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The Consequences of Cranial-Facial Anomalies

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By

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ABSTRACT (100-200 WORDS):

Elissa Andalina
Honors Thesis Abstract

Everyday communication is essential. Not being able to communicate efficiently can pose many threats to a person with a communicative disorder. Throughout the process of my literature review, I have come to realize that the world of the speech-language pathologist is one of heterogeneity, and one must be able to expect the unexpected. As a future speech-language pathologist, I will be working with adults and children from diverse cultural backgrounds. To add to me knowledge as an undergraduate, I chose to research the topic of cranial-facial anomalies, because of my deep-rooted interest in helping children and adults. I chose to conduct a literature review, which entailed a great deal of research about the various aspects and consequences that children being born with a cranial-facial anomaly are faced with during development.

The sources of my research included websites from the Cleft Lip and Palate Association and the International Institute For Birth Defects, resourceful literature from books, and plentiful support and guidance from my advisor, Dr. Seaver. Dr. Seaver fostered my knowledge by offering me with his personal experiences and intellect as a professional and specialist within the area of cranial-facial anomalies.

The aspects in which I researched included speech and language development, embryology, surgical management, and various otologic and audiologic problems. The literature review also encompasses other various aspects, including dental and feeding complications, and educational and social issues.

Upon my completion ofthe literature review, I feel that I have gained a greater appreciation and understanding of individuals with communicative disorders. This literature review enabled me to see through the eyes of these individuals, and feel what they often times are experiencing. It is devastating when the family initially learns that their child has been born with a facial disfigurement. As I approach my entrance into the graduate program for speech-language pathology, I hope to utilize my newly learned knowledge regarding these very emotional communicative issues.

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The Consequences Of Cranial-Facial Anomalies

Individuals who are born with cranial-facial anomalies are quite special.. As a professional in the field of speech-language pathology and audiology, one of the most important goals is to greatly improve these individuals' perspectives about their lives, by not only offering support, but by providing them services which offer the greatest potential for maximizing their communicative capabilities. In this literature review, various aspects that affect the individual with a cranial-facial anomaly will be discussed. These various aspects include: defining what exactly a cranial-facial anomaly is, in addition to introducing cleft lip and cleft palate; the incidence of cleft lip and/or palate; etiology, which attempts to theorize on the possible causes of these congenital malformations; the surgical treatments of cranial-facial anomalies; team care; feeding and dental problems; otologic and audiologic problems; the embryology associated with the normal development of the facial and oral structures; the embryology associated with the formation of clefts; the prominent anatomical differences associated with cranial-facial anomalies, in addition to how these anatomical differences impact speech and language; the speech, language, and rehabilitative services that the speech-language pathologist and audiologist can provide; and prevalent issues regarding where these children with cranial-facial anomalies fit into the society. In addition to the discussion about children with cranial-facial anomalies, specifically cleft lip and cleft palate, three different syndromes where many children have significant otologic and audiologic problems will be discussed. These three syndromes are; Hemifacial Microsomia, , Mandibulofacial Dysostosis, and Nager Syndrome. Children with these syndromes are also likely to have some form of a cleft.

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Cranial-Facial Anomalies

There are many different types and forms of cleft lip and cleft palate, as well as many associated disorders that can co-occur. A cranial-facial anomaly can be defined as any congenital malformation that affects the facial and cranial regions, resulting in a mild to severe facial or cranial deformity. There are many different types and severities of cranial-facial anomalies, all having a potential impact on a child's normal development. The relative impact of the cranial-facial anomaly on the child's speech and language development is dependent on the severity, ranging from mild to moderate.

Cleft lip and cleft palate are two types of cranial-facial anomalies. What exactly is a cleft lip and a cleft palate? A "cleft" literally means a 'split' or a 'separation'. A cleft lip can occur as an isolated event, or can be found in conjunction with a cleft palate. A cleft lip is a condition that is characterized as creating an opening in the upper lip between the mouth and the nose. During the initial stages of embryologic development, distinct crucial and separate areas of the developing face fuse together. If any of these crucial structures involved is disrupted, there is a possibility that those structures will not fuse at the proper time and place, resulting in this 'split' or 'separation'. If these structures do not join correctly during the formation of the lip, the result is a cleft. Many embryologists and geneticists have considered

> "post-fusion rupture, as the pathogenic mechanism for the formation of some clefts, both of the primary and secondary palate. Some interference in the formation of the epithelial seam between the structures, or in the subsequent breakdown of that seam, and replacement by mesenchyme leads to later rupture or tears, perhaps as a consequence of continued growth of the embryo." (Sperber, 1989, p22).

When a cleft does occur, the skin, muscles, and oral lining are present, but have failed to fuse together, creating this opening known as a cleft. As mentioned previously, a cleft lip can range in severity from a slight notch in the philthrum of the lip, to a

complete separation unilaterally or bilaterally. A cleft on one side is called a unilateral cleft, and a cleft on both sides is a bilateral cleft. "A unilateral cleft of the upper lip, is the result of the medial nasal prominences failure to merge with the maxillary prominences on either side of midline." (Sperber, 2000, p.52).

Cleft lips occur more often on the left side than on the right side, for reasons which are still misunderstood. A cleft of the philthrum may even extend upwards into the nose. The philthrum in a normal child is the column which extends from the bottom of the nose, to the middle of the top lip. A cleft of the alveolar ridge or gum can also occur in association with a cleft lip. This separation of the gums also ranges from a small notch, to a complete division into separate parts.

More than half of all cleft lips co-occur with a cleft palate. A cleft palate occurs when there is a failure of the two sides of the roof of the mouth to fuse at midline, enabling the proper formation of a complete palate. The back of the palate, towards the throat, is called the soft palate. Clefts of the soft palate are also known as clefts of the secondary palate. The palate towards the front (mouth) is known as the hard palate. Clefts of the hard palate are known as clefts of the primary palate. Just like a cleft lip, a cleft palate can range from just an opening at the back of the soft palate, to a nearly complete separation of the roof of the mouth. Sometimes a cleft palate can include both the soft and hard palate, known as clefts of the secondary and primary palates.

> "Clefts of the primary palate appear if the lateral nasal processes, median nasal processes, and maxillary processes do not grow to sufficient size at the right time to meet each other, or if the fusion between the paired processes and associated structures is impaired." (Sperber, 1989, p.22).

Clefts are usually a result of developmental deviations, occurring during the fourth to eighth weeks of embryonic development, in addition to complications or disruptions during the beginning of the fetal period. Clefts of both the lip and the palate are more significant than clefts which occur in isolation, as well as those clefts

combining both the primary and secondary palates. It has become evident that those individuals born with a cleft palate, are more often times likely to have other associated anomalies, when compared to those individuals who were born with just a cleft lip.

Simultaneous differing clefts occurring in conjunction with one another, is often times the case due to certain predispositions to disruptions, or environmental toxins the child is exposed to during early embryologic development. Embryologists have theorized "that the cleft of the lip and the alveolus somehow predisposes the child towards a cleft of the secondary palate." (Sperber, 1989, p. 24). The theorists' theory states, "A lack of lip closure leads to overgrowth of the pro labial tissues, which divert the tongue up into the nasal cavity in such a way, that the tongue delays movement of one or both palatal shelves to the point that the optimal time for fusion is lost." (Sperber, 1989, p.24).

It is highly understood that for the normal formation of the lip and palate, one requires a fmely tuned and timed cooperating system. Sperber (1989) says, "the embryonic structures must be the right size, and in the right place at the right time, for normal development to occur." (p.24). Indeed, having more than one structure with a malformation poses many threats to the developing infant. These facial deformities can affect the child's weight gain, feeding patterns, and the proper functioning of the middle ear during infancy and early childhood. These middle ear dysfunctions pose as potential threats to chronic middle ear and sinus infections, abnormal speech and language development, , abnormal facial growth, and can predispose children to dental complications during their preteen and adolescent years.

Incidence of Cranial-Facial Anomalies

Now that the cranial-facial anomalies have been defined and discussed, the incidence of cleft lip and cleft palate will be introduced. Who is at risk for developing a cranial-facial anomaly? In general, individuals who are born with a cleft palate have a higher percentage of siblings with malformations than people with just cleft lip alone.

There are also gender differences that vary by the type of cleft the child has. According to Sperber (1989), females are found to have a higher incidence of palatal clefts occurring without any other anomaly, but are also found to have a higher incidence of associated anomalies when compared to males. Sperber (1989) indicates that for males, it has been discovered that they have a higher incidence of being born with both a cleft of the lip and the palate, which may contribute to them also being born with more severe clefts. "More than half of all clefts are accompanied by at least one minor or one major additional anomaly." (Peterson, Jones, Kamell, 2001, p9). According to the Cleft Lip and Palate Association's website (http://www.clapa.fsnet.co.uk/La2.htm). ... associated anomalies are found to occur with a force of 30 times greater in the cleft population compared to the non-cleft population. The surprising and disturbing fact, is that about 63% of the cleft population is also suffering from an associated anomaly. It has also been discovered that the incidence of associated anomalies manifesting during embryologic development also varies by the type of cleft. Sperber (1989) stated that those children who are born with a cleft alone, have a 7% chance of being born with an associated anomaly. Those children with both a cleft lip and palate have a greater chance of an associated anomaly, at 14%. Surprisingly, the greatest number of occurrences of an associated anomaly are those children with a cleft palate, being at 24%.

There are also differences seen in how the cleft has been manifested, which can be either unilateral or bilateral. For instance, a child born with a unilateral cleft will almost always be on the left side of the face, rather than the right side. In addition, a child who is born with a bilateral cleft lip may also be born with a cleft palate. To summarize, a unilateral cleft is most often manifested on the left side of the face, and palatal clefts occur more often when the child also has a bilateral cleft.

Embryologists have found that clefts of the lip are many times the precursors to clefts of the palate. There have also been studies that have found evidence of genetically

based familial tendencies. These studies have focused on the genetic predispostions and oversions which increase the likelihood of giving birth to a child born with a cleft.

