Pierre Robin Sequence: Respiratory Considerations & Feeding Development

Carrie Drake-Luecking, MA, CCC-SLP, CLC, CNT
Kaitlyn Schmitt, MS, CCC-SLP

Riley Hospital for Children
Indiana University Health

Disclosures

- Cara Drake-Luecking
  - I am employed by Indiana University Health and a member of the Craniofacial Team at Riley Hospital for Children
  - I do not have any non-financial disclosures to report
- Kaitlyn Schmitt
  - I am employed by Indiana University Health and a member of the Craniofacial Team at Riley Hospital for Children
  - I do not have any non-financial disclosures to report

Objectives

1. Describe diagnostic criteria for Pierre Robin sequence and how these criteria may impact an infant's feeding abilities
2. Describe therapeutic feeding strategies to improve suck-swallow-breathe coordination of infants with Pierre Robin Sequence.
3. Describe how early feeding difficulties for children with Pierre Robin Sequence may impact oral motor development for functional feeding

Pierre Robin Sequence: An Overview

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Syndromes, Associations, and Sequences (Oh my!)

- Syndrome
  - well-characterized constellation of major and minor anomalies that occur together in a predictable fashion presumably due to a single underlying etiology
  - Examples: Trisomy 21 (Down Syndrome), Stickler Syndrome, CHARGE Syndrome
- Association
  - group of anomalies that occur more frequently together than would be expected by chance alone but that do not have a predictable pattern of recognition and/or a suspected unified underlying etiology
  - Example: VACTERL
- Sequence
  - A pattern of deformations and malformations which is a consequence of a single malformation
  - Examples: **Pierre Robin Sequence**, Potter Sequence

(Lowry et al., 2011)

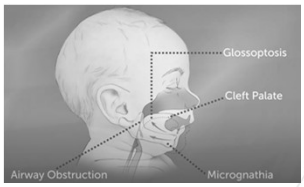
Pierre Robin Sequence (PRS)

- Occurrence in 1/8500 to 1/14,000 births
- Equally common in males & females
- High incidence in twins

(Gangopadhyay et al., 2012)
(Johns Hopkins, 2021)

Characteristics of PRS

- Mandatory characteristics
  - Micrognathia
  - Glossoptosis
  - Airway obstruction
- Common association
  - Cleft palate (66-90% of patients; Flores, 2014)



(Screenshot from: Boston Children's Hospital on YouTube)

Glossoptosis

- Displacement of the tongue base into the oropharynx and hypopharynx
- Severity is variable
- Severe glossoptosis
  - Inspiratory stridor
  - Increased respiratory effort
  - Apnea
  - Cyanotic episodes
- Functional impact may worsen during sleep or relaxation
- Endoscopy allows visualization of tongue position

Pierre Robin Sequence (PRS)

- No known singular cause
- Approximately 40-60% of cases occur as part of a syndrome or association
  - Stickler syndrome (20-25% of syndromic PRS)
  - Velocardiofacial syndrome
  - Treacher-Collins syndrome
  - Campomelic dysplasia
  - Trisomy 11q syndrome
  - Deletion 4q syndrome
  - CHARGE association

(Gangopadhyay et al., 2012)
(National Institutes of Health, 2021)

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Airway Obstruction

- Clinical signs assessed during sleep, wakefulness, & with feeding
- Sleep-disordered breathing
  - Snoring / obstructive noises
  - Increased respiratory effort
  - Diaphoresis
  - Obstructive hypoventilation
  - Obstructive sleep apnea
  - Restlessness
- Intermittent presentation may delay referral & diagnosis

Micrognathia

- Likely the initiating event
- Deficiencies in mandibular body & ramal lengths
  - CT
  - lateral cephalograms
  - plaster casts
  - 3-dimensional photography
  - Ruler & caliper
  - May also have upper jaw hypoplasia
- Evaluation is sometimes subjective; functional impact must be measured

Sleep Studies

- PSG 4-6 hours with at least 3 hours of sleep
- Apnea-Hypopnea Index (AHI)
  - # of mixed, obstructive, and central apneas and hypopneas per hour of sleep
- Currently there is no set diagnostic criteria for intervention
  - What is the functional impact on the infant/family?

