OUTLINE

- Epidemiology
- Anatomy & Embryology
- Classification of Cleft Types
- Multidiscipline team management
- Associated Pathology
- Surgical and non-surgical treatments/ timing of therapies

LEARNING OBJECTIVES

At the end of this presentation, the participant will be able to:

- 1. Describe the etiology, basic embryology, and anatomy of cleft lip & palate
- 2. Understand the treatment strategies for caring for cleft patients
- 3. Improve his/her understanding of the multidisciplinary care required to care for cleft patients, from birth to adolescence

EPIDEMIOLOGY

Incidence:
- Cleft lip & palate (CLP) occurs in approximately
  1/680—750 births in U.S.
- Isolated CP (CPO) ½ frequency compared to CLP
- North America
  - CLP 0.2—2.3 cases per 1,000
  - CPO 0.1—1.1 cases per 1,000

Gender Prevalence:
- CLP: Male : Female ratio 1.5—2 : 1
- CPO: Male : Female ratio 1 : 2
**EPIDEMIOLOGY**

Ethnicity breakdown for CL/P
- Native American: 3.6/1,000
- Asian: 2.1/1,000
- Caucasian: 1/1,000
- African American: 0.4/1,000
- Hispanic: 0.75/1,000

CPA has fairly stable incidence across ethnic subgroups.

**ETIOLOGY**

Combination of Genetic + Environmental factors

Genetic Factors:
- TGFB3 (transforming growth factor beta 3)
- MSX1 (msh homeobox gene) - increased incidence when combined with tobacco/ETOH
- IRF6 (Interferon Regulatory Factor 6) - associated with Van der Woude Syndrome; contributes to cleft in up to 12% cases

Syndromic Association:
- 50% CPA
- 30% CL/P

**PIERRE ROBIN SEQUENCE**

Triad:
- Micrognathia
- Glossoptosis
- Cleft palate (classically U-shaped)

Initial defect is hypoplasia of mandible
- May be 2/2 to oligohydramnios
1.4% of cases associated with congenital heart defects
Limb, ocular, rib, and sternal anomalies/abnormalities also described.


**STICKLER SYNDROME**

Hereditary arthro-ophtalmopathy
Associated with Pierre Robin Sequence

Ocular anomalies – high myopia presenting before age 10, retinal detachment, cataract, vitreous degeneration, astigmatism, strabismus, glaucoma
Midfacial hypoplasia with short maxilla and depressed nasal bridge and elongated philtrum
Hearing loss (both sensorineural and conductive)

**OTHER SYNDROMES ASSOCIATED WITH CLEFT**

Goldenhar (Hemifacial microsomia, oculoauriculo-vertebral dysplasia spectrum, facioauriculo-vertebral sequence)
Apert (Acrocephalosyndactyly Type I)
Shprintzen
Treacher Collins
Marfan
Orofaciodigital syndrome type I (Papillon-League-Psaume) & Type II
Otopalatalodigital syndrome
Smith-Lemli-Opitz

**22Q11 SYNDROMES**

Microdeletions or additions
Incidence 1:4,000 births
DiGeorge, velocardiofacial, conotruncal anomaly face syndrome

Other manifestations:
- Cardiac – conotruncal defects
- Defective Cell-Mediated Immune Response
- Hypertelorism, downward-slanting palpebral fissures
- Low set ears
- Choanal atresia

**FAMILY HISTORY**

Likelihood of clefting in a child given affected family members

<table>
<thead>
<tr>
<th>Family History</th>
<th>Cleft Lip &amp; Cleft Palate</th>
</tr>
</thead>
<tbody>
<tr>
<td>No family history</td>
<td>0.1%</td>
</tr>
<tr>
<td>Unaffected parents with one previously affected child</td>
<td>4%</td>
</tr>
<tr>
<td>Two previously affected children</td>
<td>9%</td>
</tr>
<tr>
<td>One affected parent</td>
<td>4%</td>
</tr>
<tr>
<td>One affected parent and one previously affected child</td>
<td>17%</td>
</tr>
</tbody>
</table>

**PRENATAL DIAGNOSIS & COUNSELING**

50% of children with cleft lip & palate have other abnormalities as well
• 22% CLP patients have abnormal karyotype