Presently, there are more adults with cleft lips and/or palates who are having children, which may contribute to an increased risk of their children being born with some form of a cleft. Researchers have found that the risk of recurrence increases if more than one family member has a cleft. According to Peterson, Jones, and Kamell (2001), it has been estimated that around one out of four of all children born within the United States has a cleft lip. Mainly, 25% of children show a cleft lip, 25% represent a cleft palate, and 50% of the children born have both a cleft lip and palate.

Fortunately, in our day and age, we are receiving better medical treatment, resulting in improved diagnostics. There have been extensive research attempts to find the exact causes of these disturbances in the child's growth during embryologic development, which will be discussed in detail in subsequent pages of the literature review,

It is extremely important to point out that the cleft lip and palate population is extremely diverse, consisting of many racial and ethnic backgrounds. According to Seaver (200 I), Korean babies are found to have the highest incidence of being born with some form of a cleft; followed by Native Americans, Caucasians, other Asians, and the incidence among African Americans being the lowest.. Many scientists believe that genetics playa crucial role. "The majority of malformations are thought to result from a combination of genetic predispositions and adverse environmental factors operating early in life." (Drillien, Ingram, Wilkinson, 1966, p.2). Drillien, Ingram, and Wilkinson (1966) consider that "the basis for many of these malformations may be a multifactorial genetic system that predisposes the embryo to react in a particular way to minor variations in the intrauterine environment.". (p.2). But what is it about the embryo that makes it so sensitive to these minor environmental variations?

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Etiology of Cranial-Facial Anomalies

In this section of the literature review, the causes of these congenital cranial-facial anomalies will be discussed, in addition to touching upon what makes the embryo so sensitive to those minor environmental variations. These studies regarding the causes of congenital malformations, is defined as etiology. Geneticists and embryologists have long been debating over the exact causes of birth defects. According to Peterson, Jones, and Kamell, (2001), presently 20% of all birth defects are due to genetics; mainly by inheriting a single gene which predisposed the embryo to abnormalities, resulting in a complete disruption in development. Mainly 3-5% of birth defects are chromosomal, and a surprising 65% of all congenital defects are due to the environment.

There are also other factors that playa major role in birth defects. One extremely important factor to point out is maternal age. Older mothers, those in the latter part of their thirties, and into their early forties, have begun to undergo menopause. If the mother becomes pregnant during the initial period of menopause, the baby can have many potential problems. These older women who do become pregnant later in life, have a greater chance of giving birth to a baby with Down Syndrome, or many other anomalies of malformation. Younger mothers are also at a higher risk, for they may not be fully mature to give birth, both in the mental and physical realm.

Peterson, Jones, and Kamell, (2001), point out that there is also a slight increased risk of having a baby with a cleft if the father is younger than 20 years of age, or if the father is greater than forty years of age.

The environment also plays a crucial role in the development of an embryo. Seaver (2001) discusses that teratogens from the environment are known to be the cause of 65% of all birth defects. Unfortunately, most environmental teratogens are ingested by the mother herself. According to the website from the International Institute for Birth Defects (http://www.cleft.net, 2001), 30-40% of birth defects are the result from the mother smoking throughout her pregnancy, in which 13% of the babies born are

premature. There have also been new data discovered, directly relating smoking to clefting. Of course, and unfortunately, the drug list goes on- ethanol, barbiturates, amphetamines, and valium. Maternal diabetes and a vitamin deficiency may also contribute to birth defects.

Fortunately, there are detection methods that are available for these mothers. One detection method which is highly used is known as amniocentesis. This test is usually performed between the 14th and 17th week of pregnancy, checking for abnormal or absent chromosomes. This method is very low risk to the mother, with the results being provided within two to six weeks post exam. Another detection method is known as Chronic Villus Biopsy (CVB). This method provides data about the embryo's risk to limb malformations, and to the risk of cleft lip and palate. This is why it is so extremely important for mothers to eat healthy, and to remain drug free, for the health of the baby and herself.

Surgical Treatment

When is a cleft lip repaired? A cleft lip is usually repaired with surgery by the time the child is two to three months of age. Timing is critical. It is highly stressed that the earlier the child gets the surgery, the less impact the cleft will have on the child's speech and language development. . Peterson, Jones, and Kamell (2001) report that the requirements for surgery are a general anesthetic, around one and a half hours time, and stress that a successful outcome will require a minimum of around four to five surgical procedures. A plastic surgeon specializing in the reconstruction of the face, due to congenital anomalies, working on a cleft lip and palate team, is referred.

> In fact, "the first surgeons to operate on children suffering from clefts of the lip or palate were only concerned with the correction of the deformity, rather than the improvement of their patient's speech. Today, the surgeons are now concerned about the effects the operation may have on the child's

> speech,, in addition to producing a palate which is capable of closing off the naso-pharynx, since excessive nasal escape is responsible for much of the speech disability suffered by patients both before and after operation." (Peterson, Jones, Kamell, 2001, p.155).

The plastic surgeon re-arranges the skin and muscles of the lip as much as he/she can, so that no other skin grafting from other parts of the body is needed. One example of a bone grafting technique, is known as alveolar bone grafting. The plastic surgeon harvests bone from other areas of the body to fill in the hole created by the cleft. "The surgical procedures are designed to create a lip and nose that are esthetically pleasing and functional, an intact palate, and an intact alveolar ridge that provides good bony support for the lip and nasal base." (Peterson,, Jones, Kamell, 2001, p. 87). The goal of primary lip reconstructive surgery for babies with a cleft lip, is to accurately create a symmetric union between the membranes of the lip, with minimal scarring.

An example of a lip surgery technique is known as Cheiloplasty. After surgery, the child is put into arm restraints to prevent them from tearing out the stitches. Modification in the child's feeding environment is managed by the nurses on the cleft team. The baby also undergoes wound care, to foster quicker healing of the scar. With any surgery, there are always potential complications. Peterson, Jones, and Kamell (2001), emphasize that with lip repair, common complications include: pneumonia, , break or tear in the stitches, hemorrhage, diarrhea,, otitis media (ear infection), and the risk of infection of 13%.

The reconstruction of the palate, also known as palatoplasty, can serve as a great benefit for the child. However, there are three variables that must be taken into consideration, , when discussing the potential problems that the effects of palatoplasty can have to the well being of the ear.

> The three variables are, "the variation in the type of palatoplasty carried out, the variation in the age at which surgical repair was performed, as

> well as the amount of time elapsed between palatoplasty, and follow-up for otologic and audiologic testing, and the inconsistency in whether, and when, tubes were placed in the ears." (peterson, Jones, KameU, 2001 p.155).

It is critical that surgery be performed as early as possible for the repair of the palate, and it is crucial for the parents to be educated about their child's condition, so that the child can receive the necessary treatment for normal speech and language development..

The main goal of primary palate repair for babies with a cleft palate is for proper and adequate closure of the palatal shelves for normal speech and language development.. However, when everything is healed, the palate may still have some scar tissue, contributing to poor mobility of the palate. Other important goals of primary palatal surgery, are for the baby to be able to intake the proper amount of food for growth, and to improve the overall functioning and well being of the middle ear cavity. Primary palatal repair should be done when the baby is between 12 to 18 months of age. Again,, the sooner the surgery is performed, the better off it is felt that the child's speech and language development will be.

After the operation, the child is put into arm restraints to prevent any unwanted finger interference. The baby is fed liquids, and is usually sedated. Fortunately, most babies experience minimal pain,, and go through a quick recovery. Medication is provided for any discomfort the baby may be experiencing. The parents are encouraged to stay in the hospital with their child, for three to five days is usually needed for the child's recovery. Feeding after surgery is usually not a problem, because the nurse and speech-language pathologist will be there to answer any questions regarding any issue. In a recent study, researchers have concluded that "breast milk helps to decrease the incidence of otitis media (ear infection), in infants with clefts. (peterson,, Jones, Kamell, 2001, p. 152).

As briefly mentioned earlier, those children who had undergone primary lip and palatal surgery, usually receive an additional secondary, surgical repair. The plastic surgeon works to improve the appearance of the nose and the lip and/or palate, including reducing the presence of any scars. This is usually done between the child's third and fifth birthday. This secondary, surgery is usually highly recommended before children start school.

For secondary, palatal repair, the plastic surgeon will again make any minor adjustments, including the correction of any maxillary (upper jaw) and/or mandibular (lower jaw) problems. Many times, children born with a cleft palate have a smaller lower jaw (mandible), which may also disrupt the ease of the child's breathing. The surgeon will also work to correct the function of the velopharyngeal mechanism. The goal in correcting the functioning of the velopharyngeal mechanism, is to improve the child's speech by reducing the amount of air and sound going into the nose. For children with clefts in their gums and/or alveolar ridge, an additional surgery may be performed for additional bone grafting along the gum line, to promote the movement of teeth into their proper positions.

Team Care

Babies who are born with any type and severity of cleft(s), and/or with other associated anomalies, will be provided with service and treatment from a Cleft Lip and Palate Team. This special team is comprised of specialists working together to make sure optimum treatment is being provided to both the child and his or her family. A cleft team can be comprised of surgeons, cleft nurses, an orthodontist, a pediatrician, , a speech-language pathologist, an audiologist, a psychologist, and a geneticist. Preferably, a member of the Cleft Lip and Palate Team will contact the family within twenty-four hours post-diagnosis of either a cleft lip and/or palate.

The surgeon is responsible for repairing the child's anomaly, before it poses potential threats to the child's speech and language development. It is the job of the speech-language pathologist to make any necessary speech and/or language modifications necessary, depending upon the severity of the child's anomaly. The audiologist will conduct audiologic testing, to rule out the possibility of a hearing loss. The child's pediatrician will provide the other members of the team with important medical information, and the psychologist will serve as a counselor and friend to the child and his or her family.

Feeding Problems

Having being born with an anomaly of the mouth and/or palate may also pose potential threats to the normal feeding of the infant. This section of the literature review will be touching upon the problems associated with feeding, that babies born with a cleft lip and/or palate may be faced with.

