
</tr>
<tr>
<td>1. We have an affected child.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>What is the probability that our next baby will have the same condition if:</td>
<td></td>
<td></td>
</tr>
<tr>
<td>a. We have no affected relatives?</td>
<td>4%</td>
<td>2%</td>
</tr>
<tr>
<td>b. There is an affected relative?</td>
<td>4%</td>
<td>7%</td>
</tr>
<tr>
<td>c. Our affected child also has another malformation?</td>
<td>2%</td>
<td>2%</td>
</tr>
<tr>
<td>d. My spouse and I are related?</td>
<td>4%</td>
<td></td>
</tr>
<tr>
<td>What is the probability that our next baby will have some other sort of malformation?</td>
<td>Same as general population</td>
<td></td>
</tr>
<tr>
<td>2. We have two affected children.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>What is the probability that our next baby will have the same condition?</td>
<td>9%</td>
<td>1%</td>
</tr>
<tr>
<td>I am affected (or my spouse is).</td>
<td></td>
<td></td>
</tr>
<tr>
<td>1. We have no affected children.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>What is the probability that our next baby will be affected?</td>
<td>4%</td>
<td>6%</td>
</tr>
<tr>
<td>2. We have an affected child.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>What is the probability that our next baby will be affected?</td>
<td>17%</td>
<td>15%</td>
</tr>
</tbody>
</table>
Classification

<table>
<thead>
<tr>
<th>Normal</th>
<th>Cleft lip</th>
<th>Bilateral cleft lip</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>Cleft lip</td>
<td>Bilateral cleft lip</td>
</tr>
<tr>
<td>Cleft palate</td>
<td>Cleft lip with partial palate involvement</td>
<td>Bilateral cleft lip with full palate involvement</td>
</tr>
</tbody>
</table>

- Unilateral vs. bilateral
- Complete vs. incomplete
Prenatal Diagnosis

- Ultrasound in 2\textsuperscript{nd} trimester when the position of fetal face is located correctly
- Diagnosing a cleft palate is more difficult, less specific → 3D

http://www.glowm.com (Global Library of Women’s Medicine)
Prenatal Diagnosis

• European study (2000):
  • 709,027 births, 7758 cases with congenital malformations
  • 751 cases reported with facial clefts
    • 553 cleft lip ± cleft palate [CL(P)] and 198 cleft palate [CP]
  • Accurate diagnoses:
    • CL(P) in 65/366 cases with an isolated malformation (18%)
    • CL(P) in 30/89 cases with multiple malformations (34%)
    • CL(P) in 32/62 cases with chromosomal anomaly (52%)
    • CL(P) in 21/36 syndromic cases (58%)
    • CP was made in 13/198 cases (6%)

Prenatal Diagnosis

- Axial 3D ultrasound of fetal palate has high accuracy in identifying prenatal cleft palate when cleft lip is diagnosed at mid-trimester 2D ultrasound screening (Baumler, et al. 2011)
  - Sensitivity 100%, specificity 90%
  - Multiplanar mode display was used for off-line analysis of (1) the primary palate in the coronal plane at the base of the retronasal triangle and (2) the secondary palate by virtual navigation in the axial plane.

- 100% cases of primary cleft palate, 86% cases of secondary cleft palate were visualized using 3D ultrasound with satisfactory false positive rate (Martinez-Ten, et al. 2011)
  - Virtual navigation of the fetal palate using the multiplanar mode display seems to be useful in the diagnosis of clefting in the 1st trimester

- Diagnosis of cleft lip was 95% (false-positive 7.7%); cleft hard palate 89.7% (FPR 15.6%); cleft alveolar ridge 84.5% (FPR 7.2%) (Sommerlad 2010)
  - For the 3D examination, lips & alveolar ridges were examined both in profile & in the frontal plane. The palate was assessed in reverse coronal view by rotating the face through 180° on the vertical axis to produce the 3D-RF view.

Prenatal Consultation

- Internet information:
  - Much of the information is useful, but none is filtered
  - Some information is grossly inaccurate
  - No interaction with trained professional
  - Empowers families to gather their own information, but may heighten anxiety
- Copies of ultrasound photos & ultrasound report
- Consultation
  - Treatment plan overview
  - View pre-op and post-op photos of unilateral/bilateral clefts of varying severity
  - Timeline for surgery
  - Overview of team care
  - Emotional preparation for having a child with a cleft lip/palate
  - Help with navigating first critical weeks after birth/adoption, minimizing likelihood of unexpected or significant medical interventions
- All families felt that this was helpful in understanding the situation (Matthews et al, 1998)
- All parents who had the opportunity to talk with the cleft team felt that the experience was valuable and would recommend it to a friend (Matthews et al, 1998)

At Birth...