PRS Secondary Comorbidities

- Dysphagia
- Failure to thrive
- Developmental delay
- GER
- CO2 retention
- Heart failure
- Brain damage
- Sudden death

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Airway Management Strategies

- Positioning modifications
  - Prone or side lying
- Supplemental oxygen
- Nasopharyngeal tubes
- Continuous positive airway pressure (CPAP)
- Intubation
- Tongue-lip adhesion (TLA)
- Mandibular distraction osteogenesis (MDO)
- Tracheostomy

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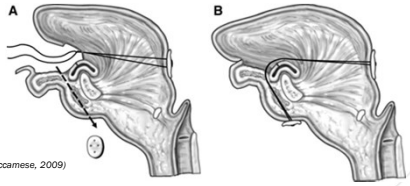
Airway Management (Li et al., 2017)

- 63 patients (2004 – 2013)
- Primary management
  - 76% managed by prone positioning
  - 11% floor of mouth release
  - 6% MDO
  - 6% tongue-lip adhesion
- Secondary management
  - 5 needed a secondary surgery (MDO or tracheostomy)
- Patients with associated airway pathologies required surgical intervention (i.e., bronchomalacia, tracheomalacia, TEF)
- Syndromic PRS resulted in poorer outcomes

Tongue-Lip Adhesion (TLA)

- Popularized in the 1940s as an alternative to tracheostomy
- High success rate
- Preferred first-line form of surgical intervention in many centers



(Quqish & Caccamese, 2009)

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Tongue-Lip Adhesion (TLA)

(Gangopadhyay et al., 2012)

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Risks of TLA

- Surgical risks
- Dehiscence of adhesion
- Scar contracture
- Poor efficacy of treatment
  - Dependent on mandibular "catch up growth"
  - May still require tracheostomy (Flores et al, 2014)
- Feeding dysfunction
  - Some controversy on the impact of feeding with negative outcomes affecting tongue mobility & swallowing mechanisms but others reporting improved weight gain after the procedure (Cote et al., 2015)

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Mandibular Distraction Osteogenesis (MDO)

- 10mm or more of overjet
- Alleviates airway obstruction early
- Higher oxygen saturation levels and lower AHI scores at 1-month and 1-year postoperative (Flores et al., 2014)
- Moves the tongue forward
- General advancement of the mandible 15-20mm based on initial deficiency
- Noticeable airway improvement within 1-2 weeks
- Distractor remains in place for ~ 8 weeks to allow for consolidation

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Mandibular Distraction Osteogenesis (MDO)

The Royal Children's Hospital Melbourne, 2018

Resnick, 2017

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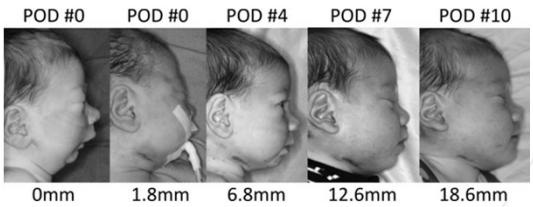
Mandibular Distraction Osteogenesis (MDO)



(Resnick, 2017)

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Risks of MDO

- Surgical risks
- High cost of care
- Device failure
- Loss of inferior alveolar sensory function
- Facial nerve injury
- Mandibular malunion
- Stiffening of temporomandibular joint
- Damage to tooth buds
- Mandibular growth disturbance

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Cleft Palate

- Wide, U-shaped cleft soft palate
  - May impact hard palate to some degree
- Cleft palate is an opening in the roof of the mouth in which the two sides of the palate did not fuse
  - Disrupts the Levator Palatini muscle
- Embryonic palate formation is disrupted during the 8th week of gestation
- Large tongue volume may act as a physical barrier to palate formation
- Most common associated abnormality in PRS

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Feeding the Infant with Pierre Robin Sequence