Although a cleft lip may sometimes get in the way, babies with a cleft lip usually do not have any feeding difficulties. Normal breast or bottle feeding is possible. The nurse will properly show the mother how to position their infant with their nipple.

On the other hand, a newborn baby with a cleft palate may need extra help. Depending upon the severity of this obstruction in the baby's mouth, it's presence can make it difficult or even impossible for the baby to form sufficient suction with the structures in their mouths, needed for successful feeding.

As newborns, these babies with a cleft lip and palate are already faced with anatomical differences that make it difficult to feed, and if you add a cleft lip or palate into the picture, it makes it even more difficult for the infant. Anatomical differences which exist in newborns making it more difficult to feed, include having a larger tongue which is housed within an oral cavity that is extremely small, and a larynx which is situated in a more elevated position, with an enlarged epiglottis.

The most prominent problems that these babies are faced with, is that feeding may be extraordinarily slow. Cleft babies may also take in too much air while feeding, or they may even bring up milk into the nose. Although breast feeding is possible, bottle feeding is usually most successful for babies with cleft palates. The team assisting the cleft lip and/or palate babies and their families, include the baby's pediatrician, the nursing staff, and the speech-language pathologist..

Dental Problems

The cleft population may experience more dental occlusion problems compared to children in the non-cleft population. The normal development of some of the child's teeth may be adversely affected by clefts, because of the simultaneous growth of both the child's teeth and jaw during the time the cleft occurs.

The severity of the dental malformation is dependent upon the cleft. Typically, teeth of children with clefts come through later than children born without clefts. In some cases where the gum is disrupted by the cleft, some teeth may be missing or extra may develop. There may be a dental malformation(s) which prohibit the top and bottom jaws to meet properly. It is not necessary for children with their baby teeth to go and see an orthodontist, rather they should be seeing their dentist for regular cleanings just like normal children. Not until the permanent teeth come, will a brace be put on to correct the irregularity .

Oral hygiene is one major concern for these children, for anatomical differences may prohibit the child from cleaning their teeth and mouths efficiently.

Otologic! Audiologic Problems

This section of the literature review will be introducing the various otologic and audiologic problems that are typically seen in the cleft population.

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It is a fact that otitis media is more common in children with cleft lip and cleft palate. Children who are born with a cleft palate may sometimes require visits to an audiologist, due to hearing problems as a result of chronic otitis media (middle ear infections). Chronic otitis media creates persistent fluid and pressure within the middle ear cavity, and can be very painful for the child causing extreme discomfort. In cases where medication has no affects, surgery to the ear may be recommended. Ear surgery involves inserting a grommet tube into the tympanic membrane (ear drum), to allow for aeration of the middle ear. After surgery, regular hearing evaluations should be arranged to monitor the child's hearing, as it is necessary for normal speech and language development. .

> "Ear disease and hearing loss constitute a major potential threat to communication in individuals with cleft palate, and many of the multi-anomaly cranial-facial disorders. More than 90% of infants with cleft palate are born with fluid already present in the middle ear cavity, and youngsters with clefts are vulnerable to middle ear disease in their teenage years and into adulthood." (Peterson, Jones, Karnell, 2001, p.148).

There are also many anatomical differences which contribute to the otologic and audio logic problems seen in the cleft population. One of the most prominent anatomical differences, is that specific tensor fibers of the hard palate are missing their 'anchor' or necessary attachments. This means that the tensor veli palatini muscle, which is responsible for the contraction of the soft palate and opening of the eustachian tube, cannot efficiently contract to open the Eustachian tube. In an infant, the Eustachian tube is positioned in a horizontal position, and as the child matures, it assumes a more tilted position. This horizontal orientation of the Eustachian tube in a neonate brings upon an even greater disadvantage, for the tensor veli palatini muscle has a decreased ability of

opening the Eustachian tube. If the Eustachian Tube cannot open properly for aeration, an excess amount ofvelopharyngeal bacteria is free to enter into the middle ear cavity.

> "The abnormal reflux of food and fluid into the nasal cavity, can set up chronic inflammatory changes about the Eustachian orifices, resulting in low grade infections, and possibly even secondary middle ear disease. The break in the mechanical barrier between the mouth and the nasal pharynx, with it's concomitant chronic inflammatory change, alters the bacterial flora of the region, permitting an overgrowth of predominantly pathogenic bacteria, resulting in otitis media" (Peterson, Jones, Kamell, 2001, p. 151).

Another problem that occurs when the Eustachian tube cannot open efficiently, is that negative pressure develops within the middle ear cavity, pulling tightly on the tympanic membrane.

> "The negative middle ear pressure also draws bacteria up through the tube. Eustachian tube obstruction is seen as the primary cause for the ear disease in children with clefts. However, abnormal compliance, or 'floppiness' of the tube is also a factor, possibly due to the quality of the cartilage, muscular dysfunction, and other factors." (Peterson, Jones, Kamell, 2001, p.150).

What is the impact that these anatomical complications pose to the degree of hearing loss experienced by these children with clefts? If these problems caused by chronic otitis media continue to persist, the child may suffer from extreme pain, and may cause permanent damage to the structures of the middle and inner ears, leading to permanent hearing loss. Most cases of cleft lip and palate are associated with a mild to moderate, bilateral conductive hearing loss. It is extremely important for the parent or caregiver to bring their child to an audiologist to monitor the child's hearing while the problem continues to persist. An audiologist can provide the necessary amplification

needed for normal speech and language development. . A referral to an ENT or "Ear Nose and Throat" doctor may be recommended, for they can prescribe medications for chronic otitis media,, or insert PE (pressure equalization) tubes into the middle ear for proper functioning. If the child's hearing mechanism is impaired in any way, the child's ability to understand and communicate with people and things in their environment is diminished. Researchers have found that children who had suffered from chronic middle ear infections, experienced the passing of harmful toxins into the anatomical structures of the inner ear, causing detrimental deformities, and contributing to sensori-neural hearing losses.

What are some variables that affect the prevalence of ear disease? Paradise, Bluestone, and Felder (1969), found that such variables as the type of cleft,, and the child's age were predictors of middle ear disease. Paradise, Bluestone, and Felder (1969), found that the "prevalence of otitis media was 96% in infants with cleft lip and cleft palate, or cleft palate only, compared with only 22% in babies with cleft lip only, and 19.5% in non-cleft babies." (p.154). As one can see, Paradise, Bluestone, and Felder's (1969) fmdings, suggest that babies born with some form of a cleft suffer a great deal more with otitis media,, when compared to the babies representing the non-cleft population.

Another important finding, is that neonates are more susceptible to chronic middle ear infections than older children. This suggests that as children get older, the risk for otitis media decreases significantly. Also, as the child gets older, the Eustachian tube moves from a more horizontal position, to a more vertical position. This vertical positioning improves the muscle and cartilage support of the tensor veli palatini muscle. (the major muscle responsible for opening the Euatachian Tube).

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Embryology

The anatomical structures that are associated with oro-facial development are extremely diverse, when compared to other regions of the body. The way in which the structures of the cranium and face develop is extremely complex, and requires an in-depth embryological description. In this section, the embryology of the facial and oral structures will be discussed. This discussion will take an in-depth look at the development of the palate and the ear, and how disruptions in their formation can cause various forms of clefts, and associated anomalies involving various structures of the ears. Sperber (1992, pl l l) stated, "These events of the first few weeks of intrauterine life, are the most important occurrences predicting one's entire extra-uterine life."

The development and health of the embryo is dependent upon a number of factors. These factors include genetic predispositions, such as an inherited genotype, and how these genetic mechanisms are able to function; environmental factors, such as nutrition and biochemical interactions, as well as temperature and pressure; and finally, functional forces, both intrinsic and extrinsic, acting upon muscles and organs.

To begin, it is extremely important to define two terms that are used to denote different modes of the joining or blending of the facial and palatal processes to form normal structures: they are fusion and merging.

> "The paired facial and palatal processes, as they grow towards their counterparts, have an epithelial covering that adheres to the covering of the counterpart process. Normal formation of the structure then depends on breakdown of that epithelial seam, and subsequent mesenchymal consolidation. This is the process known as fusion." (peterson, , Jones, Kamell, 2001, p.19).

An insufficient breakdown of this epithelium outer shell has been hypothesized to be one cause of the development of clefts. "Merging, is defined as the coalescence of growth centers in tissues where there is already a confluence of the mesenchyme that

seems to smooth out grooves in the epithelium." (Peterson, Jones, Kamell, 2001, p.19).

The formation of the face is very complex, and can be divided into developmental time periods. The face is derived from five prominences surrounding a shallow depression known as the stomodeum, which constitutes as the future mouth. These five prominences are derived from a neural region containing neural tube and crest cells, which move from a more posterior position,, to a more superior position, up to the area of the developing facial and neck regions. The five prominences involved in the formation of the facial region are; the single median front-nasal prominence, the paired maxillary and mandibular prominences, and derivatives of the first pair of branchial arches. The stomodeal chamber is responsible for dividing into the oral and the nasal cavities, when the fronto-nasal, and maxillary prominences begin to develop into more of a horizontal positioning within the chamber.

The union of these facial prominences is the result of either one of two developmental events. "1) By the merging of the fronto-nasal prominence with the maxillary and mandibular prominences, or 2) by fusion of the central maxillo-nasal components." (Sperber, 1989, p 36). In order for the prominences to merge, intervening shallow grooves proliferate, as the result of the movement of underlying mesenchyme within the groove. Successful fusion of these prominences, requires that the contacting surface epithelium disintegrate, allowing for the underlying mesenchymal cells to intermingle with one another.

The fusion of the maxillary and median nasal prominences are responsible for providing continuity, and structure to the upper lip and jaw. When there is a failure of the surface epithelium to disintegrate, the mesenchymal cells do not intermix, preventing the maxillary and median nasal mesenchyme to merge. This failure is one of the causes of an upper lip cleft and/or cleft of the anterior palate.

