Clefts, Syndromes, and Care from Prenatal to Adulthood

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Age 11
Cleft lip and palate
'playing a game'

Kinetic Family Drawing: "Draw a picture of you and your family doing something."

Disclosures

- No financial conflict of interest to disclose
- Working as part of a cleft team allows me to feel like a 'total pediatrician'
  - Behavior and development
  - Dental
  - Dysmorphology and genetics
  - Language and speech
  - Nutrition
  - Pediatric conditions
  - Psychosocial

Overview

- Overview of cleft lip and cleft palate
- Syndromes associated with clefts
- Team cleft care

Goals I

Participants will:

- appreciate the complexity of children with cleft lips and palates
- appreciate that clefts can be isolated or part of a syndrome
- appreciate the added complexity associated with syndromic clefts

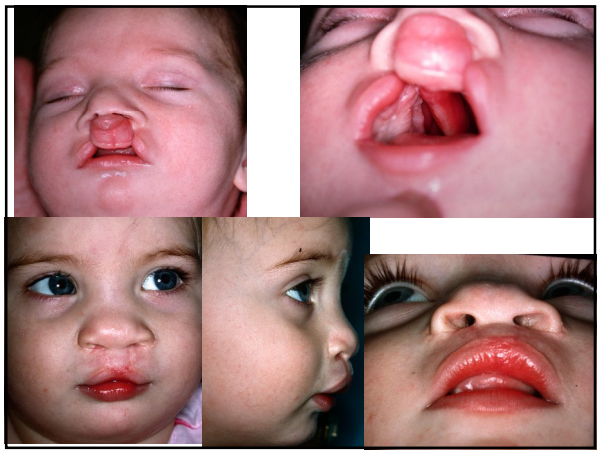
Goals II

Participants will also:

- understand the focus for care for children with clefts varies by age
- understand that hearing, speech, and swallowing issues are a few of many issues facing children with clefts
- understand that care for children with cleft lip and palate is best managed by a team approach

Overview of cleft lip and cleft palate

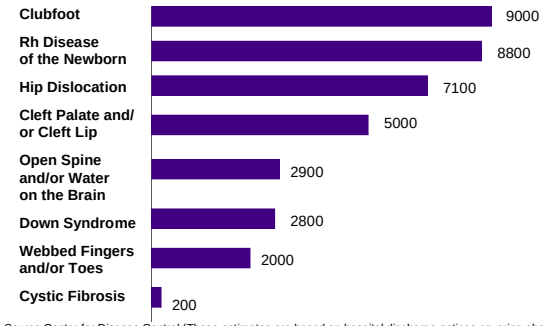
- What is a cleft?
- Range of clefts
- Isolated clefts vs.
  clefts as part of syndromes



Cleft Overview

- Fetal origins
  - Cleft lip 4-6 weeks gestation
  - Cleft palate 6-12 weeks gestation
- Over 300 different syndromes
- Teratogens (agents causing birth defects)
  - Alcohol
  - Dilantin
  - Phenobarbital
  - Retinoic acid
  - Thalidomide
  - Folic acid deficiency
- Most occur without known cause

ESTIMATED AVERAGE ANNUAL NUMBER OF LIVE BIRTHS WITH SPECIFIED BIRTH DEFECTS UNITED STATES 1970 - 1979



Source Center for Disease Control (These estimates are based on hospital discharge notices covering about 1/3 of the births in the United States. Only those structural defects evident at birth are shown.)

Incidence of clefting

- Cleft lip (with or without palate) occurs in 1/300 to 1/2500 births
  (depending on ethnicity)
- Cleft palate alone occurs in 1/1000 births
- Cleft lip more common in males
- Cleft palate more common in females
- Most children with clefts not born with other syndromes

Normal Lip - Cleft Lip

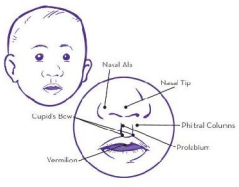


Figure 1:The Typical Lip and Nose

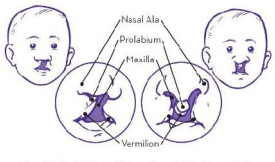


Figure 2A:The Unilateral Cleft Lip and Nose

Figure 2B:The Bilateral Cleft Lip and Nose

taken from http://www.cleftline.org/parents/cleft_lip

Normal Lip

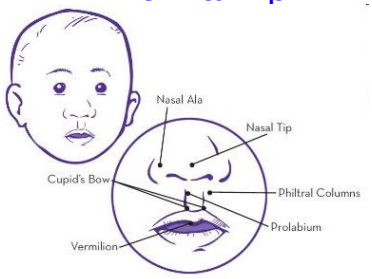


Figure 1:The Typical Lip and Nose

taken from http://www.cleftline.org/parents/cleft_lip

Cleft Lip

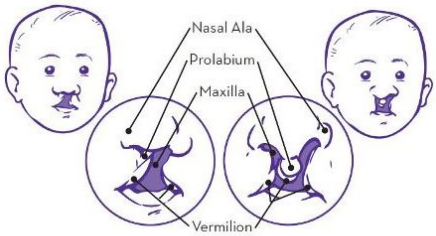
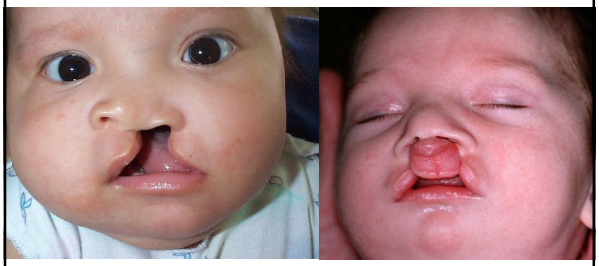