- Hong et al., 2012
  - At baseline, patients demonstrated laryngeal penetration, abnormal tongue movements, increased pharyngeal transit times, and frank aspiration (5/6 patients)
  - Following MDO, there were no further instances of aspiration and pharyngeal transit times normalized
- Rathe et al., 2015
  - Feeding difficulties were present in 95.8% of all patients
  - On average, exclusive oral feeding was achieved at 5 weeks of life for infants with isolated PRS and 14 weeks for patients with syndromic PRS
  - 7 patients were not able to exclusively oral feedings at 1 year of age
  - 10 patients never required supplemental tube feeding
- Glynn et al., 2011
  - 70% of patients required supplemental tube feedings

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Feeding the Infant with Pierre Robin Sequence

- Morice et al., 2018
  - Severe respiratory disorders and long-lasting feeding dysfunction was more common for syndromic PRS patients compared to those with isolated PRS
  - Grades of retrognathia and glossoptosis did not differ significantly between the syndromic and non-syndromic groups
  - Neurologic impairment was associated with long-term feeding dysfunction
  - Authors concluded severity of clinical conditions is more closely correlated with syndromic status, laryngeal abnormalities, and neurologic impairment than degree of micrognathia or glossoptosis

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G-tube Rates & Pierre Robin Sequence

- Lidsky et al., 2009
  - 0% of infants with isolated PRS who received early airway intervention required GT
  - 12.8% of infants with isolated PRS who received late or no airway intervention required GT
  - 87.5% of infants with syndromic PRS who received early airway intervention required GT
  - 62.5% of infants with syndromic PRS who received late or no airway intervention required GT
- Ghoul, 2018
  - GT placement in MDO, TLA, Trach
    - Trach highest (78.6%), then TLA (63.4%), then MDO (37.8%)
  - If baby had an airway operation, 60.5% had a GT
  - In non-operative group, 21.3% had a GT

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An Increase in Reflux with Pierre Robin Sequence

- Multiple studies have documented an increase in gastroesophageal reflux in infants with PRS
  - Monasterio et al. (2004) found GER in 83% of infants with PRS and apneic episodes; all experienced improvement in GER symptoms after undergoing MDO
  - Hong et al. (2012) reported GER in 100% of the infants in their study; again, all experienced improvement in symptoms following MDO
  - Baudon et al. (2002) reported asynchronous sphincter relaxation, abnormal esophagela waves, and increased resting pressures of the upper and lower sphincters
- Currently, it is believed that an increase in GER is secondary to severe upper airway obstruction
  - Increased respiratory effort leads to negative intragastric pressure
  - Immaturity of esophageal sphincter

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Feeding & Pierre Robin Sequence

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The Building Blocks of Successful Oral Feedings

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General Infant Feeding

- For ALL infants:
  - Physiologic stability is an essential underpinning to oral feeding
  - Compromise to cardiac, gastrointestinal, neurologic, or respiratory symptoms may preclude safe oral feeding
  - Feeding disrupts respiration
  - Coordinated suck-swallow-breathe pattern supports infant engagement in feeding
  - Adequate growth supports neurologic development and healing
  - Sucking reflex integrates at around 2-4 months of age
  - Early feeding times are a valuable bonding opportunity for parent & baby
  - Behaviors are the result of repetitive experiences

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Feeding the Infant with Pierre Robin Sequence

- Increased risk of airway compromise
  - Tongue-based airway obstruction
  - Apnea, tachypnea, desaturations
  - Penetration & aspiration
- **Inefficient** feedings
  - Cleft palate
  - Defensive or passive feeding behaviors
- **Insufficient** feedings
  - Fatigue/lethargy
  - Increased caloric needs
  - Refusal of feedings

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Feeding the Infant with Pierre Robin Sequence

- ACPA accredited Multi-Disciplinary Team
- Breugem et al., 2016
  - "Given the association with failure to thrive, swallowing dysfunction and developmental delays, the recognition and treatment of feeding problems in [infants with] PRS are priorities."
  - Clinical feeding assessment
    - Completed by an experienced provider
    - Initiated in the first days-weeks of life
    - Assessment includes:
      - **Efficiency of feeding**
      - **Quality of feeding**
      - **Effect on respiratory status**
      - **Quality of weight gain & growth**