The fusion of the median nasal processes, the lateral nasal processes, and the paired maxillary prominences, produces the embryonic primary (anteriorlhard) palate, in

addition to giving form to most of the upper lip. The formation of the secondary palate (soft and more posterior), is formed by the meeting and fusion of the palatine processes, commonly known as the palatal shelves. These palatal shelves also fuse with the primary palate (more anterior palate) along the premaxillary suture.

As the underlying mesenchymal cells intermix with one another, the first parts of the face to become visually recognizable are the lower jaw and upper lip, which are formed by the merging of the paired mandibular prominences. In addition, the lateral merging of the maxillary and mandibular prominences creates the comers of the mouth.

Around the seventh week of embryonic development, there is a shift in the blood supply, from a more internal position, to a more peripheral position, creating a crucial time period for the proper development of midface and palatal development. During this time, there is the potential for the blood supply to become inadequate, which will consequently lead to defects to the upper lip and palate.

Backing up one week to around the sixth week post-conception, the fusion of the median nasal, lateral nasal, and maxillary prominences creates the embryonic primary palate, in addition to constituting for most of the upper lip, a portion of the alveolar ridge, and the anterior portion of the maxilla. The premaxilla, or upper jaw, is made up of portions from the alveolar ridge and the hard palate. During the development of the primary palate, the nasal septum begins to elongate downwards, where it will eventually fuse with the palate, and separate the two nostrils. The secondary palate is formed by the fusion of medial projections from the paired maxillary processes, (known as the palatine processes or shelves). These palatal shelves will also fuse with the primary palate.

> "The palatal shelves initially develop as two vertical floppy shelves, hanging down on either side of the rapidly growing embryonic tongue. The shelves get into a horizontal position above the tongue, adhere to each other, and fuse. The shelves move upward very quickly, with the posterior portions moving upwards first. Fusion between the shelves takes place in

> the opposite direction- from front to back.. Shelf contact in the region of the soft palate seems to involve remodeling oftissues." (Peterson, Jones, Karnell, 2001, p.19).

The completion of the development of the secondary palate occurs between the tenth and twelfth weeks post-conception.

> "The process of shelf elevation and fusion takes place about one week later in girls than in boys- which may help to explain why girls are more vulnerable than boys to clefts of the secondary palate. The formation of the secondary palate takes more time in girls, providing more of a time window for adverse effects from environmental agents. (Peterson, Jones, Karnell, 2001, p.19).

The embryonic development of the facial and cranial regions can be broken down into phases, and it is crucial to explain how and what occurs along this path of development. There are three phases associated with embryologic development. Although all phases of embryologic development are important, starting the discussion at the second phase of development, the Embryonic period, is where the face and the palate begin their formations.

As stated previously, the second phase of development is termed the Embryonic period, beginning from the end of the first week, and lasting until the eighth week. This period of development can be subdivided into three periods. The first period is the Presomite period, having a time frame of eight to 21 days post-conception. During the third week of development (21 days), also known as the flexion stage, the formation of the face, head, neck, and oral cavity begins. The normal development of the head is dependent upon the proper communication of activities and information through the prosencephalic (forebrain), and rhombencephalic organizing centres. The prosencephalic centre, located beneath the forebrain, is "responsible for inducing the visual and inner ear apparatus, and upper 1/3 of the face. The rhombencephalic centre is responsible for

inducing the middle and lower 2/3 of the face, subsequently inducing the formation of the middle and external ears." (Sperber, 1989, p 31).

Associated with the development of these two important centres, is the division of the forebrain into bilateral (paired) hemispheres, contributing to the paring of the olfactory bulbs and the optic nerves. "Failure of these cerebral divisions has a profound influence on facial development, leading to many types of anomalies." (Sperber, 1989, p. 31). Also during this period, the primary layers of the embryo and fetal membranes are formed into an inner cell mass, giving rise to the ectoderm, mesoderm, and the endoderm. The ectoderm consists of the epithelium enamel or lining, hair, eye lens, the inner ear, and the central and peripheral nervous systems. The mesoderm is responsible for connective tissue, blood vessels, and the heart. The endoderm houses the digestive tract, the liver, the middle ear, and is responsible for respiratory function. A membrane, known as the oropharyngeal membrane, is formed by an endodermal thickening, and is the site at which the stomodeum, or primitive mouth is formed. The oropharyngeal membrane also serves as a junction between the "ectoderm that forms the mucosa of the mouth, and the endoderm that forms the mucosa of the pharynx." (Sperber, 1989, p.32). The oropharyngeal membrane sets a limit in regards to the depth the primitive stomodeum can form, only allowing for this complex structure to create a wide midline shallow depression. This depression formed by the oropharyngeal membrane enables the five prominences to grow ventrally to form a deep oral cavity.

The second period, known as the Somite period, lasts from 21 to 31 days, and is characterized by the appearance of the body's main systems and organs. It is extremely important to introduce a system with a number of functions, called the branchial apparatus. At the onset of the fourth week, two pairs of structures on each side of the head and neck appear. These bilateral paired structures will give rise to the development of the branchial apparatus, which is a system with a number of structures being responsible for the development of much of the lower facial regions. This branchial

apparatus is responsible for the development of the ears, maxilla, mandible, and anterior portion of the neck. "The branchial arches are formed by the segmentation of the mesoderm's lateral plate, becoming five distinct bilateral mesenchyme swellings, the branchial arches." (Sperber p., 1989, p.58). The branchial arches are positioned on the external portion of the embryo, corresponding to five internal landmarks on the elongated pharynx, known as the pharyngeal arches.

The first pair of branchial arches are known as the mandibular arches, and consist of a lower mandibular process and an upper maxillary process, which give rise to both the upper jaw (maxilla) and lower jaw (mandible). It is on the 22nd day of development, that the first pair of branchial arches are separated by a midline depression, known as the stomodeum or (primitive mouth).

The second branchial arch forms the basis for the smallest ossicle of the middle ear, known as the stapes. The second pair of branchial arches also contributes to the formation of the other two bones of the middle ear, the malleus and the incus. Also during this time of development of the second branchial arch, is the formation of a cartilaginous mass known as the hyoid arch, located in the region of the neck.

The third branchial arch is responsible for continuing with the development of the cartilaginous mass of the hyoid arch, and producing "the greater hom of the body of the hyoid bone. The mucosa of the posterior third of the tongue is also derived from this arch, which accounts for its sensory innervation by the glossopharyngeal nerve." (Sperber, 1989, P 63).

The fourth branchial arch is responsible for the formation of thyroid cartilage. During this stage, other important muscles are formed, including the cricothyroid, the palatopharyngeous and uvular muscles of the soft palate, and the palatoglossos muscle of the tongue.

The fifth branchial arch is a structure that literally disappears as soon as it is formed, and doesn't really contribute to the formation of any major facial or cranial

structures. Later development of this period will contribute to the formation of the cricoid and arytenoid cartilages of the larynx.

Each branchial arch, excluding the fifth, "consists of an artery, a cartilaginous bar, a muscle element, and a nerve going into the brain." (Peterson, Jones, Kamell, 2001, p.17). Any deficiency or disruption during the development of the branchial arches, can result in a number of specific malformations or syndromes, depending upon where the disruption in development took place.

In addition to the formation of the branchial apparatus, two other important processes are beginning to form. They are the frontonasal and maxillary processes, in addition to the nasal (olfactory) placodes. One last characteristic associated with the Somite period, is that web-like buds of limbs begin to develop, that will eventually begin to differentiate.

The third period of embryologic development is known as the Postsomite period, and lasts from the 32nd day until the 56th day. The Postsomite period is characterized by the formation of the body's external framework and features. Around the fifth week post-conception, a number of structures accelerate in their growth and development. . These structures are known as the fronto-nasal processes, the maxillary processes, and the mandibular and hyoid arches. Each of these structures are extremely important for the development of the facial region.

The frontonasal processes are responsible for the formation of the lenses for the eyes, and for the olfactory placodes. The paired maxillary processes, which are situated on both sides of the beginning facial region, contribute to the structure of the face. The mandibular arch is representative of fusion, for it forms by a constriction down midline. The hyoid arch is characterized by it's two lateral protrusions, which are also joined down midline.

Due to the extent of the rapid oro-facial development at the cranial end of the embryo, the head is responsible for nearly half of the total body size of the developing

embryo. The embryo's total crown to rump length around the fifth week is five to six millimeters. At the beginning of the sixth week, the embryo's length continues to grow, to around ten to twelve millimeters.

During this sixth week of development, nasal pits begin to divide the paired frontal processes into two separate entities. The initial, and most rapid growing divided and new process, is the medial nasal process. The medial nasal process is responsible for forming the median wall of the nasal pit. The other process to form out of the separation of the paired frontonasal processes, is the lateral nasal processes, which constitutes the lateral walls of the nasal pits. Upon the completion of this separation of the paired fronto-nasal processes into the medial and lateral nasal processes, the eyes of the embryo can now be observed in a more lateral position. In fact, the eyes of the embryo can be described as well-defined bulges.

The maxillary processes, which are wedge shaped just below the eyes, become more prominent as they continue to grow towards midline. The enlargement of the mandibular arch gives rise to the external auditory meatus, via tiny grooves that are associated with it's most lateral portions. The only distinguishing evidence of the hyoid arch are the remains of three auricular hillocks.

In the middle of the sixth week,, there is a broadening of the embryo's head, due to the movement of the nasal pits, and closer approximation of the lateral and medial nasal processes. The paired maxillary processes are continually moving towards midline, where they rest against the limbs of the median nasal processes. At this point in development, the mouth's opening is quite large. The continual fusion between the maxillary and mandibular processes help to move the lateral angles of the mouth into a more medial and closed position. The auricular hillocks, which were the remains of the developing hyoid arch,, become more prominent during the middle of the sixth week. The auricular hillocks, serve to create the formation of the pinna,, or outer ear.