Figure 2A:The Unilateral Cleft Lip and Nose

Figure 2B:The Bilateral Cleft Lip and Nose

taken from http://www.cleftline.org/parents/cleft_lip



Normal Palate

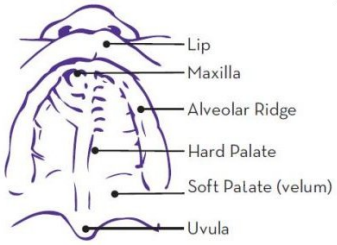


Figure 3:The Typical Palate

taken from http://www.cleftline.org/parents/cleft_palate

Cleft Palate

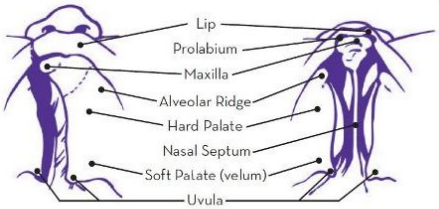
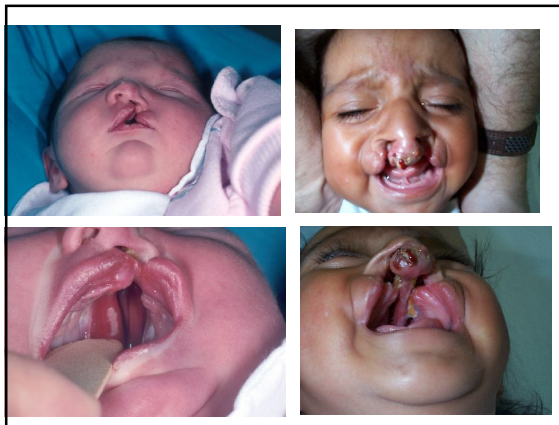


Figure 4A: Unilateral Cleft Palate

Figure 4B: Bilateral Cleft Palate

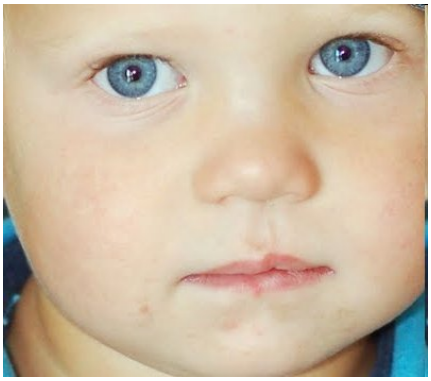
taken from http://www.cleftline.org/parents/cleft_palate

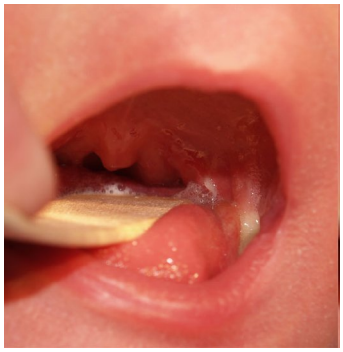


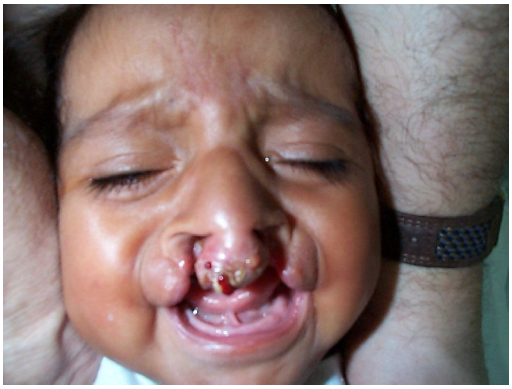
Range of clefts

CLEFT LIP
- Microform
- Unilateral incomplete
- Unilateral complete
- Bilateral incomplete
- Bilateral complete
- Extending through alveolus
- Extending up into the nose

CLEFT PALATE
- Bifid uvula
- Submucosal cleft
- Cleft of the soft palate
- Unilateral cleft palate
- Bilateral cleft palate
- Complete vs. incomplete
- Combinations







Clefts and syndromes

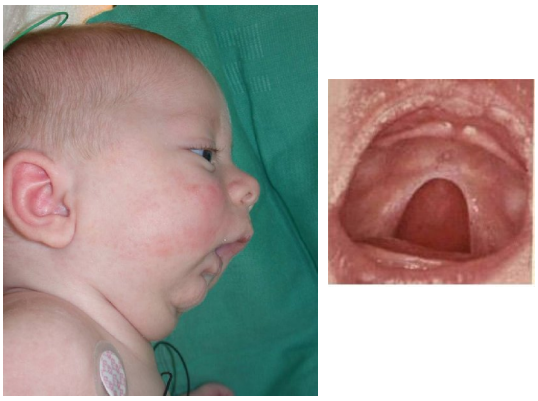
- Most clefts are isolated clefts, or clefts without any associated syndrome
- However, more than 300 syndromes are associated with clefts
- Syndromes often associated with developmental differences, but many have renal, endocrine, and skeletal abnormalities.

