

CORE CURRICULUM

for Cleft Palate & Other Craniofacial Anomalies

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American Cleft Palate –
Craniofacial Association

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CORE CURRICULUM OUTLINE

The American Cleft Palate-Craniofacial Association (ACPA) believes that children born with clefts and other craniofacial anomalies are provided optimum care when they are assessed and treated by a team of specialists with expertise in a variety of areas. Health care specialties involved with the care of clefts and other craniofacial anomalies include audiology, genetics, nursing, oral and maxillofacial surgery, orthodontics, otolaryngology/head and neck surgery, pediatric dentistry, plastic surgery, psychology and clinical social work, and speech-language pathology.

This core curriculum was created by the Education Committee of ACPA to be used as a guide for educators in these various disciplines, when planning the essential parts of their curriculum related to cleft and craniofacial anomalies. It was developed after a survey by ACPA of educators in these disciplines showed a need for such an outline. The Core Curriculum is not intended to cover all possible aspects of cleft and craniofacial management. Rather, it is intended to provide an outline of services that are appropriate for most children affected by these disorders.

The Core Curriculum is divided into two broad sections. The first covers the basics of interdisciplinary team care, classification of craniofacial anomalies, craniofacial development and etiology. The second section covers the role of each discipline in the care of a patient with a cleft or craniofacial anomaly. It is organized by patient age, within each discipline, and covers the essential aspects and knowledge bases that are essential for providing adequate care. Just as in a team there may be overlap between specialists in their observations, core knowledge and treatment expertise, this core curriculum reflects some overlap between specialties in these areas.

CORE CURRICULUM for Cleft Palate & Other Craniofacial Anomalies

SECTION 1: INTERDISCIPLINARY TEAM CARE, CLASSIFICATION, AIRWAY, AND FEEDING

I. Team Evaluation

The initial evaluation of the patient should be by a pediatrician, who is knowledgeable about all aspects of the infant's care. Optimum management of children with clefts and craniofacial anomalies is provided by a team of health care professionals with a specific interest in these anomalies. Team evaluation should be performed early in life and, ideally, the initial contact with the team should be prior to the infant's discharge from the hospital following birth. This allows the parents to receive information about their baby's problem and subsequent treatment, as soon as possible. Team members include specialists from:

- A. Audiology
- B. Genetics
- C. Nursing
- D. Oral and maxillofacial surgery
- E. Orthodontics
- F. Otolaryngology and head and neck surgery
- G. Pediatric dentistry
- H. Plastic surgery
- I. Psychology and clinical social work
- J. Speech-language pathology

II. Classification and Anatomy

The cleft or craniofacial anomaly is usually classified during the initial examination of the infant. Craniofacial anomalies, other than clefts, are discussed in Section IV.

- A. Clefts of the lip and clefts of the palate can occur simultaneously or separately.
- B. The most common classification system for clefting uses the terms primary and secondary palate to define the cleft.
- C. The dividing point of the primary and secondary palate is the incisive foramen. The primary palate is anterior to this anatomic point and the secondary palate is posterior to it.
- D. The primary palate includes:
 1. Lip
 2. Alveolus
- E. The secondary palate includes:
 1. Hard palate
 2. Soft palate
 3. Uvula
- F. Any cleft of the primary or secondary palate may be complete or incomplete,

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depending on whether or not the cleft involves the entire anatomic structure.

G. Any cleft of the primary or secondary palate may be unilateral or bilateral.

H. Submucous clefts of the secondary palate may also occur. These can be detected by visual inspection, ultrasonography or radiography.

III. Airway and Feeding

As with any newborn, the primary concerns in the neonatal period are airway maintenance, breathing, and feeding. Some of the anatomic variations in children with clefts and craniofacial anomalies may have an impact on these functions.

A. Airway maintenance

1. Cleft lip and/or cleft palate rarely cause problems with the upper airway or breathing, when there are no other associated problems.
2. Pierre-Robin Sequence is the most common anatomic deviation associated with clefting that can result in airway and breathing problems.
 - a. Pierre-Robin Sequence results in a combination of malformations, consisting of mandibular hypoplasia, glossoptosis, and midline cleft of the secondary palate.
 - b. Usually airway problems can be managed with prone positioning and time for growth to occur. However, dental prosthetic or surgical intervention, including tracheostomy, may be required in severe cases. In addition, distraction osteogenesis of the mandible has been used to treat some infants with severe airway problems due to Pierre Robin Sequence, but its use at this age is controversial.
3. Craniofacial anomalies can also be associated with airway problems. These include, but are not limited, to:
 - a. Syndromal craniosynostosis with severe midface hypoplasia.
 - b. Any syndrome associated with a severely deficient mandible, such as severe Treacher-Collins Syndrome.
 - c. Choanal atresia.

B. Feeding and Nutrition

1. Babies with isolated clefts of the lip and/or palate can usually feed by mouth with some adjustments to bottle-feeding techniques. Tube feeding is rarely required.
2. Babies with isolated cleft lip may be able to breast feed, but it is unlikely that a child with a cleft palate will be able to successfully breast feed because of nasal spillage and the difficulty in maintaining an adequate suction.
3. Despite the problems with maintaining sucking pressures, the swallowing mechanisms in children with cleft palate are usually normal. Therefore, if the milk or formula can reach the oropharynx, the natural swallowing reflexes can

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move it into the esophagus.

4. Some nasal regurgitation may occur, but this is rarely more than an inconvenience. Upright positioning during feeding may help reduce the occurrence of nasal regurgitation.
5. The strategies that have been developed to feed infants with clefts of the palate are designed to overcome the lack of negative pressure developed during sucking. These include, but are not limited, to:
 - a. Cross-cutting fissured nipples.
 - b. Squeezing a soft bottle to help with the flow of milk.
 - c. Pumping the breasts to deliver breast milk via bottle.
 - d. Developing patience in feeding.
 - e. Feeding instruction and follow-up with a feeding specialist on the cleft palate team.
6. It is important to ensure that the energy that a child expends during feeding does not exceed the nutritional and caloric intake from the feeding. This problem may occur if feeding takes more than 30 minutes.
7. Steady weight gain is the most important indicator of adequate food intake. Close follow-up with a pediatrician or other health care provider is necessary to ensure that consistent weight gain is achieved.
8. Frequently, airway problems will be exacerbated during feeding. The combination of the inability to maintain adequate sucking and airway problems may lead to the need for an alternative feeding method.
9. These same principles apply to babies with other craniofacial anomalies, even though the anatomic cause of their feeding problems may be mandibular or maxillary hypoplasia, rather than clefting.

IV. Noncleft Craniofacial Anomalies

A. Hemifacial Microsomia

1. This is the second most common congenital anomaly of the head and neck after clefting. Hemifacial microsomia includes:
 - a. Malformation of the external ear with varying degrees of microtia and/or other ear anomalies.
 - b. Malformation of the mandible, with varying degrees of shortening or absence of the ramus of the mandible. Subsequent chin deviation and malocclusion will occur.
 - c. Varying degrees of maxillary hypoplasia.
 - d. Facial nerve weakness or absence in severe cases.
2. Team management typically includes:
 - a. Protection of hearing in the normal ear.

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- b. Orthodontic management, combined with rib graft reconstruction or distraction osteogenesis of the ramus, at age 4 to 6 years.
- c. Ear reconstruction with otoplasty or rib cartilage, depending on the severity of the anomaly.
- d. Orthognathic surgery and additional orthodontic management after facial growth is complete.