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At the beginning of the seventh week, the embryo's crown to rump length is around 17 to 18 millimeters. As one can see, there is rapid growth and development during these early stages of the embryo's life. As the edges of the median and lateral nasal processes fuse, the nasal pits are being transformed into nostrils. The upper lip is also being formed by the merging of the median nasal processes, in addition to the fusion of the maxillary and median nasal processes. The fusion process of the upper lip is characterized by moving from a more superior position, to a more inferior position, where the lateral angles of the mouth move into a more medial and closed position. The mouth's opening also becomes substantially smaller, due to the meeting and fusion of the maxillary and median nasal processes at midline.

Another structure known as the nasal septum, works to divide the nostrils, which still contains remnants from the frontonasal processes. Due to the continued enlargement and growth of the head, but lack in the vertical growth of the face, the eyes move into a more anterior position, but remain at the level of the nose.

The beginning of the seventh week is also characterized by rapid growth of the internal oral structures. This rapid growth of the oral structures cause the ears to remain very low set, when compared to how they will be later in development. . The ears are characterized as being in a more posterior and low set position. Also during this seventh week of embryologic development, the maxillary processes fuse with the mandibular arch, creating rounded bulges forming the cheeks. The auricular hillocks of the hyoid arch meet, and begin to create a structure characterized as an incomplete ring around the external auditory meatus. The seventh week is also important because of it's role in palatal development. . The formation of the premaxilla is completed as the result of the merging of the median nasal processes. Another set of processes known as the Palatine Processes, are derived from the maxillary processes, and will give rise to the formation of the secondary palate.

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The final phase of embryonic development is known as the Fetal Period, and lasts from the eighth week until term. This phase can be identified by the first feelings of movement by the fetus. The fetal period is characterized by rapid growth and expansion of the structures already formed, in addition to the formation of fluid-filled cavities and membranes which physically protect the fetus, and provide for nutrition and waste disposal services. As the embryo begins to differentiate and develop into a more complex organism, a great deal of expansion occurs at the cranial end. In addition to the embryo's rapid growth and development associated with the cranium, the embryo is growing at an even greater rate at it's core, when compared to it's periphery.

The ninth week of development is characterized by having a growth spurt in the mandible, causing the tongue to drop between the palatal shelves. Due to the new positioning of the tongue, the direction of growth of the palatal shelves begins to move into a more medial position. Elevation of the palatal shelves enables the fusion process to begin. Fusion of the palatal shelves is characterized as going from an anterior to posterior position. The hard palate should be fully completed between the eighth to ninth weeks of development. It is not until around the twelfth week of development, where the soft palate and uvula will be fully developed. As one can see, the development associated with facial and palatal development is extremely complex, and requires the precise coordination and timing of crucial processes and their associated structures.

The development of the ear is also very complex and unique, and disruptions in it's development can be associated with a number of various audiologic problems in children with cleft lip and/or palate. It is important to discuss the anatomy of the ear, so that it can be compared to ear malformations later on in the literature review.

The ear is divided into three different areas; the outer ear (pinna), the middle ear, and the inner ear. The ear serves as a transducer, that changes energy from acoustical energy (sound), into electrochemical energy, (what the brain understands). The outer ear is collectively called the auricle, or pinna,, and is responsible for collecting and localizing

sounds. The pinna is the part of the ear that we can see, and is visually recognizable by cartilage covered by fat and skin. The surface of the auricle is uneven, and filled with pits, grooves, and depressions. The deepest of these depressions is called the concha, while the rim of the ear is called the helix. The helix forms the upper part of the pinna, all the way down to the ear lobe. Another part of the outer ear, known as the anti-helix, is the fold marking the entrance to the concha, as it follows the shape of the helix. The opening in the pinna is called the external auditory meatus, or ear canal. It's function is to conduct the sound waves into the tympanic membrane, or ear drum.

The ear canal is an irregular, curved, or S-shaped tube, about 25 millimeters long and eight millimeters wide. The course, or direction of the meatus is directed slightly downward, protecting against the invasion of bugs and water. The diameter of the external auditory meatus is largest at the external orifice, (beginning), and becomes gradually smaller towards the isthmus (ending). The isthmus is the site of junction between the cartilaginous framework with the bony framework. The lining of skin of the external auditory meatus connects with the tympanic membrane, where it becomes the outer layer of the eardrum. Inside of the external auditory meatus are hair cells known as cilia. Also found within the external auditory meatus is cerumen, or ear wax, which helps to protect the meautus' moisture from drying out, and is noxious to pesty bugs.

Embryologically, the external ear is formed from tissues from the first and second pharyngeal arches. Auricular hillocks form from the remnants of the hyoid arch, and become apparent as six tissue elevations. Three hillocks are the by-product from the first pharyngeal arch, and three form from the second pharyngeal arch. Each of the six auricular hillocks are responsible for carving out a distinctive portion of the external ears.

During early embryologic development, the external ears are positioned in a more inferior position relative to the lower jaw. It is not until the initial growth of the lower jaw, that the external ears begin to move into a more superior and more vertical position.

The middle ear begins with the tympanic membrane. The middle ear is an

air-filled cavity which contains the three smallest bones of the entire body; the auditory ossicles: the malleus, incus, and the stapes. The tympanic membrane reaches it's full size in utero, usually having a diameter of ten millimeters. It has a characteristic oval shape, and sits at a 55 degree angle in relation to the external auditory meatus. A ring of fibrocartilage forms a ring around the tympanic membrane, known as the annulus, which fits into a groove known as the tympanic sulcus.

The tympanic membrane has three distinct layers. The outer layer is continuous with the external auditory meatus' epithelial lining. The middle layer is fibrous, and is responsible for the compliance of the tympanic membrane. The inner layer is continuous with the mucosa lining found in the inner ear. There are also two distinct areas associated with the tympanic membrane; Pars Flaccida and Pars Tensa. Pars Flaccida is a small triangular shaped area situated at the top that is floppy. Pars Tensa is the remainder portion of the tympanic membrane which holds it tense. In conjunction with these anatomical portions of the middle ear, it's health can be determined by the appearance of the "Cone of Light". When the tympanic membrane is healthy, it is concave, smooth, translucent, and has a pearl-like gray color to it. Light shone into the ear, resulting in a wedge-shaped reflected area of light towards the bottom, indicates a healthy eardrum. Audiologists and pediatricians will use an otoscope to look into the ears of children with cleft lip and palates. If the "Cone of Light" is not apparent, the child is probably suffering from otitis media (middle ear infection).

Inside of the middle ear cavity is a six-sided structure housed inside the petrous portion of the temporal bone. The tympanic membrane is responsible for making up the entire portion of the lateral wall. The legmental wall is a paper thin piece of bone making up the roof of the middle ear cavity, and is responsible for separating the tympanic cavity from the cranium. The floor of the middle ear cavity is made up of a tympanic plate, separating the tympanic cavity from a deep groove which houses an artery. The medial

wall of this cavity gives rise to the lateral wall of the inner ear. This cavity houses the both the round and oval windows, providing connections into the inner ear.

The three bones of the middle ear are collectively called the ossicular chain. This chain of bones transmits acoustic energy from the tympanic membrane into the fluid of the inner ear. The first bone is the malleus, and is the largest of the three. The second bone is the incus, and moves in synchrony with the malleus. The third bone, which is the smallest of the three, is called the stapes. The footplate of the stapes rests in the oval window of the inner ear. There are also various muscles in the middle ear, aiding in the function of the ossicular chain. Also found in the middle ear cavity, and extremely important, is the Eustachian tube. The Eustachian tube is responsible for providing aeration of the middle ear, and permitting drainage of normal ear secretions.

During embryologic development, , as the inner ear begins to differentiate, the middle ear begins to form. The lining of the middle ear cavity and auditory tube forms from the endoderm of the first pharyngeal pouch. The ossicles of the middle ear are formed out of cartilages from the first and second pharyngeal arches.

The function of the inner ear serves as a balance center, sensing movement of our body in space through the movement of hair cells suspended in fluid in the semicircular canals. The inner ear also serves as our hearing center, because of a structure known as the cochlea. The cochlea is a bony canal which looks like a snail, because of it's coiled framework. . It is coiled around a central core called the modiolus. The internal auditory meatus is situated at the bottom of the modiolus, where the eighth cranial nerve passes through on it's way up towards the brain. The cochlea is filled with two different types of fluid, endolymph and perilymph.

> "An out growth of a saccule forms the cochlear duct. Tall, columnar epithelial cells of the growing cochlear duct becomes the organ of corti (our hearing organ). An otic pit is formed behind the second pharyngeal arch. Both otic pits are in relative proximity to the hindbrain portion of

> the neural tube. The otic pit deepens and pinches off from the surface. Below the second pharyngeal arch is a newly formed otic vesicle. The stato (vestibulo) acoustic ganglion begins to form between the otic vesicle and the neural tube. Cells that are leaving the otic epithelium are forming the neurons of the eight cranial nerve. Differentiation of the otic vesicle yields three major subdivisions of the inner ear: the endolymphatic sac and duct, and the utricular and saccular portions. The ectoderm of the embryo becomes the epithelium of the inner ear." (http://www.med.unc.eduJ,, 2001).

Another structure associated with the cochlea is known as the basilar membrane. On the basilar membrane is the organ of corti, our hearing organ. The tectorial membrane provides a cover for the organ of corti.. The organ of corti has four rows of hair cells. There are three rows of outer hair cells, and one row of inner hair cells. The auditory nerve is connected to the inner hair cells. These hair cells stick up into the tectorial membrane. When the stapes pushes against the oval window, it sets up a disturbance in the fluid of the cochlea. This disturbance causes the basilar membrane to move up and down, causing waves in the fluid of the cochlea. The basilar membrane is set up in a tonotopic organization, , so depending upon where the wave breaks, you will either hear a high or low frequency sound. If the wave breaks closer to the stapes, the person will hear a high frequency sound. If the wave breaks further away from the stapes, the listener will hear low frequency sounds. These movements of the hair cells set up an electrical impulse, which fire into the eight cranial nerve, and up into the brain for the sensation of hearing.

The various deformities associated with facial and oral development are also very complex, and require an in-depth description. These defects are the result of a multiplicity of factors; some genetic, some environmental, others a combination. The study of these anomalies is known as Teratology. The mechanisms which are involved in

facial mal-development are still puzzling, but recent research on facial development has discovered a link between the cerebral rhombencephalic and prosencephalic organizing centres.