Examples of syndromes

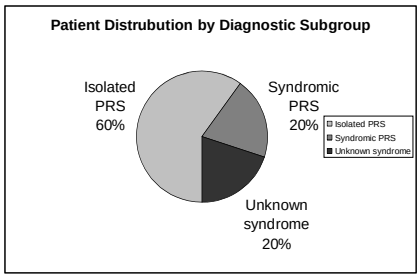
- Pierre Robin Sequence
  - Including Stickler syndrome
- DiGeorge syndrome
- Apert syndrome
- Crouzon syndrome
  - Craniofacial, but not cleft
- Treacher Collins syndrome

Pierre Robin Sequence

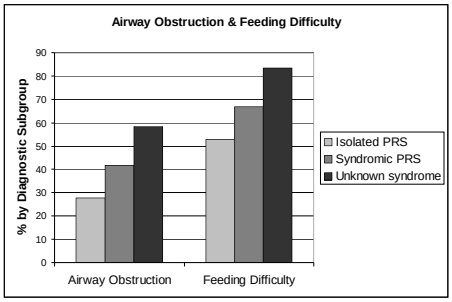
- Occurs in 1/8,500 to 1/30,000 births
- Small jaw (micrognathia)
- Tongue pushed back (glossoptosis)
- U-shaped cleft palate in many
- Airway issues may result in tracheostomy
- 60% syndromic
  - Stickler syndrome (34%)
  - Velocardiofacial syndrome (11%)
  - Fetal alcohol (10%)
  - Treacher Collins syndrome (5%)



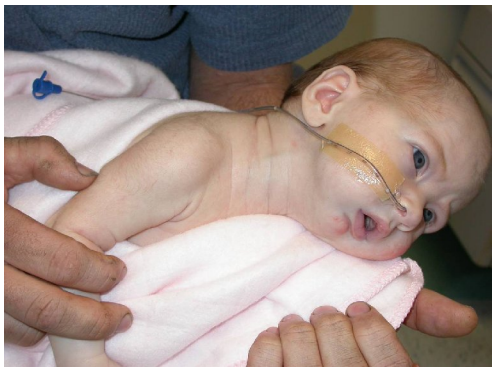
UC Davis
1993-2002 60 Patients



UC Davis experience
1993-2002 60 patients with PRS

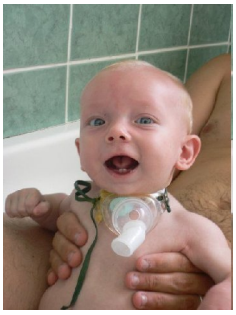


Required Intervention Required NG tube

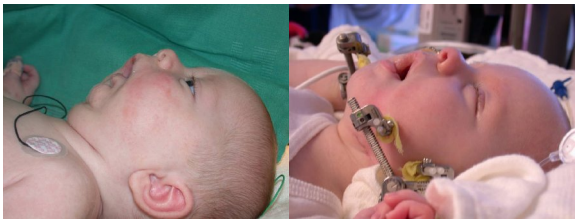


Tracheotomy

- Tracheostomy needed in 3-50% of PRS
- Tracheostomy can be removed in most, 3.1 year on average



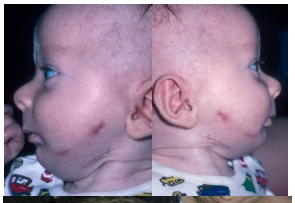
External distraction to move chin



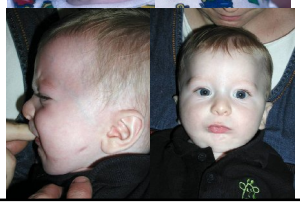
Post op day 13

Facial Scars

3 months



6 months



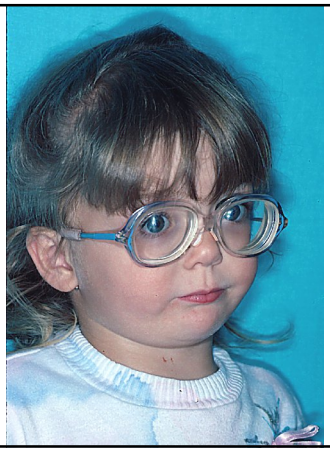
Long-Term PRS Effects

3 Years of Age

- Successful removal of tracheostomy
  85% of 17 patients (out of 60)
- Eating food by mouth normally (out of 60)