B. Craniosynostosis

1. Craniosynostosis is early fusion of the sutures between the bones of the skull where growth naturally occurs, thus, precluding growth at the suture site.
2. It can occur in isolation or as a part of several syndromes.
3. Non-syndromal craniosynostosis is classified morphologically by the suture involved and subsequent skull shape.
 - a. One or two sutures involved with different skull and upper face deformities depending on suture.
 - i. Sagittal suture-scapocephaly
 - ii. Unicoronal suture-plagiocephaly
 - iii. Metopic suture-trigonocephaly
 - iv. Bicoronal sutures-brachycephaly or turricephaly or both
 - b. Increased intracranial pressure and developmental delay is rare.
 - c. Correction usually requires one operation in infancy. Secondary surgery is uncommon.
4. Syndromal craniosynostosis is classified according to the name of the syndrome.
 - a. Turribrachycephalic skull shape is common.
 - b. Five syndromes of which craniosynostosis is a part
 - i. Crouzon syndrome
 - ii. Apert syndrome
 - iii. Carpenter syndrome
 - iv. Saethre-Chatzen syndrome
 - v. Pfeiffer syndrome
 - c. All are inherited in an autosomal dominant fashion, except Carpenter syndrome, which is recessive.
 - d. Syndactyly of the hands and feet is part of Apert, Carpenter, and Pfeiffer syndromes.
 - e. Increased intracranial pressure and developmental delay are more common than in non-syndromal craniosynostosis, but are not universal.
 - f. Multiple operations throughout life are usually required to treat these patients.

C. Other Craniofacial Anomalies

1. Orbital hypertelorism

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- a. Orbits laterally displaced making the eyes appear too far apart.
- b. Caused by nasofrontal dysplasia, encephalocele, tumor, or complex craniofacial clefts.
- c. Can be corrected with surgery.
2. Treacher-Collins Syndrome
 - a. Autosomal dominant.
 - b. Includes varying degrees of zygomatic hypoplasia, lower eyelid coloboma, mandibular hypoplasia, and microtia.
 - c. Multiple surgeries throughout life required to treat.
3. Craniofacial clefts
 - a. Clefting can occur in the upper face and forehead, as it does in the lip and palate, and can involve all anatomic layers including bone.
 - b. These clefts are very rare, may be very deforming, and may require multiple surgeries to treat.

V. Craniofacial Development

A. Developmental Craniofacial Biology

1. Molecular regulation of craniofacial morphogenesis
 - a. Facial rhombomeres, HOX and OTX2 genes
 - b. Patterns of neural crest formation, migration, fates.
2. Abnormal neural crest development (neurocristopathies), e.g., Treacher Collins syndrome (mandibulofacial dysostosis), Pierre Robin sequence, DiGeorge sequence, Hemifacial Microsomia.
3. Molecular regulation of skeletal morphogenesis, e.g., Fibroblast growth factors (FGFs) and receptors (FGFRs).
 - a. Fibroblast Growth Factor (FGF) gene
 - b. Fibroblast Growth Factor Receptor (FGFR) gene
4. Molecular regulation of eye development, e.g., PAX6, PAX2, BMP7 genes, and sonic hedgehog (shh).
 - a. PAX6 gene
 - b. PAX2 gene
 - c. Sonic hedgehog (shh)
5. Molecular regulation of palate formation, e.g. Epidermal growth factor (EGF), transforming growth factor- α (TGF α).
6. Genes and tissue interactions in tooth development.
7. "Time table"
 - a. Chronology in craniofacial embryology
 - b. Critical periods of peak morphogenesis

B. Craniofacial Embryology

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1. Development of skull
 - a. Neurocranium: membranous (desmocranium) and cartilaginous (chondrocranium).
 - b. Viscerocranium, e.g. maxilla, palate, mandible.
 - c. Morphogenesis of sutures and synchondroses.
2. Development of the face, eye, nose, lip, palate, tongue, ear
 - a. Facial prominences (5).
 - b. Roles of olfactory, optic and otic placodes.
 - c. Primary and secondary palates.
 - d. Pharyngeal arch contributions to tongue formation.
 - e. Morphogenesis of the ear – internal, middle, and external ears.
 - f. Morphogenesis of the eye
 - i. Optic cup and lens
 - ii. Retina, lens, iris and ciliary body
 - iii. Choroid, sclera and cornea
 - iv. Optic nerve
 - g. Organization and development of orofacial and tongue muscles.
 - h. Morphogenesis of velopharyngeal muscles.
3. Pharyngeal arches and pouches
 - a. Organization of arches and pouches.
 - b. Component tissues of arches and pouches.
 - c. Structures developing from arches and pouches.
 - d. Malformations related to abnormal pharyngeal arch and pouch formation, e.g., branchial fistulae and cysts.
4. Development of teeth
 - a. Stages in typical tooth morphogenesis.
 - b. Tissue interactions in tooth development.
 - c. Development and plan of primary, mixed, and permanent teeth.
- C. Basic definitions in craniofacial biology
 1. Anomaly (Major): Condition often defined as malformations (or defects) that create significant medical problems and require surgical and medical management.
 2. Anomaly (Minor): Condition often described as morphologic features that vary from those that are most commonly seen in the normal population but, in and of themselves, are not associated with increased morbidity.
 3. Association: A group of anomalies that occur more frequently together than would be expected by chance alone but do not have a predictable pattern or unified etiology.
 4. Critical Periods: Intrauterine (chiefly embryonic) periods of peak organogenesis

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- during which time the embryo is at high risk for teratogen exposure.
5. Disruptions: A condition where a fetal structure is growing normally and then growth is arrested by a factor(s) that disrupts the normal development process.
 6. Deformation: A condition (often temporary) caused by an abnormal external force on the fetus during in utero development that results in abnormal form and growth of the fetal structure or region.
 7. Dysplasia: Anomalous development related to an underlying tissue disturbance where the cellular architecture or growth of a tissue is not normally maintained throughout development.
 8. Ectoderm: Outermost of the 3 primary layers that forms the nervous system and outer skin (epidermis).
 9. Endoderm: Innermost of the 3 primary layers that forms the lining of the gut.
 10. Etiology: Underlying factors and causes for congenital anomalies or birth defects. Note that the same apparent conditions may have different etiologies in different individuals.
 11. Facial Prominences: These are the five major building-block structures (frontonasal [1], maxillary [2], and mandibular [3]) that play important roles in the formation of the embryonic head and face.
 12. Fibroblast Growth Factor (FGF): A family of signaling key roles in embryogenesis, including that of the limbs, skeleton, and head and face.
 13. Fibroblast Growth Factor Receptor (FGFR): Protein receptor sites located on cell membranes which bind with specific signaling molecules (e.g. Shh, FGF) that transmit molecular signals to the cell nucleus and the specific development of the cell(s).
 14. Field Defects: Term often used to describe related malformations in a particular region and sometimes used interchangeably with the term "sequence".
 15. Genotype: This is the fundamental genetic constitution or composition of an individual.
 16. HOX genes: A set of homeobox genes with identified DNA sequences controlling those that play important roles in morphogenesis of the body, in general, and in specific structures of the head and face.
 17. Malformation: Malformation signifies that fetal development and growth did not progress normally due to underlying genetic, epigenetic, or environmental factors that altered development of a specific structure or structures.
 18. Mesoderm: Middle layer of the 3 primary layers that forms the dermis, bone, cartilage, blood vessels and connective tissue.
 19. Neural Crest: Layer of cells superior to the developing neural tube that migrate to become part of nearly all major structures and organ systems in the body.
 20. OTX gene: A homeobox containing gene that plays an important role along with

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HOX genes in the embryogenesis of the brain and the morphogenesis of the first pharyngeal arch and its derivatives, especially the craniofacial regions.