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Clinical Feeding Assessments & Pierre Robin Sequence

- Respiratory assessment
  - At rest, with non-nutritive stimulation, with oral feeding attempt
  - Pulse oximetry in clinical settings
  - Watch & Listen
  - Parent report
- Manage airway first
  - Positioning considerations
  - Supplemental oxygen considerations
  - Is airway intervention necessary?
- Only attempt NNS and PO feeding with a stable airway

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Clinical Feeding Assessments & Pierre Robin Sequence

- Begin with an assessment of stability during non-nutritive stimulation
  - Respiratory rate, heart rate, oxygen saturation as able
  - Evaluate rooting & latching
  - Suction & compression
- Coordination of Suck-Swallow-Breathe pattern during nutritive sucking
  - Rate
  - Strength
  - Coordination
  - Stability
- Finish with a reassessment of stability at rest or with non-nutritive sucking
- Monitor for distress cues & adjust plan as needed

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Bottle Selection & Pierre Robin Sequence

- Optimal feeding systems
  - Give baby control of expression rate & bolus size
  - Are efficient (<20-30 minutes)
  - Consistent across feedings & caregivers
  - Promote an organized feeding pattern
  - Allow for safe swallow strategies
- Special consideration for infants with clefts
  - Utilize specialty feeding systems to maximize compression
  - Consider positioning
- Positioning
  - Elevated Side Lying
  - Upright

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Cleft Specialty Bottles

- Assisted delivery bottles
  - Caregiver controls the feeding by squeezing the bottle
  - Infant can be a passive participant
  - Enfamil Cleft Palate Nurser, Medela Special Needs Feeder (Haberman)
- Infant directed bottles
  - Baby controls the compression depth & sucking frequency
  - Most consistent across feedings & feeders
  - Nipple flow rate can often be adjusted
  - Medela Special Needs Feeder (Haberman), Pigeon, Dr. Brown's Specialty Feeding System

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Cleft Specialty Bottles




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Aspiration

- Poor growth
- Poor feeding cues
- Refusal of feedings / "shutting down"
- Persistent distress cues
- Prolonged feeding times
- Persistent dream feeding
- Wet/gurgly vocal quality
- Cough/sputter
- Stridor or wheezing
- Increased work of breathing
- Desaturations/apnea/bradycardia

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Oral Motor Development

- PRS Contributing factors
  - Tongue position
  - Airway obstruction
  - Orophayrngeal dysfunction
  - Insufficient suction
  - Pharyngonasal reflux
  - Cleft palate
- Oral motor skill development
  - Birth – 4 months: Suckle; anterior to posterior motion
  - 4-6 months: sucking; up-and-down motion
  - 6-9 months: emerging Munch; tongue lateralization
  - 9-12 months: munch; emerging rotary
  - 15-24 month: rotary chew, jaw stabilization for cup drinking

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Persisting Feeding Difficulties (Reid et al., 2006)

- Who
  - Babies with cleft lip, cleft palate, cleft lip and palate.
    - Cleft condition in isolation, isolated PRS, and PRS + congenital defect or syndrome
- Feeding difficulties persisting beyond 14 months
  - All babies with diagnosed PRS

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Persisting Feeding Difficulties (Reid et al., 2006)

- 14 months
  - Good
    - Established cup feeding
    - Eating finger food/solid consistencies
  - Satisfactory
    - Primarily breast/bottle feeding, but starting cup drinking
    - Soft solid consistency
  - Poor
    - Primarily tube feeding while working towards solids
    - Or if full PO, primarily eating purees
    - No cup

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Persisting Feeding Difficulties

- Early Intervention
  - Developmental milestones
  - Feeding Milestones
    - Progressing oral motor skills with purees and solids
    - Cup drinking

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Case Study: Baby D

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Case Study – Baby D

- Born at 38+2 weeks gestation
- No complications with pregnancy
- Nuchal cord x1
- Apgars 6, 8
- Required CPAP w/100% O2 in delivery room to attain sats >90%
- Admitted to the NICU due to respiratory distress
- Initial neonatal notes state pt has increased tone and is "jittery at thimes"
- Incomplete hard & soft palate cleft (Veau II)
- Preauricular tags
- Feeding goals: exclusive breast feeding
  - Mother pumping