Isolated PRS	91%
Named syndrome	92%
Unknown syndrome	78%

Smith MC, Senders CW, Int J Ped Oto; 2005

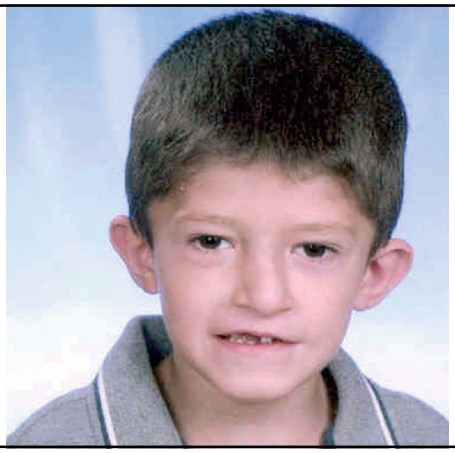


Stickler syndrome

- 30-40% of children with PRS have Stickler
- Type 2 collagen disorder
  COL2A1, COL9A1, COL11A1, COL11A2
- Significant vision issues
  - Progressive myopathy (8-10 diopters) in 75%
  - Retinal detachment (by age 20) in 70%
- Joint issues – hyperextensibility in 35%
- Growth retardation in 25%

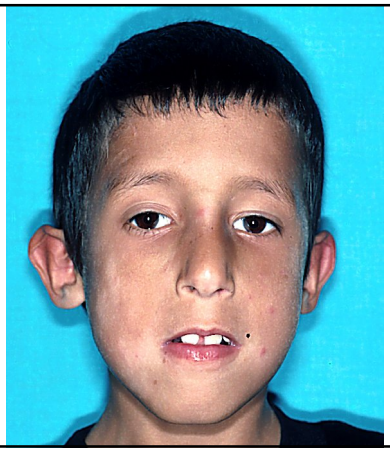
Care of Pierre Robin Sequence

- Cleft care
- Airway issues
- Feeding issues
- Speech and swallowing therapy
- Screen for additional associations



Velocardiofacial Syndrome
AKA DiGeorge syndrome

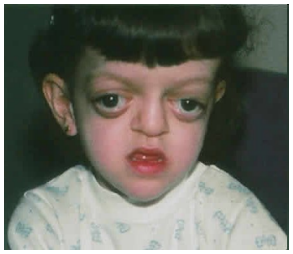
- Clefts, Pierre Robin sequence (~70%)
- Congenital heart defects (~75%)
- Feeding issues (~30%)
- Immunologic issues (~75%)
- Growth (~40% are below 5th%ile)
- Metabolic issues, including calcium
- Developmental issues in 70-90%
- Deletion of one copy of 22q11.2



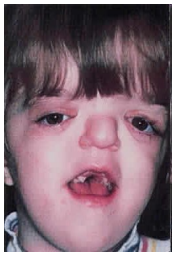
Care of VCFS

- Cleft care
- Screening for and treatment of other associated conditions
- Developmental and educational support

Crouzon



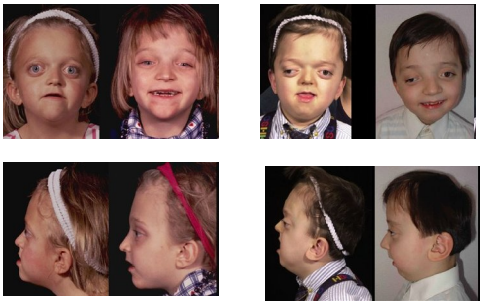
Apert



Crouzon's syndrome

- Craniosynostosis (fused skull bones)
- Wide-set, bulging eyes
- Characteristic jaw
- Less associated with clefts (than Apert)
- Hearing loss
- CNS: occasional MR, seizures, frequent HA
- >35 different mutations in FGFR2, gene found on 10q26.13
- Autosomal dominant, but 50% a new mutation

Before and After - Crouzon



http://www.craniofacial.net/syndromes-crouzon

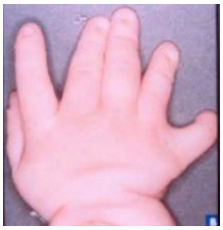
Apert syndrome

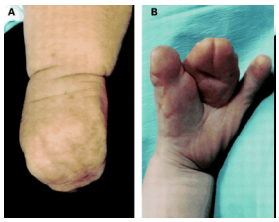
- Craniosynostosis
- Wide-set, bulging eyes (< Crouzon)
- Syndactyly of fingers and toes
- Hearing loss common
- Average IQ of 72
- Clefts in 30%
- 2 mutations of FGFR2 (755C>G, 758C>G) resulting in aa subst: Ser252Trp or Pro253Arg
- Autosomal dominant, but usually new mutation

Before and After - Apert

http://www.craniofacial.net/syndromes-apert

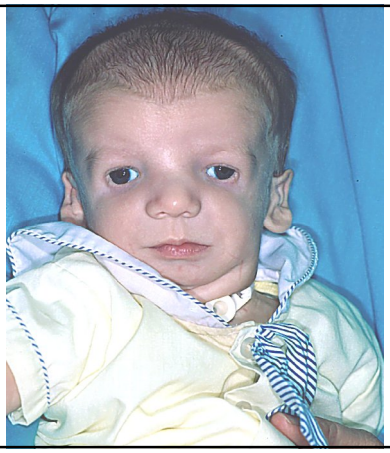
Apert syndrome

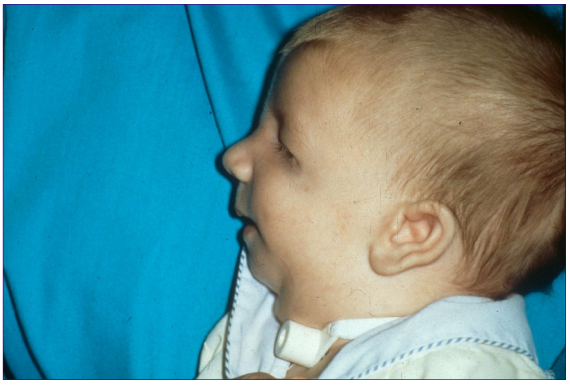




Care considerations Crouzon and Apert syndromes

- Both likely require skull surgeries
- Both have hearing concerns
- Both may have speech concerns
- Apert syndrome more likely to have clefts
- Apert syndrome has orthopedic concerns
- Apert syndrome also may have cardiac, and gastrointestinal issues.
- Both may have spinal issues.