21. Pathogenesis: The cellular basis of abnormal development associated with known or hypothesized etiologies.
22. Pharyngeal Arches: Paired arches in embryonic neck region separated by pharyngeal grooves which play important roles in development of the head and neck.
23. Pharyngeal Grooves: Deep depressions between pharyngeal arches in embryonic neck region. The first groove persists and forms the external acoustic meatus.
24. Pharyngeal Pouches: Outpocketings from the embryonic pharynx wall that play important roles in development of structures, such as the tympanic membrane, tonsils, thymus and parathyroid glands.
25. Phenotype: Phenotype is the observed result of the interaction of the genotype with environmental factors, i.e. the observable expressions of a particular gene or genes.
26. PAX gene: The PAX gene family (e.g. PAX2, PAX6) is an important group of genes that play key roles in the morphogenesis of such structures as the ear, eye, and nose.
27. Rhombomeres: Blocks of tissue located lateral to the embryonic hindbrain (rhombencephalon), which provide for fundamental organization of hindbrain and eventually play key roles in facial development.
28. Sequence: A group of related anomalies that generally stem from a single initial major anomaly that alters the development of other surrounding or related tissues and structures.
29. Sonic Hedgehog (shh): A protein "signaling" molecule that plays the most important role in shaping the entire embryo, and in specific structures of the head and face, including teeth.
30. Syndrome: A condition generally recognized and defined as a well characterized constellation of major and minor anomalies that occur together in a predictable fashion presumably due to a single underlying etiology (e.g. genes, chromosomes, teratogens).

SECTION II: GENERAL ROLE OF THE VARIOUS DISCIPLINES IN TREATING PATIENTS WITH CLEFTS AND CRANIOFACIAL ANOMALIES

I. Audiology

The audiologist on the cleft and craniofacial team provides information regarding hearing sensitivity and mechanical function of the ears. Many syndromes with cleft lip and palate as a feature also have a risk for hearing loss. In addition, the function of the Eustachian tube (which connects the space behind the eardrum to the back of the throat) may be impaired by the cleft of the palate, putting the patient at increased risk for frequent ear infections. Stable hearing sensitivity is required for the proper development of speech and language. Children with cleft lip and palate are already at risk for speech and language problems due to anatomic abnormalities of the “articulators.” For this reason, it is important to identify hearing loss early by monitoring their auditory sensitivity on a regular basis, in order to minimize complications of abnormal or fluctuating hearing on speech development.

A. Early Identification

1. Newborn hearing screening: Techniques have been developed to test hearing regardless of age. Hospitals in many states routinely screen the hearing of all newborns. Diagnostic testing is performed in cases where the infant does not pass the screening test.
2. High-risk testing: In states where universal screening is not available, children with craniofacial anomalies are tested because of their high risk for hearing loss status. Although specific test protocols will vary from one facility to another, “high risk” infants should be tested prior to age 4 months.
3. Diagnostic testing: In cases where the infant does not pass the screening test, diagnostic testing will be performed in order to determine the severity of hearing loss as well as the type of hearing loss (“nerve deafness” vs. hearing loss due to ear infection), and whether the hearing loss is in one ear or both.

B. Management

1. Sensorineural hearing loss, or “nerve deafness”, is managed in most cases with hearing aids. The type of hearing aid recommended will depend upon the severity of hearing loss as well as any physical deformity of the external ear. In addition to amplification, early intervention educational services may be recommended with emphasis on language acquisition in light of the hearing loss.
2. Conductive hearing loss due to ear infection or effusion will be managed by a physician, usually either the pediatrician or an otolaryngologist. Once the infection is appropriately treated, the hearing should return to normal. Conductive hearing loss due to anatomical abnormality of the mechanical structures of the ear may be managed by surgery, amplification, or a combination

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of the two.

C. Monitoring

1. Sensorineural hearing loss. Children with cleft lip/palate and sensorineural hearing loss should be tested every 4-6 months in order to assess any progression of hearing loss and to make adjustments to amplification as needed for proper fit as the child grows.
2. Conductive hearing loss. In cases of conductive hearing loss periodic assessment will assist the managing physician by providing feedback regarding the efficacy of treatment in achieving and maintaining normal hearing status.

II. Genetics

The geneticist is responsible for identifying the etiology and/or pathogenesis of the cleft or craniofacial anomaly. The information is then used to discuss overall prognosis for the patient as well as recurrence risk for the parents, patient, and other family members. As with other birth defects, clefts and craniofacial disorders may be the result of chromosomal abnormalities, single gene disorders, and/or environmental factors/agents. Most commonly they are the result of multifactorial inheritance involving the interaction of an individual's genetic background with the environment.

Considerable progress has been made in the identification of causative factors over the past 10 years particularly in the area of single gene disorders. The genes responsible for several of the most well known genetically determined syndromes have been recently identified. However, at the time of this writing, molecular testing is infrequently utilized in clinical management. Working drafts of the human genome sequence have recently been published in Nature and Science. Several surprises have emerged. The number of human genes (roughly 30,000) is far less than originally estimated. Through a variety of genetic mechanisms including alternative splicing and regulation of transcription, the 30,000 genes code for an enormously complex array of proteins. Clearly, biology is no longer "one gene – one protein." It is now known that mutations in different genes may produce the same phenotype (e.g. FGFR1 and FGFR2 in Pfeiffer Syndrome). Different mutations in the same gene may result in different phenotypes (e.g. FGFR3 and achondroplasia, hypochondroplasia, thanatophoric dysplasia, and Crouzon Syndrome with acanthosis nigricans). The tissue distribution of a mutation may produce a range of phenotypes from a multisystem disorder to a tumor (e.g. GNAS1 and McCune-Albright Syndrome, fibrous dysplasia, and pituitary adenoma).

With respect to environmental factors, there are some agents, such as with the acne drug, Accutane, which are potent human teratogens with a high risk for craniofacial malformation in prenatally exposed fetuses, regardless of the infant or mother's genetic

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background. There are factors, such as cigarette smoking, that increase the risk for cleft lip with or without cleft palate only in susceptible individuals. However, the genes that confer susceptibility to most cleft and craniofacial conditions remain to be elucidated. There is currently considerable interest in folic acid as a pre- or peri-conceptual treatment that might reduce the risk for cleft lip and palate as it does for spina bifida. Further study is needed to confirm early reports.

Three types of genetic mutations are under investigation in craniofacial disorders:

- A. Those that increase an individual's susceptibility for a given error in morphogenesis but produce a phenotype only through interaction with other genes or environmental factors;
- B. Those that produce phenotypes directly; and
- C. Those that modify expression of disease producing genes and thus alter the phenotype.

Genetic advances are likely to improve the ability to diagnose and test for syndromes impacting craniofacial development. Understanding of the molecular pathogenesis of a condition will hopefully translate into novel strategies for treatment through manipulation of cellular pathways. Recognition of the factors impacting susceptibility and risk may lead to more effective strategies for prevention.