Prenatal Ultrasound:
• 24 weeks gestation: identify cleft lip 30-55% of time (some variability due to US protocols)
• Ability to identify CPO on US lower – closer to 1-2%, although imaging modalities improving
Prenatal Diagnosis & Counseling

Goals of prenatal detection:
- Obtain genetic counseling
- Provide emotional support
- Treatment planning

Important to discuss recurrence risk with subsequent pregnancies

Parental education regarding feeding, comorbidities, and counsel regarding expectations/timing of procedures
- Pre-order special feeding supplies

Anatomy

Cleft Anatomy

Cleft Lip
- Unilateral:
  - Orbicularis oris muscle abnormally inserts onto anterior nasal spine on the non-cleft side, and onto the nasal alar base on the cleft side

- Bilateral:
  - Premaxilla positioned more anteriorly
  - Prolabium does not contain orbicularis muscle

Cleft Palate
- Deficiency of Primary/secondary palate + musculature
- Abnormal insertion of Levator veli palatini muscle
LEVATOR VELI PALATINI MUSCLE

In cleft palate, the paired LVP muscles run from the posterior palate in an anterior-posterior direction.

3 abnormal insertion points:
1. Posterior edge of hard palate
2. Tensor veli palatini aponeurosis
3. Superior pharyngeal constrictor muscle

EMBRYOLOGY

Development of nose/upper lip begins during 4th week of gestation
Several pathways involved:
- Sonic hedgehog (SHH), wingless type (Wnt), bone morphogenic protein (BMP), fibroblast growth factor (FGF)

Fusion of frontonasal & maxillary processes begins at 32 days

Upper lip development typically complete by 48th day gestation

EMBRYOLOGY

8th week: tongue withdraws from position between lateral maxillary prominences to shift from vertical to horizontal position and fuse
- Several mechanisms believed to contribute to migration
  - Increased mesenchymal proliferation + increased tissue fluid content
  - Signaling pathways: platelet-derived growth factor, FGF10, SHH, and TGFbeta-3

- University of Indiana – Development of Face and Palate
  - Judith A. Stoffer, MA; Commissioned by Valerie Dean O’Loughlin, Ph.D, Professor of Anatomy
  - Indiana University Bloomington
  - http://www.indiana.edu/~anat550/hnanim/face/face.html
EMBRYOLOGY & ANATOMY

CLEFT CLASSIFICATION

CLEFT PALATE
- Unilateral vs. Bilateral
- Primary vs. Secondary
- Complete vs. Incomplete
- Submucous Cleft

CLEFT LIP
- Unilateral vs. Bilateral
- Complete vs. Incomplete
- Microform Cleft

UNILATERAL INCOMPLETE CLEFT LIP

UNILATERAL COMPLETE CLEFT LIP & PALATE
ISOLATED CLEFT OF SECONDARY PALATE

SUBMUCOUS CLEFT PALATE

Triad
- Bifid uvula
- Zona pellucida
- Notched hard palate or absent palatal spine

CLEFT NASAL DEFORMITY

Deviation of nasal bones toward cleft side
- Shortened columella
- Foreshortened medial crus of cleft side lower lateral cartilage
- Displacement of the caudal septum to Non-cleft side
- Lateral, inferior, and posterior displacement of cleft side alar base

MULTIDISCIPLINARY CARE OF CLEFT-RELATED ISSUES

CLEFT MANAGEMENT

Multidisciplinary Approach
- Medical management
- Surgical management
- Adjunctive treatments/therapies

MULTIDISCIPLINARY CLEFT TEAM

Goals:
- Convene regularly to evaluate & discuss patient/family needs
- Coordinate multi-specialty care
- Consolidate case loads to maintain expertise
- Facilitate continuing education for all team members
- Plan short & long-term patient management outcome measures
FEEDING THE CLEFT PATIENT
Wide complete clefts prevent seal on bottle or breast
• Inefficient sucking, tire more readily
• Swallow significant air
• Breast feeding feasible in CL only
Early SLP involvement
Special nipples and valve flow control system
• Meade-Johnson Cleft Palate Feeder
• Pigeon Cleft Palate Nipple and Bottle
• Special Needs Feeder (Haberman Feeder)

FEEDING DIFFICULTY
Modified Feeding Techniques
• Upright positioning
• More frequent/aggressive burping
• Placement of nipple in buccal area of opposite cheek
Devices
• Palatal feeding prosthesis
• Syringe w rubber tubing
• Special Nipples/valve flow systems