Treacher–Collins Syndrome

- Characteristic skull changes
- Characteristic eye findings
- Ears findings include low set, cup shaped, narrow ear canals, small ears, and conductive hearing loss
- Cleft palate, often with PRS (35%), VPI (60%), macrostomia (15%)
- CNS – normal IQ, low IQ related to hearing loss
- TCOF1 gene (5q32-33.1)
- Autosomal Dominant

http://www.craniofacial.net/syndromes-treacher-collins

Care of Treacher Collins

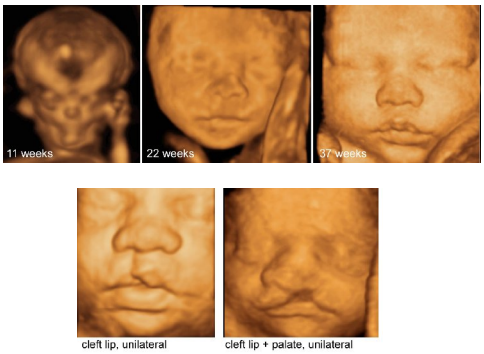
- Cleft care
- Airway issues
- Feeding issues
- Hearing
- Cosmetic surgery

Cleft Care

- Early years
- Preschool years
- School years
- Transition to adulthood

Cleft Care - Early Years

- Initial diagnosis
  - Prenatal
  - At birth
- Early care
  - Feeding and growth
  - (Sometimes breathing)
  - Hearing
  - Development
  - Dental
  - Surgeries – lip, palate, ears

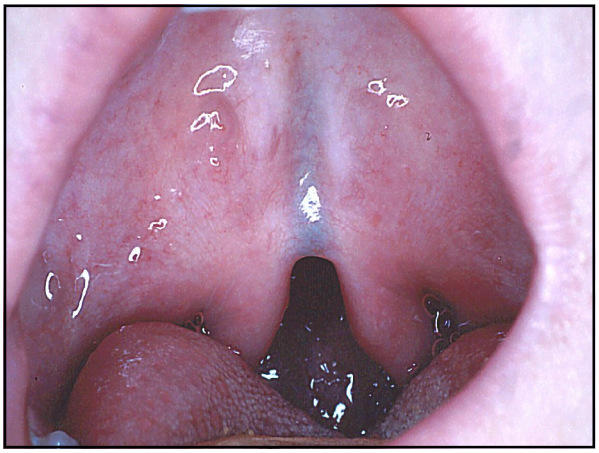


Prenatal diagnosis

- Isolated cleft palate harder to detect
- May alert families and doctors to associated conditions/syndromes
- Allows preparation for feeding and other early issues, including linkages to cleft team

Diagnosis at birth

- Parent reaction
  - Grief: Loss of the 'perfect baby'
  - Guilt: Was it something I ate (took, did)? Or was it something that I did not do?
  - Blame: How did this happen?
- Feeding
  - Less of an issue for isolated cleft lip
  - Breastfeeding usually problematic

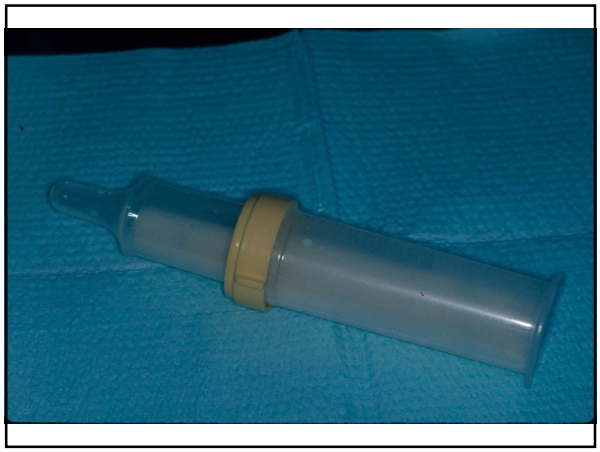


Bifid uvula with submucosal cleft

- One of the more mild cleft palates, but still …
- Issues with feeding and later speech
- Lack of suction due to palatal dysfunction
- May be missed
- May be over-called (thought to be a problem when it is not)

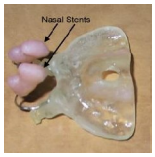
Early feeding

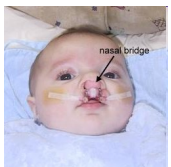
- Basically, pour the food into an infant's mouth (due to the lack of suck)
- Special bottles or nipples are used
- Breastmilk can be pumped, then given
- Weight gain is the sign of success (lack of weight gain, the sign of failure)