A. Cleft lip with or without cleft palate (CL_±P)

1. Incidence in the general population is roughly 1:1000, but varies in different racial groups.
2. Although the majority of CL_±P occurs in an otherwise normal individual, between 10% and 20% of affected individuals have the condition as part of a syndrome with broader implications to the individual and family. These conditions need to be identified such that appropriate follow-up is instituted and accurate recurrence risk counseling is offered.
 - a. The majority of syndromes are diagnosed clinically through history and physical examination.
 - b. Chromosomal testing may be indicated when CL_±P occurs with other malformations, growth deficiency, or developmental delay.
 - c. Molecular (DNA) testing is available for a very few specific conditions.
3. For isolated CL_±P, multifactorial inheritance is likely. Empiric risk for recurrence for unaffected parents with one affected child is 4:100 or 4%. This risk also applies to the affected individual's own chance for similarly affected offspring.
4. Prenatal diagnosis for isolated CL_±P depends upon the ability of ultrasound to visualize the fetal face. For syndromes in which CL_±P represents one feature, prenatal diagnosis should be tailored to the underlying etiology of the syndrome.

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B. Cleft palate alone (CP alone)

1. Incidence in the general population is roughly 1:2000.
2. Although the majority of CP alone occurs in an otherwise normal individual, up to 50% of affected individuals have the condition as part of a syndrome with broader implications to the individual and family. These conditions need to be identified such that appropriate follow-up is instituted and accurate recurrence risk counseling is offered.
 - a. The majority of syndromes are diagnosed clinically through history and physical examination.
 - b. Chromosomal testing may be indicated when CP occurs with other malformations, growth deficiency, or developmental delay.
 - c. Molecular (DNA) testing is available for a very few specific conditions.
 - d. Stickler syndrome is a common enough disorder that ophthalmologic evaluation of at-risk individuals is recommended.
3. For CP alone, multifactorial inheritance is likely. Empiric risk for recurrence for unaffected parents with one affected child is 3:100 or 3%. This risk also applies to the affected individual's own chance for similarly affected offspring. The risk is for an infant with CP alone, not CL_±P.
4. Prenatal diagnosis for CP alone is currently not possible.

C. Non-cleft craniofacial abnormalities

1. This group of conditions is highly heterogeneous and runs the gamut from disorders of unknown etiology with a negligible recurrent risk (e.g., amnion rupture sequence) to those in which single gene mutations play the determining role (most of the syndromic craniosynostoses) with a substantial risk for recurrence in some families. Since prognosis and recurrence risk information is specific to each condition, genetic evaluation is encouraged in this population.
2. Prenatal diagnosis may be possible for a few conditions depending upon the etiology, the phenotype produced, and the availability of chromosomal and molecular diagnosis.

III. Nursing

The role of nursing in the care of patients/families affected by craniofacial anomalies is multifaceted including education, case management, consultation, research, and primary care. Early intervention consists of assistance with infant feeding, access to team care, and family education. The nurse continues to interact with the family throughout all phases of the treatment period to assist them in understanding and complying with the recommended treatment plan, as well as providing crisis intervention and anticipatory guidance.

A. Prenatal

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1. Assist with family education about clefting and other craniofacial disorders and team care after birth.
 2. Provide information about potential feeding issues.
 3. Introduction to Parent Support Network if applicable.
 4. Provide direct contact information for team evaluation.
- B. Neonatal
1. Initial contact with newborn in birth hospital – discussion of newborn care, team care, and early cleft management, feeding, resources, support group.
 2. Modeling acceptance of child with craniofacial malformation.
 3. Ongoing follow-up of feeding and weight gain after discharge, directly or through consultation with primary care physician.
- C. Infant/Toddler
1. Preoperative preparation for surgical procedures, discharge teaching, and follow-up.
 2. Ongoing coordination of team services/care.
 3. Ongoing support of family.
 4. Feeding issues – introduction of solid foods, prevention of bottle caries, weaning from the bottle, cup feeding.
 5. Anticipatory guidance regarding growth and development issues; particularly encourage parenting techniques that promote speech development.
- D. Preschool/School-Aged/Adolescents
1. Preoperative preparation that involves both the child and family.
 2. Assistance with initiation of speech therapy and advocacy in the IEP process in obtaining services from the school district.
 3. Ongoing evaluation of audiology and ENT concerns.
 4. Referrals for social skills, self image concerns.
 5. Introduction to other similarly affected patients/families.
 6. Continued emphasis on multidisciplinary team care services.
 7. Referral of adolescent/adult for genetic counseling.

IV. Oral and Maxillofacial Surgery

This discipline is concerned with the occlusion and facial form of patients with cleft and craniofacial anomalies. They work with other members of the team to ensure harmonious and appropriate dental arch form and facial form. Although there may be overlap with plastic surgery and otolaryngology and head and neck surgery in some areas, the oral and maxillofacial surgeon manages the alveolar cleft and skeletal problems related to cleft and craniofacial anomalies such as maxillary hypoplasia and other skeletal malocclusions.

A. Prenatal

May counsel families regarding prenatal diagnosis and implications.

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B. Neonatal

May be involved in airway management. See Otolaryngology section for details.

C. Infant

Early bone grafting of the cleft alveolus has been performed by some at this time but this is a controversial procedure that has been associated with poor mid-facial growth and class III dental malocclusion.

D. Toddler

Occasionally primary teeth in the line of the alveolar cleft will need extraction at this age.

E. Preschool

See Toddler section above.

F. School-Aged

1. Bone grafting of the alveolar cleft is usually done during the period of mixed dentition.
 - a. Age 6 to 10.
 - b. Orthodontic care prior to bone grafting to align the dental arches on either side of the cleft.
 - c. Sometimes teeth in and around the cleft can be salvaged with bone grafting saving the need for prosthetic dentistry later.
 - d. Depending on the size of the cleft in the alveolus the source of the bone graft may be the iliac crest, calvaria, or bone bank.

G. Adolescents

1. It is during this time when skeletal maturity is reached that consideration is given to maxillary or mandibular osteotomies to normalize occlusion and facial form.
2. Patients with cleft lip and palate have a significant incidence of class III skeletal malocclusion with mid-face hypoplasia.
3. This can be corrected with a Lefort I osteotomy of the maxilla sometimes combined with an osteotomy of the mandible. For slight anterior dental cross bites and orthodontic mid-facial protraction, a facial mask may be used.
4. Distraction osteogenesis has been used to correct these skeletal occlusal problems as well.
5. Skeletal surgery may be carried out in adulthood as well.

V. Orthodontics

Orthodontists are involved with the study and guidance of the growth and development of the face, and dentition of the child with a cleft or craniofacial anomaly from birth to maturity. Their role includes diagnosis of changing facial morphology and function due to treatment and growth. They provide orthodontic and orthopedic treatment and general expertise for consultation with all of the other members of the cleft and craniofacial team.

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Due to the long-term treatment required for the majority of these patients, different phases of active treatment, interspersed with periods of retention or no treatment, will be necessary.

A. Prenatal – none

B. Neonatal

1. Pre-surgical infant orthopedics is sometimes used to reposition the segments of the cleft maxilla prior to lip repair. This can vary in complexity from lip taping to narrow the cleft, to a bonnet with elastic to ventroflex a protruding premaxilla, to more complex pinned appliances.
2. These appliances can make lip closure easier. While this short-term benefit is clear, long term effects are unclear and controversial.
3. Some clinicians use orthopedic appliances to alter the appearance of the nose and/or columella to improve the shape prior to lip repair.