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Case Study – Baby D

- Initial feeding evaluation with SLP at DOL 2
- Weaned to room air & in crib on same date
- Oral mech exam confirms cleft palate
- PO feeding assessment
  - Pigeon nipple (? size)
  - Upright position
  - Coordinated SSB
  - Minimal anterior bolus loss
  - No nasal regurgitation and no desats
  - Unknown PO volume

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Case Study – Baby D

- DOL 5-21
  - Poorly advancing PO volumes (20-60ml)
  - Prolonged feeding times (30-60 minutes)
  - Poor growth (reached birth weight at DOL 20)
  - Inconsistent feeding cues
  - Desaturations occurring outside of feedings
  - Infrequent ST visits while in OSH NICU
    - Recommendation for Regular Pigeon nipple

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Case Study – Baby D

- Transferred to NICU at Riley Hospital for Children at IUH on DOL 21 for feeding evaluation & craniofacial consultation
  - Still taking 30% PO
  - Riley NICU concerned for micrognathia
- Consults: Plastics, ENT, Optho, Genetics, Speech, OT

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Case Study – Baby D

- ST assessment completed at RHC NICU on DOL 22

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Case Study – Baby D

- ST treatment completed at RHC NICU on DOL 23

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Case Study – Baby D

- VFSS Results (DOL 24)
- Trial 1: Thin liquid via Dr. Brown's Cleft bottle with DB-Newborn nipple
  - 4ml consumed, 29 swallows visualized
  - 4 penetrations, no aspirations
- Trial 2: Thin liquid via Dr. Brown's Cleft bottle with DB-Level 1
  - 4ml consumed, 14 swallows visualized
  - 4 penetrations, 1 aspiration off-camera
- Trial 3: Thin liquid via Dr. Brown's Cleft bottle with DB-Newborn nipple
  - 11ml consumed, 27 swallows visualized
  - 3 penetrations, no aspirations

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Case Study – Baby D

- VFSS Recommendations:
  - PO x15 minutes with Dr. Brown's Cleft bottle with DB-Newborn nipple
  - Side lying feeding position
  - Supplemental NG feeds as needed
  - Outpatient f/u through Craniofacial clinic

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Case Study – Baby D

- ST visit on DOL 25 (Day after VFSS)
- 1400 feeding: Offered PO with Dr. Brown's Cleft Bottle with DB-Newborn nipple in elevated side lying
  - Slow latch, Improved SSB coordination with adequate self-pacing, minimal catch-up breathing, no tracheal tugging, Intermittent tongue thrusting
  - PO x12 minutes prior to cough, color change, and desat to 63% requiring sternal rub by SLP & RN to recover, cessation of feeding cues after that time
  - SLP planned to return for the next feeding time on that date for reassessment
- 1700 feeding: Offered PO with Dr. Brown's Cleft Bottle with DB-Newborn nipple in elevated side lying
  - Good feeding cues, timely latch, timely initiation of a nutritive feeding pattern
  - Desat to 79%, self-recovered, feeding cues then muted but returned after several minutes. MOB reoffered bottle and pt continued to feed x10 minutes, drops in 02 to 92% but self-recovered
  - PO feedings with ST only and treatment frequency increased to 5x per week
  - D/w medical team consideration of GT placement

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Case Study – Baby D

- ST continued to follow 5x per week until the time of discharge
  - Respiratory status initially worsened after PO feedings were discontinued (head bobbing, tracheal tugging, and intermittent tachypnea), medical team initially considering distraction
  - Desaturations ceased after several days of NG feeds only and PO with ST only, distraction was avoided
  - During ST visits, baby consuming <10ml in 10-minutes feeding time
  - G-tube was placed on DOL 31
  - With improved respiratory status, LIMITED PO feedings were reinitiated
    - PO feedings 1-2x per day up to 10-minutes, no more than 10ml via Dr. Brown's Cleft with DB-Newborn
- Patient discharged home on primary GT feedings and limited PO practice plan on DOL 36