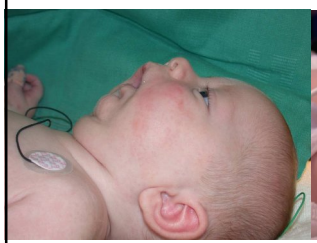
Nasal alveolar molding

- Used prior to repair of cleft lip
- Used to pull the cleft together prior to surgery
- Does not help with feeding



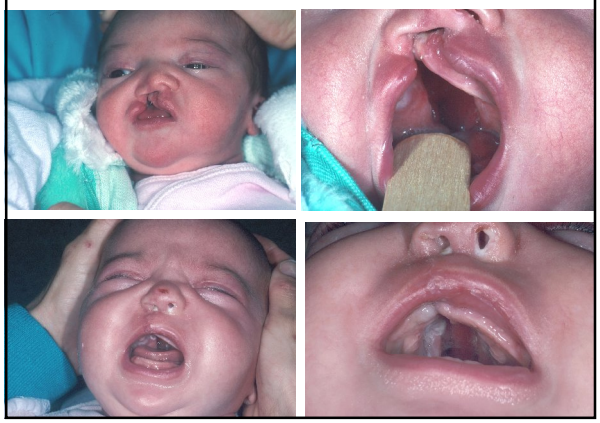


Jaw distraction



Cleft lip repair

- Rule of 10's
  - 10 (to 14) weeks
  - 10 pounds
  - Hemoglobin of 10
- Often with placement of ear tubes



Timing of palate repair

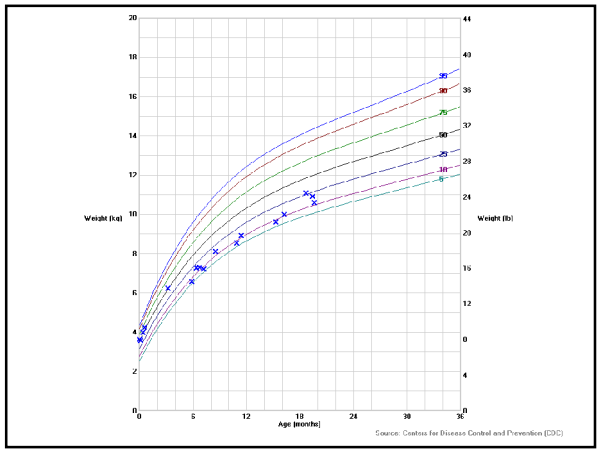
- Cleft of soft palate can be repaired @ 3mo
  - Early closure of clefts in soft palates does not affect facial bone growth
- Hard palate clefts repaired @ 10-14 mo
  - Early closure of clefts in hard palates may affect facial bone growth
  - Late closure of these clefts will affect speech

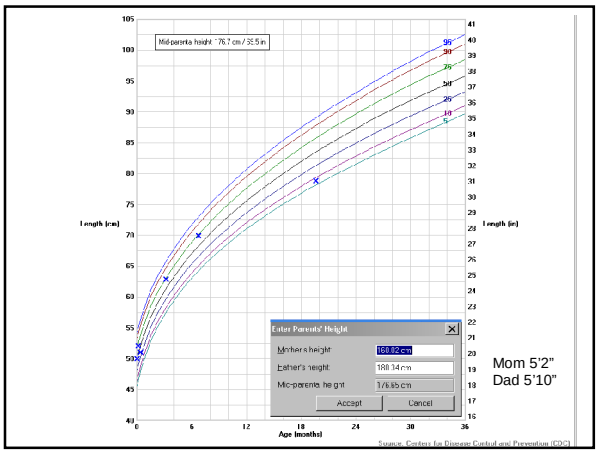
Problems with palatoplasty

- 9-50% require pharyngeal flap
- Non-uniform method to evaluate speech function
- May affects facial growth

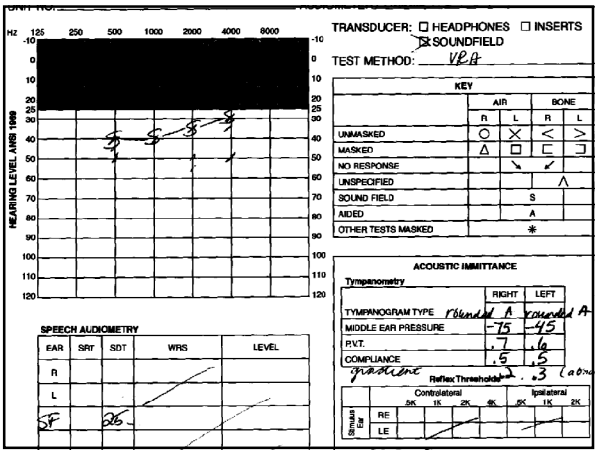
Cleft Care – Preschool Years

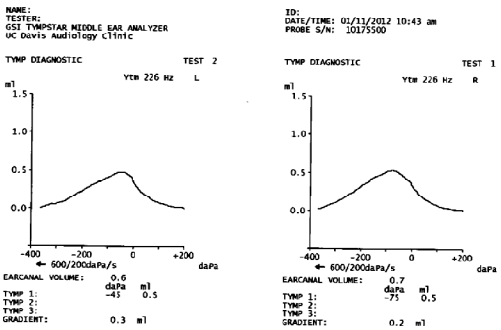
- Feeding and growth
- Speech and hearing
- Development
- Dental
- Surgical considerations
  - Ear tubes
  - VPI (velopharyngeal insufficiency)
  - Fistulas (holes in the repaired palate)