C. Infant

When the primary teeth begin to erupt, the parents are advised as to the possibility of dental irregularities, particularly an incisor or supernumerary tooth erupting into the palate. The long-term sequence of treatment is outlined in general terms.

D. Toddler

No specific treatment is indicated, but digit habits and functional shifts may be addressed. Communication with the primary care dentist/pedodontist is established and future concerns outlined.

E. Preschool

1. In some cases, the maxilla may be expanded in order to improve dental function, eliminate functional shifts, to provide access for restorative care to carious teeth impacted in the cleft site, and/or to improve the nasal airway. However, long term retention is needed to maintain the expansion.
2. Oronasal fistulae are sometimes a concern because of liquids escaping through the nose. The anterior part of the cleft may have become hidden as the maxillary segments moved together after lip repair, and this area may not have been repaired during palatoplasty. Consequently, palatal expansion may expose this oronasal communication. Surgical closure is often difficult, and the orthodontist may elect to use an obturator to close off the fistula.
3. A reverse pull headgear may be considered to protract the maxilla and maintain normal jaw relations. This is an effective treatment modality but requires considerable compliance on the part of the patient. Overall success is also uncertain due to the difficulty in anticipating future jaw growth when trying to compensate for inadequate maxillary growth.

F. School-Aged

1. Fixed appliance therapy usually occurs in the mixed dentition between the ages

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of 7 and 9 years, with the goal of preparing for alveolar bone grafting.

2. This phase usually involves aligning malpositioned incisors and expanding the maxillary arch to an appropriate relationship with the lower dental arch. When this is complete, an alveolar bone graft is placed and any oronasal fistulae closed. Maintenance of expansion with a palatal bar or removable appliance is required for some time since the grafted maxilla is unable to maintain the corrected arch form.
3. Reverse pull headgear therapy may be initiated or continued during this time period.

G. Adolescents

1. When the permanent teeth have erupted, definitive orthodontic treatment begins.
2. Treatment may involve surgical or orthopedic repositioning of the jaws to optimize jaw relations and occlusion. Close cooperation between the orthodontist, surgeon, prosthodontist (if necessary), and general dentist is required during this time.

H. Adults

Adults generally require the same treatment as children and adolescents with some possible exceptions. Since adults have completed growth, no possibility exists for influencing jaw growth through orthopedics. Additional or more extensive surgery may be required to achieve the same result. Alveolar bone grafts are less successful in adults, and thus may not be indicated if a graft would not carry significant benefits. Otherwise, a properly treated patient should have the same dental status as a non-cleft person. All aesthetic and functional goals can and should be addressed.

I. Record keeping

This is an important part of the orthodontist's role on the cleft and craniofacial team, as it is necessary for assessment of treatment results.

1. Infant – Photographs should be taken regardless of any treatment. Casts should be made prior to and following any pre-surgical orthopedic treatment. Infant casts are important to assess the wide variability of cleft morphology and to compare the results of different treatments over time as growth occurs.
2. Preschool – Records taken during this time period depend upon treatment rendered. If palatal expansion is done, casts, photos, and a posteroanterior cephalogram are important to assess the result of treatment.
3. School aged – Full or orthodontic records should be taken prior to any orthodontic intervention, including incisor alignment and palatal expansion. These records should include, but are not limited to casts, photos, radiographs (panoramic, occlusal, periapical, and lateral/submentovertex/posteroanterior

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cephalograms), and clinical examination. Further, appropriate records, such as casts and photos, should be taken after treatment.

4. Adolescents – Full orthodontic records as above should be taken before and after definitive orthodontic treatment. Progress records should be taken before and after orthognathic surgery, and more often as necessary.
5. Adult – Full records should be taken as described above.

VI. Otolaryngology

Otolaryngology is a surgical and medical specialty concerned with head and neck disorders, including congenital malformations and the problems associated with them. In patients with craniofacial disorders, including cleft lip and palate, these areas include: surgical management of the cleft lip and/or cleft palate, airway and breathing problems, feeding and swallowing dysfunction, disorders of the ears and hearing, as well as voice, speech, and resonance disorders. Otolaryngologists interact closely with plastic surgeons, oral/maxillofacial surgeons, pediatricians, speech pathologists and audiologists on the Cleft Team due to the extensive overlap in their areas of expertise.

A. Prenatal

The otolaryngologist may be one of the specialists to provide prenatal counseling to parents whose child has been found to have cleft lip/palate on screening or high resolution ultrasonography. Many parents have considerable anxiety and concern about the cleft, the treatment and the necessary surgical procedures their infant will face soon after birth. This consultation can provide reassurance, assistance with anticipated feeding or breathing difficulties, and information regarding the planning of the primary surgical procedures for reconstruction of the cleft lip and palate. The parents can also be counseled about anticipated ear or hearing problems. During this consultation parents can be prepared for their child's appearance by showing them pre and post treatment examples of patients with cleft lip and cleft palate.

B. Neonatal

In the initial period after birth, it is critical for the child to breathe and feed well.

Neonates with cleft palate may demonstrate some degree of respiratory distress, especially if micrognathia is present. The otolaryngologist is often involved with the evaluation and management of the airway. Initial management of the airway may involve infant positioning, use of nasal airways, oral appliances or feeding tubes to help hold open the airway. Tests such as sleep studies, pulse oximeter monitoring and blood gases are often beneficial. When these techniques are insufficient to maintain the airway, more invasive procedures may become necessary including: intubation,

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mandibular distraction, tongue-lip adhesion, and tracheotomy. If children are being considered for these procedures airway studies with rigid endoscopy are often used to rule out other unexpected airway pathology.

It is important to differentiate between primary feeding problems and feeding problems secondary to airway issues. Babies who have difficulty maintaining a stable airway may exhibit feeding difficulties. Initially, some children with cleft palate may show some discoordination of the “breathe, suck, swallow” reflex resulting in nasal regurgitation of feeds or intermittent choking spells. These episodes often are self limited and the infant soon learns to prevent nasal regurgitation on his own and coordinate his swallowing . Assistive devices may involve use of orogastric or nasogastric tubes. Because the infant with a cleft palate is unable to generate suction, he usually has difficulty breastfeeding and using standard nipples and bottles. Specialty bottles, such as the one produced by Mead-Johnson™ and one way valve nipples such as the Haberman™ and Pigeon™ systems are very helpful in feeding babies with cleft palate. Some centers may fabricate acrylic obturators to close off the palate to allow sucking and facilitate feeding. Nasogastric feeding tubes may be placed to supplement nutrition. Gastrostomy feeding tube placement may be recommended when feeding is significantly dysfunctional.

With universal infant hearing screening programs mandated in most states, the majority of infants will have their hearing screened prior to discharge from the hospital. If the baby is stable, it is best to perform this hearing screening within the first few days of life, as many children with cleft palate initially aerate their middle ear, only to have effusion develop soon afterward. The screening methods are most useful during this effusion-free period, as conductive hearing loss is associated with the presence of effusion resulting in a failed screening. Consultation with the otolaryngologist is recommended for children who fail to pass their hearing screen.

C. Infant

Surgical management of the cleft defects in the lip and palate is performed by most surgeons within the first year of life. Most cleft surgeons follow the surgical guidelines described in Infant management of the Plastic Surgery section.