Defects or disruptions that occur within the Rhombencephalic Organizing Centre, are associated with anomalies occurring in the middle and lower thirds of the face. These anomalies are caused by disruptions or displacements with the structures involved, causing various defects to the developing embryo. Three terms are used to describe these glitches in development; malformations, deformations, and disruptions. It is important for the purpose of this literature, to point out the differences associated with these three terms.

> "Malformations arise from abnormal development, predominantly during the embryonic period, and are not self-correcting. Deformations represent normal development thwarted by mechanical constraints. Disruptions are the result of frustrated development of normal organs. Both deformations and disruptions occur in the fetal period of development, and may correct themselves." (Peterson, Jones, Kamell, 2001, p. 50).

It has also been discovered that the membranous portions or bones of the facial skeleton are more susceptible to anomalies, than are the harder cartilaginous portions or bones in the skull.

Ear Malformations

During this section of the literature review, microtia and atresia, which are outer ear pathologies, and middle ear congenital deformities that arise in children in the cleft populations will be discussed. These various congenital deformities may eventually give rise to potential audiologic and otologic problems. Basically, the manifestation of an anomaly can occur anywhere in the outer, middle, or inner ears. Outer ear pathologies consist of microtia, atresia, and anotia.

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Children who are born with microtia have no outer ear (pinna), due to it's lack of development. Microtia, which means "small ear", is typically due to disruptions in development, involving the structures from the first and second branchial arches. Microtia generally affects more girls than boys, and is more common to manifest on the right side rather than the left. Microtia varies in severity, depending on the time frame when the structures involved in development were disrupted. There are three types of microtia. In Type 1 microtia, the auricle is still recognizable. Type 2 is characteristic of a very long and vertical auricle. There is some recognizable cartilage, and a sign of the formation of a helix (upper rim of ear). In Type 3 microtia, the cartilage and tissues involved appear soft, and no longer take on the characteristic shape of an auricle.

It is the parent's decision if they want their child to undergo surgical intervention to correct the deformity. Plastic surgeons will usually perform reconstructive surgery when the child is between 6 and 12 years of age. Reconstruction of the child's external ear usually requires three to four follow-up surgeries. Depending upon the severity of microtia, and as long as no cartilage is blocking the intact structures of the middle and inner ears, children born with microtia shouldn't suffer from any threats to their development of speech and language.

Another common outer ear pathology is known as atresia. With atresia, the child has been born with a blocked, or closed off, external auditory canal. Blockage due to a membranous mass, and blockage due to an osseous mass, are two different causes of atresia. The membranous mass is less common than the osseous mass. With the osseous mass, there is literally a solid mass of bone which has formed into a lateral wall, blocking the middle ear cavity, allowing no sound waves to enter beyond that point.

As discussed previously, middle ear pathologies are more common in children with cleft lip and/or palate. These pathologies of the middle ear include: ossicular abnormalities, Eustachian tube malfunction, and otitis media.

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Ossicular abnormalities affect the three bones in the middle ear cavity; the malleus, incus, and stapes. The ossicles could be fixated with other structures, commonly associated with the footplate of the stapes becoming stuck or fixated within the structure of the oval window. Ossicular discontinuity can also occur. Discontinuity can occur when the ossicles may have become separated from each other. Both of these problems can result in distortion in one's hearing, and can harm a child's speech and language development. . The causes of ossicular abnormalities are generally due to genetics, and to teratogens found in the environment. .

Eustachian tube malfunction is the most prominent problem associated with the cleft population. The Eustachian tube is extremely important for the proper functioning of the middle ear. The eustachian tube has three primary functions; (1) it provides for aeration of the middle ear cavity, (2) it provides for equalization of pressure between the middle ear cavity and the outside environment, and (3) it allows for drainage of middle ear fluid of secretions into the nasopharynx. Thus, chronic otitis media can be the result of the malfunctioning of the eustachian tube.

The main muscle that is responsible for opening the Eustachian tube is the Tensor Veli Palatini. "The high susceptibility of children with clefts to middle ear disease, is that the cleft interferes with the ability of the tensor veli palatini to open the Eustachian tube." (Peterson, Jones, Kamell, 2001, p.148).

The two most prevalent causes of malfunction are obstruction, , and a dysfunction in the opening of the structure. Obstructions to the Eustachian tube can be inflammatory due to illnesses such as; sinusitis, allergies, and nasopharyngitis. Obstructions to the Eustachian tube can also be lymphatic. The most common cause of this problem is due to extensive swelling, involving the structures of adenoidal and lymphatic tissues used in opening the Eustachian tube. The dysfunctions of the opening mechanism arise from anatomical differences associated with muscle function. "Faulty anatomy leads to faulty function." (Seaver, 2001).

As discussed previously, otitis media is due to these malfunctions and dysfunctions of the anatomical structures involved. The severity of otitis media is associated with stages. At first, the Eustachian tube is unable to open for aeration. As this is happening, the middle ear is beginning to absorb more and more air, creating negative pressure in it's cavity. Because the tube cannot open, the necessary release of fluids during aeration has been obstructed, contributing to an excessive accumulation of fluid in the middle ear cavity. When the pressure and fluid build up to an excess, the tympanic membrane is perforated.

Otitis media can be treated with antibiotics, antihistamines, and decongestants. Peterson, Jones, and Kamell (2001) suggest that when a child has had three incidences of otitis media in one year, has had excessive fluid in the middle ear cavity for more than eight weeks, then surgery is usually recommended. Typical surgeries involve inserting (PE) tubes into the middle ear cavity to restore the equalization of pressure.

As touched upon before, the effects of otitis media can have a severe impact on a child's speech and language development. "Clinicians have speculated that the mild to moderate hearing loss that occurs secondary to otitis media with effusion, may be associated with delays in cognitive and language development, as well as with delays in academic performance." (peterson, Jones, Kamell, 2001, p. 192).

During a child's development of speech and language, chronic otitis media can have a profound influence on the way the child perceives speech. The hearing loss associated with the otitis media creates an even greater obstacle for the child with a cleft palate. Because children are in the process of learning speech and language, they don't have the necessary language foundation, preventing them from using contextual cues and experiences to decipher speech.

> "Fluctuating hearing loss may result in an inconsistent auditory signal, which may lead to inaccurate encoding of information. When disruption to the auditory signal is prolonged or frequent, a child's ability to perceive

> and discriminate certain speech units may be impaired. This problem may also lead to attention problems if a child decides to simply "tune out" the signal. Illness that is associated with middle ear disease may restrict or alter the child's interactions with people and objects in the environment, resulting in fewer opportunities to establish a knowledge base from which speech develops." (Peterson, Jones, Kamell, 2001, p.192).

That is why is so important for early surgical and behavioral intervention.

Speech and Language

Speech-language pathologists are concerned with a child's entire speech and language development, regarding his/her effectiveness in communicative abilities. As children grow and develop, they learn how to move their articulators; the lips, tongue, teeth, and palate, in synchronous coordination with the muscles of respiration and phonation, to produce speech. When a child is born with a cleft lip or palate, these structures required for the normal development and articulation of speech is interrupted. This section of the literature review will discuss the services that the speech-language pathologist can provide for the child with a cleft, in addition to touching upon the importance of parental involvement in the therapeutic process.

Once an infant has been diagnosed as having some form of a cleft, early intervention is critical, for this is the time to educate the parents on how to establish the promotion of normal speech and language development. The parents should be provided with information regarding the anatomical role of the structures at play, in addition to their contributory function of the proper development of their child's speech and language. The parents should also be informed about any restrictions the cleft may cause to their child's overall development.

It is important to mention, that to be an effective clinician, one must remain objective, and accept the heterogeneity that is evident within this population. During the

initial evaluation by a speech-language pathologist, the child's motor development, communication development, the functioning of the oro-motor/speech mechanism, and the child's phonation abilities should all be assessed. It is extremely important for the speech-language pathologist to assess the child's motor development, as any noticeable delays may be indicative of a specific motor impairment, leading to the diagnosis of a more global developmental delay or malformation syndrome. Delays in motor development may also serve as predictors of possible future developmental delays.

The speech-language pathologist should also monitor the child's initial milestones, specifically in the amount of babbling the child takes part in, as well as the amount of expansion seen in the child's phonemic and phonetic repertoire prior to their first successful words.

While assessing the child's communication development, it is important for the speech-language pathologist to take a speech sample from the child during a play activity. This will provide an indicator of the child's language abilities and word usage, that does not rely on a highly structured, and unnatural activity.

While assessing the child's speech mechanism, the speech-language pathologist should also differentiate the role of structural restrictions placed on speech function, due to the physical impairments. This means that the speech-language pathologist should have the child use as many different articulators and aspects of the speech mechanism as possible, in order to establish a global representation of it's overall functioning and capabilities. The speech-language pathologist can then identify any developmental motor delays that may be putting the child behind, in regards to their speech and language development. Examples of activities that are commonly used to assess the speech mechanism are tasks which involve word imitation, with the use of such as games as Peek-a-boo, and Patty Cake.

The speech-language pathologist can also assess a child's protrusion of lips during a blowing activity, a child's range of tongue motion, and a child's phonation ability, by

assessing the quality of laryngeal function during speech. Abnonnallaryngeal quality may be a sign of an airway obstruction, or some other complex syndrome.

As mentioned at the onset of this section, the parents should also be informed of any restrictions an unrepaired cleft may cause. The parents should be involved as much as possible, for they are the ones who are going to be around their child the most. Parents should stimulate their child with speech and language as much as possible. The parents should talk to their baby as much as possible, using both visual and tactile cues. Parents should help their baby begin to understand words, using toys as a reference, and should help their child say the words he/she already knows, using picture books, and referencing to things that are part of baby's everyday schedule. By helping the child put words together while reading nursery rhymes and songs, the child will learn even more words and meanings to objects. It is also important to encourage the child to talk with other family members, such as grandparents, who are generally relevant in the child's life. Let the child play telephone, so he/she can babble and play. The greater number of opportunities the child has to conversate, the better the child's speech and language development will be.