Mom 5'2"
Dad 5'10"





Dental care issues

- Many children with clefts require braces
- Braces accelerates existing dental decay
- Often hard to find dentist who will treat children with clefts
- Even harder to find dentists to treat children with clefts and syndromes, especially those with developmental delay

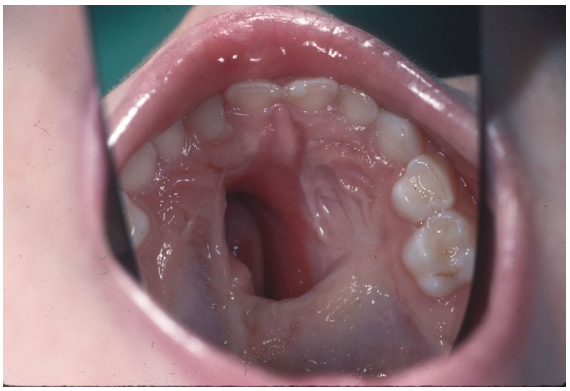
Speech issues

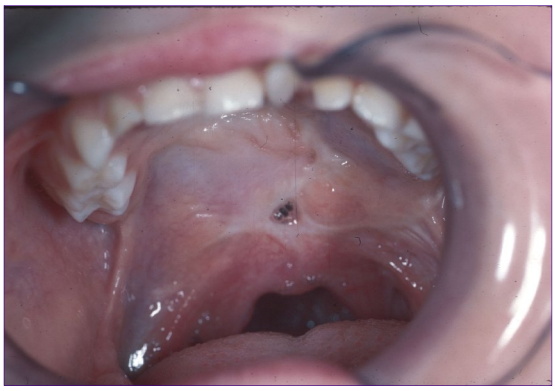
- Some due to cleft
- Some related to surgery
- Some hearing related
- Some developmental

- Early intervention, special education
- Swallowing therapy need for some

Velopharyngeal insufficiency

- Structural insufficiencies
  - Clefts; fistulae
  - Submucous clefts; short palate
  - Mechanical interferences; scarring
- Functional incompetencies
  - Neurogenic problems
  - Mislearning
    - Hearing loss; phone specific VPI





Treatment of VPI

- Some improvement with speech therapy
- Surgery often required to decrease the nasal airflow
- Concern about creating obstructive sleep apnea
- Surgery usually done in 2 stage

Age 8
Bilateral cleft lip and palate
'playing ball'

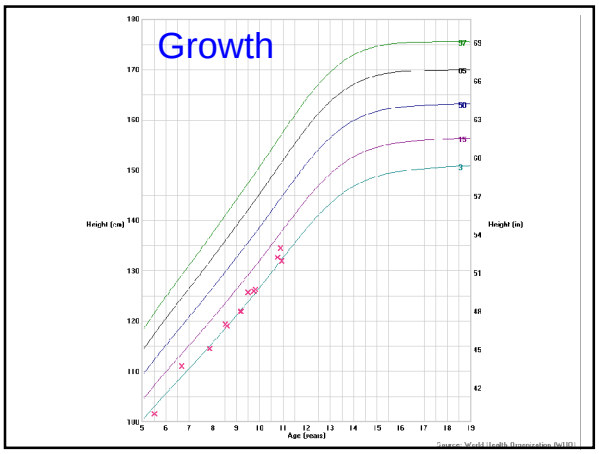
Kinetic Family Drawing: "Draw a picture of you and your family doing something."

Cleft Care – School Years

- Speech
- Growth
- Cosmetic results
- Bullying
- Dental, orthodontic, oral surgery
- Surgical considerations
  - ear tubes or patching ear drums
  - velopalatal insufficiency (VPI)
  - fistulas
  - bone grafting
  - revisions of lips and noses

Speech

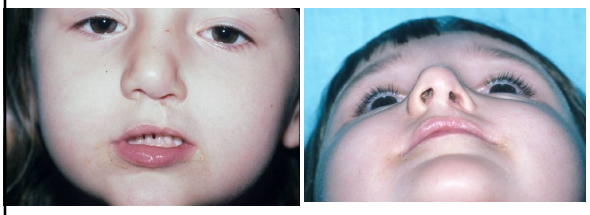
- Speech therapy
  - Largely delivered at schools
  - Sometimes requires surgery for VPI
  - Graduation from speech