Nearly all babies with cleft palate (and some with cleft lip only) will develop Eustachian tube dysfunction with development of middle ear effusions and associated conductive hearing loss. Surgical placement of tympanostomy tubes (or pressure equalizing tubes, PET's) is the currently recommended treatment to relieve the middle ear fluid. The timing of tube placement varies between team-

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based otolaryngologists, with most being placed at the time of the cleft lip repair, or with the cleft palate repair. The tube functions by allowing air to enter the middle ear through the hole in the tube, preventing fluid buildup and reducing conductive hearing loss due to the fluid. Hearing tests are recommended following the tympanostomy tube placement to confirm normalization of hearing.

Ongoing management of existing and identification of new airway or breathing difficulties following the primary repairs of the lip and palate continue through the first year of life.

D. Toddler through school aged children

Regular visits every six months are recommended to monitor the status of tympanostomy tubes and signs of normalization of middle ear function. Audiometric assessment is advised with a 6-12 month interval, or sooner if hearing loss is suspected. Tube replacement is often needed into the preschool years to treat persistent middle ear effusion. Amplification of hearing loss not due to effusion is recommended as soon as it is diagnosed. Possible interventions for hearing loss (other than due to middle ear effusion) include: traditional hearing aids, bone anchored hearing aids (BAHA), and cochlear implants.

Obstructive sleep apnea and nasal airway obstruction may occur during this time period and should be screened regularly. These symptoms include loud snoring, gasping or pauses in sleep breathing pattern, restlessness during sleep, enuresis after successful toilet training, and witnessed apnea. Diagnosis may require clinical sleep studies. Treatment is directed to the cause of the obstruction, with tonsillectomy being the most commonly recommended procedure. Except in very special circumstances documented by detailed study of airway pathology, adenoidectomy and septal surgery should be avoided in this age group due to speech and facial growth concerns.

Speech is developing in the toddler age group and frequently a referral to the speech pathologist is made for monitoring and diagnosis of disorders of articulation and resonance. Resonance disorders are screened for and may be evaluated with nasopharyngeal endoscopy and/or videofluoroscopy. Surgical or prosthetic treatment of hypernasal resonance may be warranted. Please see the section in Plastic Surgery regarding options of surgical management. Voice disorders such as hoarseness and breathiness may require further evaluation by the otolaryngologist.

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Revision lip and nasal reconstructive surgeries may be considered for residual lip or nasal deformities prior to entering school.

Children with clefts of the alveolus usually require treatment of the alveolar cleft at this time. Please see the orthodontic and oral and maxillofacial surgery section for details of this treatment.

E. Adolescents and Adult

Teens and adult patients with cleft related nasal deformities are candidates for cleft septorhinoplasty, once facial growth has neared completion around 15-16 years of age. Correction of septal deviation and hypertrophied turbinates can be performed simultaneously with improvement of the nasal airway. Surgical management is also directed towards correction of any residual cleft nasal deformities.

Adult cleft palate patients with significant sinus and nasal problems may have undiagnosed nasopharyngeal reflux of food and liquids that causes nasal contamination. It is advised that these patients undergo an assessment of NP function with nasopharyngoscopy with a liquid swallowing challenge during the procedure.

Ear problems and obstructive sleep airway conditions may persist in this age group and require continued follow-up and treatment.

VII. Pediatric Dentistry

The role of pediatric dentistry in treating individuals with cleft and craniofacial anomalies is the comprehensive preventative and therapeutic oral health care of children from birth through adolescence and special patients beyond the age of adolescence who demonstrate mental, physical, and/or emotional problems. In addition, the pediatric dentist should provide preventative counseling and caries control to maintain the child's oral cavity in a state that maximizes the outcomes of therapies provided by other team members.

A. Prenatal

1. Parental information and support.
 - a. Maximize the families' support network.
 - b. Minimize the transmission of cariogenic bacteria from the parents to the child.
2. Provide information to parents about neonatal treatment options.
 - a. This will maximize their ability to make informed decisions about treatment options such as pre-surgical infant orthopedics.

B. Neonatal

1. Parental information and support.

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2. Pre-surgical infant orthopedics (see the orthodontic section for more information).
 3. Growth and development monitoring.
 4. Caries prevention.
- C. Infant
1. Caries prevention counseling.
 2. Peri-operative care.
 3. Infant orthopedics continued.
 4. Growth and development monitoring.
- D. Toddler
1. Caries prevention.
 2. Growth and development monitoring.
- E. Preschool
1. Caries prevention.
 2. Growth and development monitoring.
 3. Behavior modifications.
 4. Routine dental care.
 5. Interceptive orthodontics where appropriate.
 6. Restorative procedures.
- F. School-Aged: Same as preschool plus preparation for alveolar bone grafting where necessary.
1. Oral hygiene guidance.
 2. Removal of primary dentition in surgical site.
- G. Adolescents
1. Oral hygiene.
 2. Periodontal concerns.
 3. Support during comprehensive orthodontic treatment.
 4. Caries prevention.
- H. Adult
1. Preparation for transfer to general dentist or other dental specialist.
 2. Monitor third molar and refer to OMFS where appropriate.

VIII. Plastic Surgery

Plastic surgery is the surgical discipline concerned with the restoration of normal form and function for patients with cleft and craniofacial anomalies. This is accomplished through appropriately timed operations throughout the patient's life. Some deformities can be reconstructed with one operation early in infancy and others require multiple surgical treatments as growth and development occur. There may be overlap with oral and maxillofacial surgery, and otolaryngology, head and neck surgery in the performance of

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these procedures. The goal is always to have normal function and appearance throughout a patient's life, realizing that this cannot always be accomplished because of anatomic or developmental considerations and how they relate to the timing of surgery.

A. Prenatal

1. Prenatal diagnosis of cleft and craniofacial anomalies is becoming more frequent with ultrasound.
2. Counseling regarding the implications and subsequent treatment may be carried out prior to birth.
3. Although fetal surgery has been done in animal models for cleft repair, this is not an accepted procedure for cleft repair at present.

B. Neonatal

1. Please see Section I for a detailed discussion of team evaluation and cleft classification.
2. Some surgeons are advocating cleft lip and palate repair in the neonatal period. The advantages of this approach have not been proven and the risk of complications is higher.

C. Infant

This is the typical time when surgical closure of the lip and palate is accomplished.

1. Cleft lip is usually surgically closed in the first 2 to 3 months of life when it is clear that the baby is healthy and thriving. Most surgeons still use the rule of tens to plan the timing of closure.
 - a. Ten weeks.
 - b. Ten pounds.
 - c. Hemoglobin of ten.
2. Some surgeons perform lip adhesion prior to definitive lip repair. This procedure is a partial lip repair that does not rearrange the structures into normal anatomic position. Its purpose is to narrow the cleft making the final lip repair easier.
3. The goal of the lip closure is to create a lip that functions well and approximates the physical characteristics associated with a non-cleft lip. The physical characteristics of the nose will also be improved by the lip closure. Sometimes lip revision will be required to improve the result, but the first operation generally provides a dramatic and lasting improvement in the function and appearance of the baby's lip.
4. The timing of palate closure varies from team to team but is usually carried out from 6 months to 18 months of age.
5. In children with airway problems or extremely wide palatal clefts, closure may be delayed.
6. The reason to close the palate is so that speech will develop normally and the patient will not regurgitate liquids and solids into the nose when eating.