OTITIS MEDIA WITH EFFUSION — CLEFT PALATE
Conductive hearing loss
• Speech and language development
Eustachian Tube Dysfunction
• Eustachian Tube (ET) connects middle ear to nasopharynx
• Depends on proper function of both TVP and LVP muscles
• Children have shorter ET oriented in a horizontal plane with smaller opening

EAR DISEASE/EUSTACHIAN TUBE DYSFUNCTION
Cleft children:
• Shorter ET compared to non-cleft children, and larger angle between cartilage & TVP
• Deformed ET cartilage with less elastin
• TVP and LVP muscles have less contractile tissue & more connective tissue
Results in more negative middle ear pressure which leads to fluid accumulation = middle ear effusion

EAR DISEASE/EUSTACHIAN TUBE DYSFUNCTION
All cleft palate patients have ETD
• Improves with palatoplasty, but functional obstruction still exists
70% of children with cleft palate obtain normal ET function 6-10 years after repair
Cleft patients are more likely to develop cholesteatoma due to retraction/ETD
• Prevalence of cholesteatoma in children with cleft palate 9.2% (compared to 6/100,000)

SPEECH & LANGUAGE DEVELOPMENT
Considerations:
• Hearing Loss
• Velopharyngeal dysfunction
• Dimplism
Early Speech Therapy involvement
• Frequent monitoring/re-evaluation
**VELOPHARYNGEAL COMPETENCE**

During phonation, velopharyngeal closure contributes to oronasal resonance. Complete velopharyngeal closure requires coordination of soft palate (velum) movement with lateral and posterior pharyngeal walls. Velopharyngeal closure necessary for production of:

- Consonant sounds
- Nasal resonance of all sounds except nasal sounds /m/, /n/, /ŋ/

**VELOPHARYNGEAL INSUFFICIENCY**

Evaluate pharyngeal closure pattern and size of gap (nasal endoscopy):

- Consonant movement of velum primarily responsible for velopharyngeal closure, little contribution from lateral pharyngeal walls
- Sagittal movement of the lateral pharyngeal walls primarily responsible for closure, little posterior movement of velum
- Circular: movement of velum and pharyngeal walls contribute to sphincteric or purse-string closure

Management:

- Speech therapy
- Prosthetic devices (Obturator)
- Augmentation of pharyngeal wall
- Surgical procedures

**ANATOMY OF VELOPHARYNGEAL COMPETENCE**

Muscles of palatal sling:

- LVP, TVP, palatoglossus, palatopharyngeus, musculus uvulae

Lateral and posterior pharyngeal movement:

- Salpingopharyngeus and superior pharyngeal constrictors

Muscles innervated by pharyngeal plexus (CN IX and X) except for TVP (CN V)

**HOARSENESS**

Some data suggests higher prevalence of hoarseness/vocal cord pathology in cleft palate patients. Laryngeal compensation for velopharyngeal dysfunction

VPI compensatory mechanisms:

- Attempt to overcome insufficient air pressure in oral cavity
- Mechanisms include mid-dorsum palatal stops, posterior nasal fricatives, velar fricatives, pharyngeal stops, pharyngeal fricatives, and glottal stops.
- Glottal stops implicated in VC abnormalities and voice disturbances/nasal strait

**AIRWAY ISSUES IN CLEFT PALATE**

Higher risk for upper airway obstruction, particularly in syndromal patients

Nasal obstruction due to deviation

- PES: Micro/Retognathia, glossoptosis
- Hypopharyngeal obstruction due to collapse of epiglottis & tongue base
- Protrude positioning, nasal or oral airway

Lower threshold for airway endoscopy in syndromal children

Children will attempt to compensate to minimize nasal air escape

- Facial grimacing, hoarseness, low speech volume, compensatory misarticulations

SLP – document resonance, nasal air emission, consonant production errors, speech intelligibility, speech acceptability.
Normal pediatric OSA prevalence 2-3%
• 3-12% with primary snoring
Cleft palate children more likely to have Sleep Disordered Breathing
• SDB prevalence 22-65%
• OSA 8.5% (almost 3 x as likely as general peds pop)
Low threshold for Polysomnography (PSG) testing