When the child gets a little older, the parents or caregiver should try and restate the correct production of a word with added stress, after the child had attempted to articulate it, so the child is auditorily bombarded with the word or sound's correct production. After each occurrence of the restated correct production, the child should start to get the hang of how to articulate that particular sound or word correctly. The parents should also try and expand their child's vocabulary by adding in words.

A child who has a cleft lip and/or palate are going to be receiving services from a speech-language pathologist. However, the therapy shouldn't end at the completion of the therapy session; speech correction should continue at home. As previously discussed, it is extremely important that the parents, or primary care givers, be actively involved in the proper development and management of their child's speech and language. Most

importantly, the parents or caregivers should make practicing speech at home a "fun time", and should be done in calming and relaxing environments. It is important for the child to perceive these speech exercises as fun, otherwise the child may develop a negative perception about speech and communication. In addition, parents should try to use positive verbal reinforcement, and show extreme patience. The parents should make speech exercises into a game. The child can color, cut and paste things, or take part in reading their favorite stories. The speech-language pathologist may recommend that the parents keep a notebook to write about their perceptions, feelings, concerns, and/or improvements that they are noticing in their child's speech and language development in their natural home environment.

Speech and Language Development

This section of the literature review will discuss the developmental stages of speech and language that children go through. Normally developing children pass through a series of developmental stages during the first year of life, in response to developmental changes in both the nervous and musculo-skeletal systems.

> "The same environmental factors which influence the development of speech in healthy children, are important in those who suffer from clefts of the lip and/or palate: social class, place in family, the talkativeness of the parents, the kind of company with other children, and other environmental factors, all make a difference to the rate of the child's speech development. These factors are likely to influence the rate of speech development even more significantly in children with a speech handicap." (Peterson, Jones, Karnell, 2001, p. 177).

The first 12 months of speech and language development is termed the pre-linguistic period. This period of development is characteristic of the child not yet being able to produce words, because vocal tract differences restrict effective vocal

productions. A child during this period of development is attracted to prosodically varied tones.

The pre-linguistic period can be divided into different stages. The first stage is called phonation, and lasts from birth to one month of life. During this stage, there are no true speech sounds. The baby is restricted to vegetative and comfort state productions. There is little constriction occurring in the vocal tract, meaning that the sounds that are produced from the child represent a more closed production. However, the baby has the ability of discriminating between different phonemes.

The second stage is known as the cooing or gooing stage, occurring around two to three months of age. The child is now producing velar consonant like sounds with relative frequency, but have not yet mastered adult consonant sounds. Acoustically, the cooing and gooing sounds are similar to rounded back vowels, such as *lui,* as in boot.. By the second month of life, the baby can distinguish his/her mother's voice, and can discriminate between changes in frequency. Infants are strongly attracted to the sound of their own language, known as motherese. The infant's close attention to the mother's speech, plays a major role in the child's identification of relevant units of adult speech.

The third stage of development in this first year of life is known as expansion, , occurring between four and six months of life. During this stage, postural changes result in the child gaining increasing control of both laryngeal and oral articulatory mechanisms, due to changes in the shape of the oral cavity, facilitating in greater tongue mobility. The baby uses squeals, growls, yells, and raspberry vocalizations, which can be described as spitting and vibrating out of the lips. Also during this stage of development, the child begins to produce adult vowels.

> "The latter part of this expansion stage, and the subsequent canonical babbling stage, are sensitive periods for perceptual and motor learning. Indeed, it is during this period of phonetic development that differences begin to emerge between normally developing infants, and infants with a

hearing impairment and clefts." (Peterson, Jones, Kamell, 2001, p. 175).

The fourth stage of the pre-linguistic period is termed canonical babbling, occurring around six to eighth months of life. During this stage, the child uses true consonants in combination with a fully resonated vowel; consonant + vowel = babble; examples include, Baba, Gaga, Dada. At seven months, the child can discriminate between varied intonational patterns. The final stage of the prelinguistic period, occurring between six months and 12 months, is known as variegated babbling. During this stage of development, the child increases his/her use in productions of varied consonants.

The second period of speech and language development is called the pre-representational period, occurring between 12 and 24 months of life. This stage is also known as the "50 word stage". During this period of development, the child's receptive language exceeds his/her expressive language. The child can understand around 200 words, but can only produce a small number of them. This first "50 word period" begins with the first meaningful word, and ends when the child begins to put true words together. A true word is "a relatively stable phonetic form that is produced consistently by the child in a particular context, and is recognizably related to the adult like word form." (Meredith, 2002). The child is likely to produce more consonants in the initial position, rather than medial or final consonant positions. Common tendencies that are typically seen during this period, are the child's use of invented or protowords, and phonetically consistent forms (PCF's). These are "consistent vocal patterns that accompany gestures prior to the appearance of words." (Bliele, 1996, p.37).

Also during this stage, the child is typically producing syllable structures such as consonant-vowel (cv), vowel-consonant (vc), and consonant-vowel-consonant (eve). In learning and acquiring novel sounds, the child develops individualized acquisitional patterns. There is much variability here, for a child may use salience (selection), avoidance. or both. In salience (selection), a child will select words to use that are

already found in their phonological inventories. In avoidance, the child will avoid words that are not yet found in their phonological inventories.

The third period of speech and language development is called the early representational period, with a time frame from two to five years of life, and is characteristic of the pre-school age child. During this period, there is a large growth in the child's correct production and usage of consonants and vowels, due to their reorganization of phonologies into systematic patterns, or phonological processes. Children during this stage are more ready physiologically, and more cognitively able and alert. Towards the beginning of this period, the child moves from producing 50 words to 150-300 words, and can understand between 200 to 1200 words. By five years of age, the child can produce around 2500 words, and can understand around 10,000. As the child begins to use two word utterances, they show use of contrastive stress. The use of contrastive stress, is recognizable when a syllable in a word, or a word in an utterance becomes more prominent.

The final period of a child's development of speech and language is called the late representational period, occurring around five years of age and greater, and is characteristic of the "school age child". During this time, the child may still exhibit some phonological processes, but their speech rate increases dramatically. The child learns to adapt to phonetic characteristics of home and peers, regarding dialect and cultural differences. Children's perceptual abilities continue to improve, in association with learning appropriate oral-motor control and timing skills for more complex words.

Obviously, children go through very complex developmental periods, and a facial deformity involving one of the many articulators may pose various threats to the normal development of speech and language. "Compared with their same age non-cleft peers, babies with unrepaired cleft palate vocalize less frequently, and demonstrate fewer total consonant productions, and a more restricted consonant inventory during babbling." (Peterson, Jones, Kamell, 2001, p. 178). Even for those babies who have had the

reconstructive palatoplasty surgery, differences in development for these babies continue to persist, influencing subsequent phonetic development. Because these children demonstrate delays in canonical babbling, have a reduction in the total number of babbles produced, and have a limited consonant inventory, they are of major concern. These problems "limit the opportunities that the child has to establish the feedback system needed to produce and monitor speech." (Peterson, Jones, Kamell, 2001, p.178). Babies who have not undergone any type of surgery for the reconstruction of the palate, are forced to execute and practice sound productions with an inadequate articulatory surface, resulting in a distorted oral-motor movement, and a distorted auditory output. Steel-Gammon (1992) and others have emphasized the role of feedback and practice on normal vocal development.

> "Speech has a skill component, and as with any skilled activity, practice increases the control and precision with which movement is performed. Thus, the more often a baby produces the movements that shape the vocal tract to produce particular sounds and sound sequences, the more automatic those movements become, and ultimately the easier it is to execute them in producing meaningful speech." (Cited in Peterson, Jones, Kamell, 2001, p. 451).

This is why the babbling period is so important. The more consonant-vowel like productions the child is able to produce and practice early on, the more the child can attach meaning for later acquisition and development of words. When a baby systematically executes repeated productions, it gives them oral and motor feedback. Both limited oral and motor practice, and distorted auditory feedback, can be one of the causes for early delays in speech and language development.

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Speech and Language Impairments

Now that speech and language development has been discussed in children, this section of the literature review will be discussing the characteristics typically seen in the speech and language of children in the cleft population. Children who are born with a cleft lip, typically go through the stages of speech and language development without any complications. For children who are born with a cleft palate, the majority usually develop efficient speech following the repair of the cleft. However, a repaired cleft palate can still have an impact on the child's articulatory abilities, mainly affecting the quality of speech.

Recent investigations have concluded that individuals with a cleft palate have significant improvement in their articulatory performance with age.

> "The development of speech may be altered in many different ways by many different factors. Speech sounds may be defective in later life, even when anatomical defects have been corrected. This may be due to the use of faulty methods of speech sound production acquired in earlier life when speech was developing, where the child was having to compensate for such difficulties." (peterson, Jones, Kamell, 2001, p.155).

In general, children with clefts are at an increased risk for developing both phonetic and phonological based disorders, in conjunction with having an extremely slow rate of early speech development. .

> "Phonetic errors occur as a result of structural deviations associated with the anatomical differences associated with the cleft. Phonological errors may occur in relation to either general expressive language delays, or physical constraints imposed by the cleft. Articulatory errors become integrated into the child's developing phonological system over time." (Peterson, Jones, Kamell, 2001, p. 181).

It is evident that speech and language deviations are based on the type of cleft the child is born with. The findings of many studies suggest that the articulation abilities of

children with a cleft lip is almost perfect, while those children with clefts of the palate suffer from poorer articulatory abilities. In addition, children with clefts of both the soft and hard palate have poorer speech than those children with a velar cleft, and those children with a unilateral cleft have better speech abilities than those children with a bilateral cleft.

> "Children who are born with cleft lip and/or palate are at risk for resonance, articulation, and expressive language problems that may impair communication for many years. The most remarkable speech production problems demonstrated by children with cleft palate, are those related to velopharyngeal inadequacy, including hypernasality, audible nasal emissions, weak pressure consonants, and compensatory articulation patterns." (Peterson,, Jones, Kamell, 2001, p. 162).