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7. Every sound in the English language except M, N, and NG resonates orally.
8. When a palate and cleft is not closed, resonance is hypernasal and multiple errors in speech development will occur.

D. Toddler

1. Despite closure of the palatal cleft, many children with cleft palate will still require speech therapy and approximately 10 to 20 percent may require secondary surgery for persistent hypernasal speech after closure.
 - a. This is called velopharyngeal insufficiency (VPI).
 - b. VPI becomes evident at age 2 to 3.
 - c. Rarely can occur without cleft palate.
 - d. Secondary surgical procedures to correct this problem are
 - i. Posterior pharyngeal flap
 - ii. Pharyngoplasty
 - iii. Augmentation of the posterior pharyngeal wall
 - iv. Speech prosthesis
 - e. See Speech Language Pathology section for more details on evaluation.

E. Preschool

Nasal reconstruction may be performed just prior to kindergarten, possibly combined with a lip revision. These procedures are performed to improve function of the lip and nose, and to ensure that the child will look their best at a critical time of increased peer interaction when they begin school.

F. School-Aged

1. Dental concerns usually are primary during this time as orthodontics and alveolar cleft bone grafting are carried out.
2. See the Orthodontics and Oral and Maxillofacial sections.

G. Adolescents

1. Some children with clefts develop maxillary retrusion requiring jaw surgery to align their dental arches after their facial growth is complete (usually age 14 to 18).
2. After this is accomplished a final septorhinoplasty may be performed to improve breathing and nasal aesthetics.

H. Adult

1. Most patients have completed treatment by the time they reach adulthood.
2. Surgical revision is usually successful in treating any residual problems.

IX. Psychology and Clinical Social Work

The psychologist provides evaluation of, and treatment for, emotional, learning, developmental, and adjustment disorders. This generally occurs within the context of the patient's family. Particular attention is focused on the manifestations of appearance and

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speech on the patient's self esteem and coping strategies for the patient and family in dealing with issues related to multiple operations. The clinical social worker may also focus on many of these concerns as well as using their expertise to obtain services when needed for patients.

A. Prenatal

Assist with prenatal counseling regarding future expectations of development and coping with the unexpected intrauterine diagnosis of a child with a cleft or craniofacial anomaly.

B. Neonatal

1. May assess high risk infants for risk of developmental disorders.
2. May also assist parents with stresses related to children with facial deformity or other developmental problems.
3. Family support groups such as groups of parents of children with clefts or craniofacial anomalies can be very important to some parents in helping them cope with the birth of a child with a cleft or craniofacial anomaly. This can continue throughout childhood and adolescence.
4. In children with high risk for developmental problems, early referral to an infant program may be beneficial.

C. Infant

1. Infant assessment includes developmental assessment of motor and language development, and social responsiveness.
2. Continue to assess the family.

D. Toddler

1. Toddler assessment of self help skills, social development, and motor/language development.
2. Continue to assess the family.

E. Preschool Development

1. Evaluate language and intellectual development.
 - a. Expressive vs. Association language disorders are frequent and need to be carefully monitored.
 - b. Early verbal IQ deficits are common and may affect overall IQ scores.
2. Early Social Interactions.
 - a. Parent-child interactions.
 - b. Overprotectiveness may be present in parents of children with facial deformities and this should be monitored and counseling provided when needed.
3. Developmental Assessment.
 - a. Need for early assessment due to high frequency of early delay.
 - b. Validity problems of early assessment make it necessary to avoid rigid

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- establishment of intellectual ability.
4. During the preschool years delays in development frequently first manifest themselves. The psychologist and speech language pathologist are the team members most likely to diagnose and recommend specific interventions to maximize the patient's potential development when delay is present.
- F. School-Aged Child
1. Learning disorders in children with clefts and craniofacial anomalies.
 - a. Reading disorders
 - i. Need for early screening intervention and remediation
 - ii. Reading problems related to speech problems are common
 - iii. Reading comprehension problems related to language problems may occur, thus reading evaluation should include assessment of both word recognition and reading comprehension
 - b. Memory disorders
 - i. Late development of auditory memory
 - ii. Learning problems related to short term memory are frequent, therefore screening of short term memory or word finding problems is important (dysnomia)
 - c. Language disorders (common types)
 - i. Dysnomia (word finding problem)
 - ii. Expressive Dysphasia (verbal expression of ideas problem)
 - iii. Associative Dysphasia (understanding of language problem)
 - iv. Behavioral Problems
 - d. Acting out behaviors
 - i. Related to early parent overprotection
 - ii. Related to language disorders
 - e. Behavioral inhibition
 - i. Anxious withdrawal
 - ii. Passivity (non-anxious) to avoid teasing
 2. Teacher Expectations
 - a. Teacher perceptions of ability is often underestimated.
 - b. Self fulfilling academic expectations of teachers translates to underachievement.
 3. It is during this time that children and their peers become aware of how they look. Deformities can lead to problems with teasing and self esteem in patients with clefts and craniofacial anomalies. The psychologist can help with coping strategies for patients and their families and can give the surgeon advice about the timing of operations for appearance during this critical time. The clinical social worker can aid in appropriate placement within the educational system.

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G. Adolescents

1. Self-esteem
 - a. Realistic vs. unrealistic self perception of appearance and/or speech.
 - b. Social skills training may help to overcome hypersensitivity.
2. Behavioral Inhibition and Social Introversion
 - a. Depression/anxiety treatments may be indicated.
 - b. Social introversion as a way of life may result in lowered self expectation.
3. Dating and Self-Concerns
 - a. Cognitive behavior modifications may help through the use of self talk to provide strategies for coping with anxiety-provoking social situations.
 - b. Group counseling can be especially helpful with groups of peers with similar conditions.
4. During adolescence there is a heightened self-awareness of body image and greater existential worry about “who am I?”, “what is my identity”. Most adolescents experience these issues; however, the adolescent with facial differences or speech problems may experience a greater sense of “being different”, leading to greater emotional turmoil.

Also, adolescence is a period when there is often a decrease in open communication with parents and other adults. Therefore, it is important for the team psychologist, and/or social worker, to communicate and screen adolescents for possible emotional/social concerns. Monitoring of school achievements, peer activities, and social interactions may reveal when problems are occurring.

H. Adulthood

1. Social Adaptation
 - a. Marriage aspirations
 - b. Activities
2. Educational/Vocational Aspirations
 - a. Achievement motive
 - b. Work aspirations

X. Speech and Language Pathology

The speech/language pathologist provides evaluation and treatment of four communication parameters for patients with clefts and craniofacial anomalies, from infancy through adulthood. These parameters include resonance, articulation, phonation, and language development. The goal of the speech/language pathologist is to facilitate normal speech and language development. This is achieved by providing education concerning speech and language development, recommending and providing speech therapy, and as the child matures, by providing more direct perceptual, acoustic, sound pressure, radiologic, and aerodynamic measurements of the velopharyngeal mechanism. Dental, hearing, prosthetic,

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and surgical interventions must be factored into all management considerations. If velopharyngeal insufficiency is suspected, and palatal management is considered, direct visualization of the velopharyngeal mechanism during speech production is required, with repeat studies following surgical or prosthetic management.