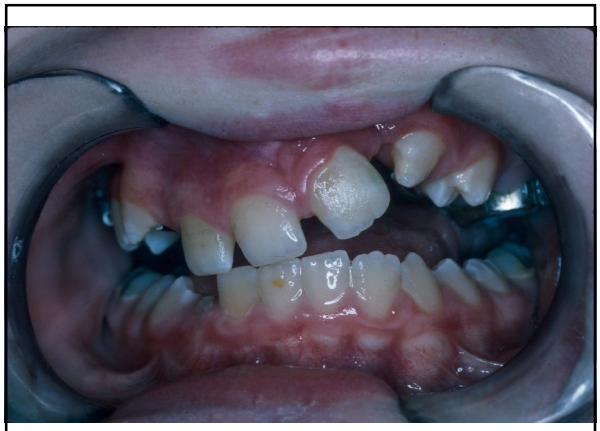


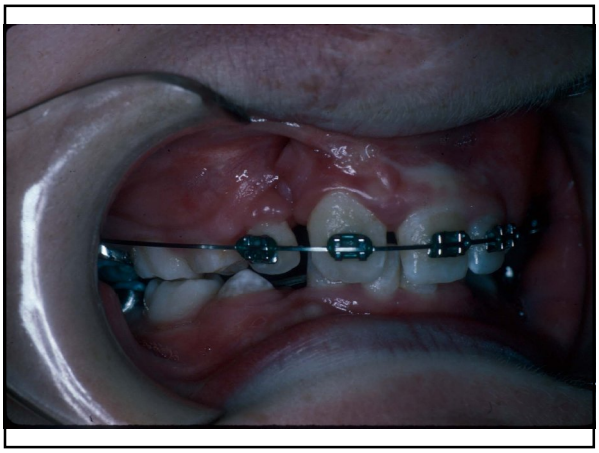
Cosmetic concerns

- Time of self-awareness
- Cleft scars
- Nasal asymmetry
- Dental/orthodontic issues

- Differences leads to bullying or self-isolation







Surgical considerations

- Often school age is a respite from surgery
- But …
  - ear tubes or patching ear drums
  - Pharyngeal flaps for VPI
  - Repair fistulas
  - Bone grafting to build up gum lines
  - revisions of lips and noses

Kinetic Family Drawing: "Draw a picture of you and your family doing something."

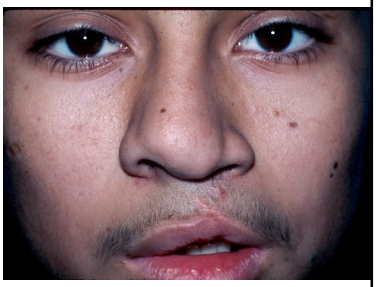
Age 9 years 9 months
Bilateral cleft lip and palate

Cleft Care: Transition to Adulthood

- Cosmetic results
- Risk of transmission
- Dental, orthodontic, oral surgery
- Insurance issues
  - CCS (California Children's Services)
  - Pre-existing condition

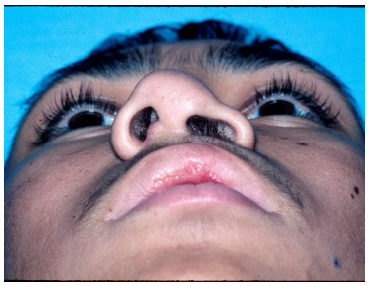
Adult Cleft Nasal Deformity

- Asymmetric tip
- Ala
  - thickened
  - hooding
  - base

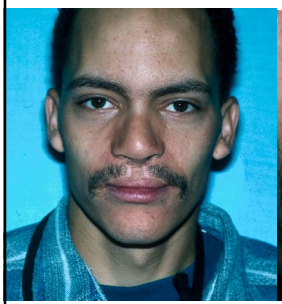


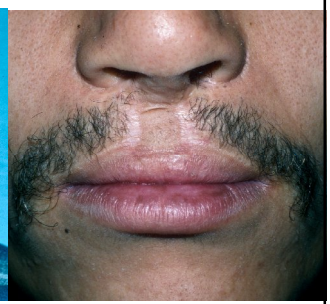
Adult Cleft Nasal Deformity

- Nostril shape
- N/L fistula
- Septal deviation



Prolabial Hair





Risk for Clefting

	%
One affected sibling	2-4
Two affected sibling	4-9
One affected parent	3-4
One affected parent & sibling	13-17
Severe (BCLCP)	doubles risk

Components of Cleft Team Care

- Panel evaluations provide
  - Comprehensive assessments
  - Specialized care
  - Screening for associations
  - Longitudinal assessments
  - Referrals to other specialists

Cleft and Craniofacial Program

Team Members

- Audiology
- Clinical Nurse Specialist
- Facial Plastic Surgeon
- Geneticist
- Oral Surgeon
- Orthodontist
- Otolaryngologist
- Pediatrician
- Social Worker
- Speech Therapy
- Team Coordinator

UC Davis
Cleft and Craniofacial Panel

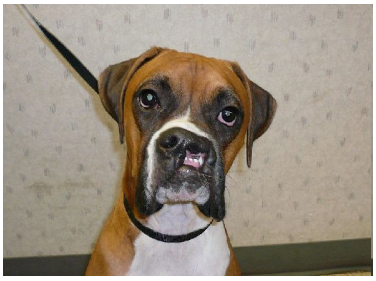
- Panel assessments 2-3 x per month
- Case discussions weekly

www.ucdmc.ucdavis.edu/otolaryngology/specialty/cleft/cleft.html

Notables with clefts

- King Tut
- Tad Lincoln
- Tom Brokaw
- Stacey Keach
- Jesse Jackson
- Payton Manning
- Joaquin Phoenix

Thank you for your attention



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Credit to Dr. Craig Senders for many of the slides