1. Risk of aspiration due to communication between oral and nasal cavities
2. Airway obstruction (esp. Pierre Robin sequence with cleft palate + micrognathia and normal tongue)
3. Difficulties with feeding due to nasal regurgitation

- Influenced by other major/minor anomalies that may represent 1 of 400 known cleft syndromes
The Early Years...

• Breathing
  • When the palate and jaw are malformed, breathing becomes difficult. Treatments include surgery and oral appliances.

• Feeding
  • Problems with feeding are more common in cleft children. A nutritionist and speech therapist that specializes in swallowing may be helpful. Special feeding devices are also available.

• Ear infections and hearing loss
  • Any malformation of the upper airway can affect the function of the Eustachian tube and increase the possibility of persistent fluid in the middle ear, which is a primary cause of repeat ear infections. Hearing loss can be a consequence of repeat ear infections and persistent middle ear fluid. Tubes can be inserted in the ear by an otolaryngologist to alleviate fluid build-up and restore hearing.

• Speech and language delays
  • Normal development of the lips and palate are essential for a child to properly form sounds and speak clearly. Cleft surgery repairs these structures; speech therapy helps with language development.

• Dental problems
  • Sometimes a cleft involves the gums and jaw, affecting the proper growth of teeth and alignment of the jaw. A pediatric dentist or orthodontist can assist with this problem.
Feeding

Because of the cleft, baby is unable to create suction needed to efficiently pull milk from the bottle or breast.

- Breastfeeding
  - Most children born with cleft lip and palate are unable to breastfeed
  - In a unilateral cleft, the child may be effectively positioned so the cleft is obstructed by the breast

- May take longer to feed
- May have nasal regurgitation
- May swallow more air while feeding

Improving cleft feeding:
- Feed baby in more upright position to decrease the amount of regurgitated feeds
- Infant sneezing and coughing can clear the nose
- If feeding lasts > 45 min, valuable calories may be burned as infant works hard to feed
- Feeding well every 3-4 hours leads to better weight gain than short feeds every 1-2 hrs
- Burp frequently to clear swallowed air

Higher fistula rates in underweight children

http://www.cleftline.org
Feeding

- Cleft palate nursers

- Orthodontic nipple can be crosscut

http://www.cleftline.org
Multidisciplinary Team

• Comprehensive care:
  • Pediatrician
  • Plastic surgeon
  • Pediatric dentist
  • Orthodontist
  • Otolaryngologist
  • Audiologist
  • Genetic counselor
  • Speech pathologist
  • Maxillofacial surgeon
  • Social worker
  • Psychologist
Cleft Lip

- Microform cleft (forme fruste)
  - Furrow along vertical length of lip
  - Small vermilion notch
  - Minor imperfections in white roll
- Incomplete cleft lip
  - Varying degree of vertical lip separation
  - Intact nasal sill (Simonart band)
- Complete cleft lip
  - Full-thickness defect of lip & alveolus
  - Extends to base of nose
  - Often accompanied by cleft palate
  - Premaxilla typically rotated outward & projects anteriorly
Cleft Palate

- Complete cleft palate
- Partial cleft palate

- Submucous cleft palate
  1. Bifid uvula
  2. Notching of posterior border of hard palate
  3. Muscular diastasis of soft palate with intact mucosal layer (zona pellucidum)
# Treatment Overview

- Cleft lip repair
- Cleft palate repair
- Cleft lip revision
- Alveolar cleft closure & bone grafting
- Closure of palatal fistula
- Palatal lengthening
- Pharyngeal flap
- Pharyngoplasty
- Columellar lengthening
- Cleft lip rhinoplasty & septrhplasty
- Lip scar revision
- LeFort I maxillary osteotomy

| Age | 0m | 3m | 6m | 9m | 1y | 2y | 3y | 4y | 5y | 6y | 7y | 8y | 9y | 10y | 11y | 12y | 13y | 14y | 15y | 16y | 17y | 18y |
|-----|----|----|----|----|----|----|----|----|----|----|----|----|----|-----|-----|-----|-----|-----|-----|-----|-----|
| Palatal obturator | | | | | | | | | | | | | | | | | | | | | |
| Repair cleft lip | | | | | | | | | | | | | | | | | | | | | |
| Repair soft palate | | | | | | | | | | | | | | | | | | | | | |
| Repair hard palate | | | | | | | | | | | | | | | | | | | | | |
| Tympanostomy tube | | | | | | | | | | | | | | | | | | | | | |
| Speech therapy/pharyngoplasty | | | | | | | | | | | | | | | | | | | | | |
| Bone grafting jaw | | | | | | | | | | | | | | | | | | | | | |
| Orthodontics | | | | | | | | | | | | | | | | | | | | | |
| Further cosmetic corrections (Including jaw surgery) | | | | | | | | | | | | | | | | | | | | | |