A. Neonatal and Infancy

1. Monitor and assess feeding, swallowing, and hearing ability.
2. Discuss the following areas with family: language, cognition, and speech development with and without a cleft palate or palatal dysfunction.
3. Monitor and stimulate receptive and expressive language and cognitive development.
4. For babies with cleft palate, facilitate oral communication by emphasizing all vowel sound production and those consonants produced by the lips and anterior tongue, which are nasal, or require little intraoral air pressure (/m/, /n/, /w/, /l/, and "y"). Avoid consonant constrictions that are made in the back of the throat, in the glottal area, or made by the posterior tongue to posterior pharyngeal wall. Also, avoid excessive yelling and screaming.

B. Toddler (≤ 3 years)

1. Monitoring the patients' general communication development, motor skills, and cognition.
2. By this age, patients have usually undergone lip and/or palate repair, and their speech, language, resonance, and voice needs to be assessed with consideration for early speech and/or language therapy; with more global delays, an early childhood program should be instituted.
3. Nasal consonant substitutions may be observed. These occur when the speech articulators are placed appropriately for the intended oral consonant, but due to incomplete palatal closure, the speech sound is produced as a nasal consonant (/b/ becomes /m/; /d/ becomes /n/).
4. Compensatory substitutions may be noted. These are unconsciously learned speech patterns that occur when the articulators are positioned inappropriately in an effort to produce oral consonants. These are commonly heard in attempted production of plosives (sounds created by complete blockage of airflow followed by buildup of pressure which is suddenly released, such as /b/) and *fricatives* (sounds characterized by turbulent noise, such as /s/).

If adequate oral pressure cannot be achieved with typical placement of the articulators, then an alternative constriction site may be used and pressure is created below the level of constriction. Some common compensatory articulations include:

- a. Glottal stops – closure of the vocal folds at the level of the glottis.

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- b. Pharyngeal fricative – posterior positioning of tongue to posterior pharyngeal wall, occurring on fricatives and affricates.
 - c. Pharyngeal stop – posterior positioning of lingual base to pharyngeal wall, occurring on /k,g/.
 - d. Posterior nasal fricative – coarticulated nasal snort/flutter with any pressure consonant.
 - e. Mid-dorsum palatal stop – usually made in an approximate place of consonant /j/ in attempt to valve airflow.
5. Obturation of any hard palate fistulas may result in elimination of nasal leakage, improved resonance, and VP closure. Use of a speech bulb may be indicated for patients demonstrating reduced intraoral pressure, resulting in difficulty producing pressure consonants despite speech therapy.
6. Monitor phonation for vocal hoarseness, volume, and pitch levels with speech therapy for remediation or referral to otolaryngology.
- C. Preschool, School-Aged, and Adult: As speech articulation is acquired, the speech/language pathologist can begin differential diagnosis of velopharyngeal functioning.
1. Continued monitoring of hearing acuity.
 2. Speech disorders related to velopharyngeal function.
 - a. Hypernasality – the perception of excessive nasal resonance during production of vowels and semi-vowels resulting from inadequate separation of the oral and nasal cavities.
 - b. Hyponasality – reduction of normal nasal resonance usually resulting from blockage of nasal airway by various causes.
 - c. Mixed hyper/hypo – simultaneous occurrence in the same speaker, usually resulting from incomplete velopharyngeal closure and high nasal resistance that is not sufficient to block nasal resonance completely.
 - d. Cul-de-sac – variation of hyponasality associated with tight anterior nasal constriction, often resulting in muffled quality.
 - e. Nasal air emission – nasal escape associated with production of high oral pressure consonants. Occurs when air is forced through incompletely closed velopharyngeal port, and can be audible or visible (evidenced by mirror fogging, nasal grimace, and/or nasal flaring).
 - f. Compensatory articulations.
 - g. Reduced intraoral pressure – reduced build up of air in the oral cavity during production of pressure consonants due to inadequate valving of the VP mechanism.
 3. Phonation: Voice quality, the perceptual characteristics of voice.
 - a. Hoarseness – a periodic vibration of the vocal folds producing a “rough” vocal quality.

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- b. Breathiness – excessive leakage of air through the glottis during phonation.
- c. Pitch – sound property determined by the frequency of vibration of the vocal folds, either high, optimal, or low.
- d. Volume – acoustic power or intensity.
- 4. Assessment
 - a. Perceptual
 - i. Standardized articulation testing
 - ii. Assessment of perceived oral-nasal resonance balance during connected speech
 - b. Nasometer – nasalance, which provides a numeric output indicating the relative amount of nasal acoustic energy.
 - c. Aerodynamic Measurements – pressure flow studies estimating the sectional area of VP orifice (i.e., PERCI).
 - d. Assessment of oral structure and function.
 - i. Face: symmetrical structure and function; drooling
 - ii. Lips: degree of bilabial contact, non-speech function, position during quiet breathing
 - iii. Dentition: occlusion, crossbite, open/closed bite, over/underbite, ectopic teeth, missing, rotated, or supernumerary, dental arch collapse, dental appliances
 - iv. Tongue: deviation, lobule, frenulum, tongue thrust, non-speech function (range, strength, and symmetry of motion)
 - v. Hard palate: height, contour, width, oronasal fistulae
 - vi. Tonsils/faucial pillars: size, position, and symmetry of tonsils, movement of pillars
 - vii. Soft palate: symmetry at rest and during phonation; lateral and vertical degree of movement, uvula
 - viii. Submucous cleft palate: bifid/notched uvula, zona pellucidum or transparency of the palate at midline, bony notching at the posterior border of the hard palate
 - ix. Pharyngeal walls: vertical/lateral/symmetry of movement
 - e. Imaging Studies

If VP dysfunction is suspected, direct visualization is required to evaluate velopharyngeal functioning during speech production using oral and nasal consonants in words, phrases, and sentences.

 - i. Nasopharyngoscopy (videos: submucous cleft palate, velopharyngeal insufficiency; occult submucous cleft palate; sphincter flap; pharyngeal flap): degree of velopharyngeal closure for speech production and swallowing, velopharyngeal closure pattern, symmetry, velar contour,

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- movement (velum, lateral pharyngeal and posterior pharyngeal walls, Passavant's ridge), adenoids/tonsils, laryngeal structure, and function
- ii. Multiview videofluoroscopy
- f. A midsagittal lateral view: movement of the velum and posterior pharyngeal walls, height and length of velum, point of velar closure, and velar relationship to adenoids and posterior pharyngeal wall; posterior tongue valving
- g. Frontal view: lateral pharyngeal wall movement
- h. Basal/Towne's view: all of the above, except vertical movement

The speech/language pathologist reviews both perceived speech characteristics and physiological status of the velopharyngeal mechanism during speech production, with possible recommendations for surgical or prosthetic management, speech therapy, and/or continued monitoring of VP function. If surgical management is recommended, perceptual evaluation should occur 3-6 months following surgery, with repeat imaging studies 6-12 months post management. Speech therapy for VP function should be deferred for 6-12 weeks following secondary palatal management, while therapy for developmental or compensatory articulations may be resumed in 3-4 weeks.

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2. <http://www.geneticalliance.org/> Organization of parent support groups. A searchable catalogue for support group information on a variety of genetic conditions.
3. <http://www.nidcr.nih.gov/cranio/index.html> Educational information section of the National Institute of Dental and Craniofacial Research website.
4. <http://www.gene.ucl.ac.uk/nomenclature> Hugo gene nomenclature committee.
5. <http://www.ncbi.nlm.nih.gov/genome/guide> Human genome resources; genome guide.
6. <http://www.ornl.gov/hgmis/publicat/primer2001/index.html> *Genomics and its*

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