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Case Study – Baby D

- Baby presented for first Craniofacial Clinic Visit on DOL 42

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Case Study – Baby D

- Baby presented for second Craniofacial Clinic Visit on DOL 64

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Case Study – Baby D

- Outcomes to date at 5-months of age:
  - Primary nutrition & hydration via G-tube
  - Growing well
  - Holding head up independently
  - Cooing, smiling, engaging with parents
  - Using a feeding pump, does not tolerate bolus feedings well
  - Beginning limited tastes of baby food
  - Involved in First Steps
  - Continues to follow in Craniofacial clinic
  - Planning for palatoplasty around 12-months of age
  - Genetics results still pending

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Case Study: Patient C

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Case Study – Patient C

- Patient C is a female born full term with a PMH of Pierre Robin Sequence, cleft palate, micro/retrognathia s/p mandibular distraction, small for gestational age, patent foramen ovale, patent ductus arteriosus, multiple congenital anomalies, moderate to severe obstructive sleep apnea, laryngomalacia s/p supraglottoplasty, FTT and oropharyngeal dysphagia s/p gtube.
- Required a 1 month NICU stay due to respiratory failure secondary to micro/retrognathia cleft palate and feeding difficulties.
- Discharged from NICU and PO/NG feeding plan.
- Seen through the multi-disciplinary Craniofacial Clinic.
  - First visit full PO
  - Second visit family advanced to Dr. Brown SFS + level 2 nipple
    - Arching, finger splaying, increased congestion

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Case Study – Patient C

- Swallow Study 1 – 8 weeks old
  - Aspiration of thin, semi thick and nectar thick liquids from bottle systems with varying leveled nipples
  - Aspiration before the swallow - oor oral control resulted in overflow to the pharynx with all consistencies
  - Aspiration durirng – inadequate airway protection
  - Recommended NPO with NG tube feeds for all nutrition/hydration
- Swallow Study 2 – 4 months old
  - Aspiration thin, semi thick and nectar thick liquids from bottle systems with varying leveled nipples
  - Aspiration of puree
  - Highly disorganized oral motor pattern, inadequate oropharyngeal pressure due to open cleft palate, and inadequate airway protection.
- Recommended NPO and placement of gtube due to persisting feeding difficulties.

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Case Study – Patient C

- Outpatient evaluation
  - 9 months
  - NPO/tube feeds for all nutrition and hydration
  - Mom reported the patient was very interested in others while they eat.
- Oral Mech Exam
  - Unrepaired Veau II cleft palate
  - Poor management of oral secretions – frequent drooling; sucking on middle three fingers persistently
- Puree trials
  - 1 tbsp
  - Used suckle on 3 fingers to promote A/P transfer
  - Decreased labial seal and increased lingual pumping and significant anterior loss of bolus without fingers in mouth
  - Slight increase in congestion

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Case Study – Patient C

- Impressions
  - Pt presents with oropharyngeal dysphagia requiring gtube feeds for primary nutrition and hydration.
  - At a high risk for aspiration.
  - Given approval from MD, plan to trial minimal PO puree practice plan to target oral motor skill advancement to prepare for repeat VFSS.
  - Will need updated VFSS. Based on ongoing assessment and timing of palate repair surgery will determine timing of VFSS.
  - Ongoing parent education

Case Study – Patient C

- Recommendations
  - Continue gtube feeding plan per MD and RD recommendations for all nutrition and hydration.
  - 10 minutes before gtube feed sit patient in supportive highchair and may offer 1 tbsp stage 2 puree via infant spoon 1 x a day.
  - Please stop puree with overt signs and symptoms of aspiration and wait for reassessment at next therapy visit.

Case Study – Patient C

- How is she doing now?
  - 2 tbsp puree 2 x a day
    - Significantly improved oral motor skills with puree.
    - Rarely puts fingers in her mouth while eating puree.
    - Good weight gain, no respiratory symptoms, no overt s/s of aspiration with trials of puree.
  - Working on open cup drinking with puree and meltable solids in feeding therapy.
  - Scheduled for palate repair in May.
  - Goal to repeat VFSS after palate repair.

What questions do you have?

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