Children may experience increase in airway obstruction at time of palatoplasty
• Decrease in functional airway space caused by closure of palate
• Tongue may be more swollen due to retractor placement
• Placement of nasal airway prior to extubation after palatoplasty may help
Some patients have worsening of OSA symptoms after cleft palate repair or after pharyngeal flap/VPI surgery
Role for mandibular distraction osteogenesis or tongue-tip adhesion in PRS.
• Also consider trach and g-tube, if necessary

Timing: 0-4 months
Helps to narrow cleft distance in preparation for surgical repair
• Reduce tension for repair
May also help reposeion pro-labium
Can be used alone or with Pre-surgical Orthopedics (PSO)

Timing: starting at 2-4 weeks
Naso-Alveolar Molding (NAM)
Latham Device
Nasal Splints
LIP ADHESION

Timing: 2-4 weeks
Goal is to convert complete cleft lip to incomplete cleft
- Allows definitive surgical repair to be simpler and under less tension
- Can delay definitive lip repair to allow for solidification of maxillary platform
- May be used with or without use of orthopedic devices

POSTOPERATIVE CARE

Normal bottle or nipple feeding resumed on POD #1
Adhesive strips left on wounds for ~3 days
Soft arm restraints as appropriate
Wounds cleaned with dilute H2O2 and antibiotic ointment
Postop massage around 3 weeks for scar contracture

CLEFT LIP REPAIR

Timing: 2-3 months
Goals of Repair
- Reconstruct orbicularis oris muscle
- Camouflage scar
- Begin to restore nasal symmetry
Key Steps
- Closure of nasal floor
- Approximate orbicularis oris muscle
- Closure of lip and re-creation of philtrum

TYMPANOSTOMY TUBE PLACEMENT

Timing: 6 -12 months
In conjunction with palatoplasty
Indications:
- Conductive Hearing Loss (CHL)
- Chronic Otitis Media with Effusion (COME)
- Eustachian Tube Dysfunction (ETD)
- Need for repeat sets of tubes
- May require T-tube placement
- Long term sequelae (cholesteatoma)

CLEFT PALATE REPAIR

Timing: 9-14 months
Goals of Repair
- Separation of nasal & oral cavities
- Creation of competent velopharyngeal valve for swallowing & speech
- Preservation of midface growth
- Development of functional occlusion
**PALATOPLASTY**

**Objectives**
- Layered closure with minimal tension
- Reconstruction of the levator sling

**Palatoplasty techniques**
- Primary veloplasty (Schweckendiek)
- Bipedicled flap palatoplasty (van Lengerbeck)
- V-Y pushback palatoplasty
- Two-flap palatoplasty
- Furlow (double-opposing Z-plasty)

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**VPI SURGERY**

**Timing**: 3-5 years

**Pharyngeal flap**
- Large central gap with sagittal closure pattern and adequate lateral wall movement
- Goal: to create central obstruction to allow lateral pharyngeal walls close against flap, and allow residual nasal airflow to pass through lateral ports.

**Furlow double-opposing Z-plasty palatoplasty**
- Small central gap with notched soft palate (indicates malposition of levator veli palatini)

**Sphincter pharyngoplasty**
- Coronal closure pattern

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**PHARYNGEAL FLAP**

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**SPHINCTER PHARYNGOPLASTY**

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**CLEFTH RHINOPLASTY**
CLEFT RHINOPLASTY

TIMING OF THERAPEUTIC PROCEDURES

- Lip Adhesion: 2-4 weeks
- PSO/NAM: 2-4 weeks
- Cleft Lip Repair: 2-3 months
- PE Tube placement: 6-12 months
- Cleft Palate Repair: 10-14 months
- Correction of VPI: 3-5 years
- Intermediate rhinoplasty: 3-5 years
- Orthodontia: 6-12 years
- Alveolar bone grafting: 8-12 years
- Definitive Septorhinoplasty: post-adolescence

REFERENCES

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- Johnson JT, Rosen CA, Bailey BJ. Bailey's Head & Neck Surgery – Otolaryngology. 5th ed. 2014. Ch. 103 and 107 (Comprehensive Cleft Care, The Syndromal Child)

Photo credits:

- Brackmann DE et al. Otologic surgery. 3rd ed. 2010. Chapter 6 (Surgery of Ventilation and Mucosal Disease)