*Note: The table represents the recommended ages for various procedures.*
Nasoalveolar Molding (NAM)

- Pre-surgical treatment used to improve final results of surgical repair for cleft lip & cleft palate
- Takes advantage of malleability of immature cartilage of the nose
- Non-surgically molds the columella with tissue expansion
Lip Adhesion

- Indications
  - Wide unilateral complete clefts
  - Complete clefts with poorly aligned maxillary segments
  - Bilateral cleft deformity with:
    - Markedly separated elements
    - Alveolar segments displaced
    - Lip margins of cleft under tension when brought together

- Disadvantages
  - Primary repair is more effective
  - Excess scarring & more surgeries

Surgical Management

• Orthodontic treatment as early as a few weeks after birth
• Definitive repair around 3 months
  • 10 lbs
  • 10 Hgb
  • 10 wks old

• Microform clefts
Surgical Management CL

- **Unilateral cleft lip**
  - Straight-Line, Triangular-Flap, Rotation-Advancement Repairs

- **Bilateral cleft lip**
  - Straight-Line, Z-Plasty, Millard, Modified Manchester Repairs
Surgical Management CP

- Two-Flap Palatoplasty

- Double-Opposing Z-Plasties
Post-operative Care

- Oral feedings
  - Uninterrupted breastfeeding after surgery
  - Resume feedings with favorite bottle
- Study shows no complications with breastfeeding post-op
  - Highest weight gain
  - Hospital stay shorter by 1 day
- Activity
  - Avoid pacifiers or toys x 2 wks
  - No-Nos (elbow immobilizers) x 4 weeks
  - Nasal stents x 4 weeks
- Wound care
  - Bathe, clean with cotton swabs & dilute H₂O₂
  - Topical antibiotic ointment
  - Permanent sutures removed day 5-7

VPI (Velopharyngeal Insufficiency)

- Air leaks into nasal cavity resulting in hypernasal voice resonance and nasal emissions while talking
- Speech articulation errors (eg, distortions, substitutions, omissions) and compensatory misarticulations and mispronunciations (eg, glottal stops, posterior nasal fricatives)
- May worsen with adenoid involution at puberty, LeFort I advancement
- Non-operative management:
  - Articulation therapy
  - Sucking & blowing exercises
  - Electrical & tactile stimulation
  - Prosthetic appliances
  - Biofeedback techniques
- Operative management:
  - Augmentation of posterior pharyngeal wall (autogenous, allogeneic, synthetic)
  - Lengthening of palate (Furlow z-plasty if not done already)
  - Pharyngeal flap
  - Pharyngoplasty
Craniofacial Differences
What is Craniosynostosis?

- Condition in which one or more sutures on a baby's head to close earlier than normal
- Results in head deformity that is permanent and can be severe
- Increased intracranial pressure, seizures, and developmental delay can occur
- Ideally repaired surgically at 3-6 months of age
Craniosynostosis

• Single suture
  • Sagittal synostosis → Scaphocephaly 40%
  • Unicoronal synostosis → Anterior plagiocephaly 15%
  • Metopic synostosis → Trigonocephaly 7%
  • Lambdoid synostosis → Posterior plagiocephaly 1%-2%
Craniosynostosis

- Multiple sutures (1 in 25,000 - 1 in 100,000)
  - Bicoronal → Anterior brachycephaly
  - Bilambdoid → Posterior brachycephaly
  - Sagittal + metopic → Scaphocephaly
  - Bicoronal + sagittal + metopic → Turribrachycephaly
  - Multisuture → Kleeblattschädel
Positional Plagiocephaly

- Can be prevented if identified early (2-3 mo), with aggressive positioning
  - Look for torticollis and treat aggressively; PT if necessary
- Helmet therapy ideally starts at 5mo
- Active helmet therapy - 3 mo duration
  - vs. passive helmet therapy - 6-9 mo duration
Other Congenital Differences...

- Tessier clefts
- Microstomia/macrostomia
- Pierre Robin Sequence
- Microtia
- Prominent ears
- (Giant) congenital nevi
- Sebaceous nevus of jadahsson
- Hemangioma
- Port-wine stain
- Breast asymmetry
- Poland Syndrome
- Ectopic breast tissue
- Virginal breast hypertrophy
- Gynecomastia
- Tubular breasts
Pop Quiz

1. Isolated cleft palate is more often associated with syndromic conditions than combined cleft lip and palate
   • True
2. Only a cleft lip can be diagnosed prenatally, not a cleft palate
   • False
3. Cleft lip results from failure of fusion of the medial nasal and maxillary processes
   • True
"We restore, repair, and make whole those parts.... which nature has given but which fortune has taken away, not so much that they may delight the eye but that they may buoy up the spirit and help the mind of the afflicted."

-Gaspar Tagliacozzi 1597