Children born with a cleft palate who have not undergone palatal surgery may experience inadequate maneuvering of the musculature of the velopharyngeal system, meaning that the muscles are not in synchrony with the other surrounding crucial muscles. Crucial muscles functioning within this velopharyngeal system include: the Levator, which functions to raise the soft palate; the Tensor, which functions to tense the soft palate; the Palatoglossus, which runs from the soft palate to the tongue, and functions by pulling the soft palate down, and raising the back of the tongue; the Palatopharyngeous, which is the main muscle of the soft palate, functioning to lower the soft palate; and the Superior Constrictor, which is the upper most muscle of the pharynx, forming the sides and back walls of the nasopharynx, and the oropharynx.

The velopharyngeal mechanism is usually open during breathing, and closed during speech. This mechanism is also involved in non-speech activities which requires closure, by sealing off the nose from the mouth. These non-speech activities include swallowing, blowing, and sucking. In fact, the velopharyngeal mechanism controls the oral-nasal distinction of speech.

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Speech production problems that are associated with cleft palate and velopharyngeal inadequacy are; resonance problems, articulation difficulties, including compensatory articulation patterns, and nasal emissions. Children with resonance problems can sound hypernasal, hyponasal and/or denasal.. Hypernasality refers to a "resonance alteration of vowels and vocalic consonants, that occurs when the oral and nasal cavities are abnormally coupled." (Peterson, Jones, Kamell, 2001, p. 162). The speech of a child who has hypernasality associated with their speech, sound as though they are speaking through their nose. The causes of nasal escape following an operation may occur for a number of reasons.

> "In some cases, it may be impossible to repair the original defect completely, and escape may continue to occur through it. In others, the process of repair may result in shortening of the soft palate, so that nasal escape occurs behind it. Sometimes there is considerable scarring of the palate after operation, particularly if post-operative infection has occurred. As a result, the palate is relatively immobile, and functional nasal escape occurs during speech." (Peterson, Jones, Kamell, 2001, p.160).

Hyponasality and denasality "refer to a reduction in nasal resonance, when the nasal airway itself is partially blocked, or the entrance to the nasal passages is partially occluded." (peterson,, Jones, Kamell, 2001, p. 163).

Children born with a cleft lip and/or palate may also have articulation difficulties associated with their speech. According to Peterson,, Jones, and Kamell (2001), children who are born with a cleft lip and/or palate generally demonstrate poorer articulatory competence, than do normally developing children.

> "Descriptive accounts of articulation in speakers with cleft palate have demonstrated that omissions and substitutions occur more frequently than other error types in young children,, although errors related to distortion occur most frequently in older children and adults. Numerous

> investigations have demonstrated that children with cleft palate have more difficulty producing pressure consonants than other classes of consonants. They typically misarticulate fricatives and affricates most frequently, followed by plosives, glides, and then nasals." (Peterson, Jones, Kamell, 2001,p.171).

Early language delays which are typically seen in children with a cleft are the deletion of final consonants, reducing the number of syllables in a word, and backing. These early developmental delays usually become less apparent by the time the child has reached four to five years of age.

Children with a cleft palate also tend to utilize compensatory articulation patterns. Compensatory articulation patterns are defined as atypical patterns of articulation, formed by the child before the surgery had taken place, where they had continuously attempted to produce and articulate proper speech. Some of these compensatory articulation patterns may be so deeply engrained in the child's speech behavior, that when the surgery has been completed and healed, the child may still continue to utilize them. These compensatory patterns may also develop, or continue to persist due to the decreased surface area of the palate for proper articulation, and often times because of velopharyngeal inadequacy.

Examples of these compensatory patterns are often defined as passive, compensation, , and camouflage. In a child who shows passive compensatory articulation patterns, they make no attempt to try and fix their problems. In a child who shows compensation, they may substitute or omit sounds that they have trouble producing. In a child who demonstrates camouflage, he/she "attempts to mask the perceptual consequences ofvelo-pharyngeal inadequacy, through the use of weak articulation." (Peterson, Jones, Kamell, 2001, p. 165).

Tentative conclusions have been drawn regarding the prevalence of articulation disorders within this population. "The prevalence of articulation problems in the cleft

palate population, is probably greater for those children and adolescents who do not receive continuing management by a cleft palate team." (Peterson, Jones, Kamell, 2001, p.l64). Sperber (1989) enforced that it is expected that up to 25% of preschoolers who have under gone lip or palate repair, and who have continued treatment with the cleft palate and cranial-facial team, should have normal articulation. However, there are always those significant number of children who continue to experience articulation difficulties.

Speech-Language Therapy

Although children born with a cranial-facial anomaly may suffer from a developmental delay in language, speech, or use of voice, there are many treatments for these problems. This section of the literature review will discuss the various speech and language treatments, and therapy options that are provided by the speech-language pathologist..

The most important factor in the rehabilitation of children with an anomaly is early intervention. The main goal for a speech-language pathologist working on a cleft palate team is the importance of educating the parents or caregivers. When the parents or caregivers arrive for their first evaluation, they are often unsure of how the anomaly will impact their child's development, and in turn, are not sure of what they should be expecting from their child. Thus, it is extremely important for the speech-language pathologist to provide the parents with accurate and appropriate information early.

The speech-language pathologist should provide the parents with information on normal language development, and should give the parents suggestions on ways to help their child achieve successful speech development at home. "First and foremost, parents of babies born with cleft palate should be informed of the impact that the cleft has on the child, and how this may be expected to affect the child's early communicative efforts, as well as speech production performance." (Peterson, Jones, Kamell, 2001, p. 289).

In addition to providing the child's parents with accurate information, the speech-language pathologist also acts as a counselor, regarding the expected impact of the anomaly on speech and language development, and also encompassing social and emotional issues. The parents are indeed upset about what has happened to their child, and they are most likely to feel guilty. For one, the parents have had their dream of a perfect child taken away from them. They were expecting their child to be born normal, and the presence of their child's anomaly has shattered that possibility.

As early intervention and parent education are extremely crucial components in helping the child, it is also very important for the speech-language pathologist to teach the parents how to carry over the therapy being provided to the home, by encouraging aggressive language stimulation. Language stimulation that the parents can engage in, can be used in babbling games and vocal play. Also, language stimulation targeting the enhancement of early vocabulary development may also facilitate the enlargement of the child's phonetic repertoire. "Parents are going to teach speech and language, so they need proper guidance and direction. If the parents aren't involved in the management process early on, they may inadvertently assign all responsibility for management to the professionals involved with the child." (Peterson, Jones, Kamell, 2001, p. 289).

Peterson, Jones, and Kamell (2001) set forth an example of an optimal therapeutic program for a child's speech and language development which includes: the counseling of parents in regards to language development; a home program delivered by the child's parents and caregivers, who provide an extensive language stimulation program; direct speech therapy provided by the clinician, depending upon the individual needs of the child; and for the Cleft Lip and Palate Team to practice a team management approach.

Another example of an optimal therapy program is direct articulation and phonological therapy. As with any therapy being provided, the frequency of the therapeutic visits is dependent upon the individual needs of the child. With this type of therapy, the best results are seen when accomplishing short term goals of a larger, and

more complex long term goal. The speech-language pathologist teaches the child how to produce sounds in syllables and words, with the main goal of establishing automatic and fluent use of the sounds in conversational speech.

The material used to teach the child should be age appropriate, with the speech-language pathologist providing auditory and visual cues, and responding to the child's correct productions with positive verbal reinforcement. . For younger children, tasks which involve blowing bubbles or blowing through a whistle, are often helpful in teaching the concept of oral airflow. "Young children who exhibit a restricted consonant inventory, and those who produce compensatory articulatory patterns, are frequently enrolled in therapy designed to increase the strength of oral musculature." (peterson, Jones, Kamell, 2001, p. 292).

Another example of an optimal therapeutic program is electropalatography. This procedure is used with children and adults with cleft palate, and is used to study lingual articulation, , and to treat disorders of articulation in children.

> "Instrumentation involves an acrylic palatal plate fabricated for each client, in which electrodes are embedded throughout the plate, and are exposed on the lingual (tongue) surface. When the electrodes are contracted by the tongue, a signal is sent to an internal processing unit through lead-out wires. Real time visual feedback of the location and timing of tongue contacts with the hard palate is provided on a computer monitor." (Peterson, Jones, Kamell, 2001, p. 298).

Clinical fmdings suggest that the patients treated with electropalatography showed rapid and dramatic improvement in their communicative abilities. In fact, electropalatography offers adults and children with cleft palate an alternative feedback system. It is helpful for the patients who are unable to modify their behavior through verbal reinforcement from the clinician, and from tactile / kinesthetic cues.

The modification ofhypernasality , and nasal emission for those children and adults with velopharyngeal inadequacy is another example of a therapy approach. The main goal for this type of therapy is to attempt to manipulate the muscles and articulators of the oral cavity in such a way, to achieve a change in oral resonance.

One way of manipulating the oral cavity to see a change in resonance, is by increasing the client's opening of their mouth. As the client opens their mouth wider during speech,, the amount of oral airflow increases, decreasing the amount of nasal airflow. This decrease in the amount of nasal airflow results in speech which is perceived as less nasal. The clinician should provide the client with materials representing a wide range of articulatory movements which foster greater mouth opening. One helpful way for the clinician to make their client more aware of their mouth opening, is to have them practice speech in front of a mirror.

Specific Syndromes

Up to this point in the literature review, different types and severities of the otologic and audiologic problems that are common in children born with a cleft lip and/or palate have been discussed in great detail.. The wide variety of treatment options and services that speech-language pathologists working on a cleft lip and palate team can provide, has also been discussed. Different aspects and characteristics affecting the speech and development of language have also been discussed. In this section of the literature review, three different syndromes that can co-occur with cleft lip and palate will be discussed. These syndromes are: Hemifacial Microsomia, , Mandibulofacial Dysostosis, and Nager Syndrome.

llem~acialAlicrosomia

Children who are born with Hemifacial Microsomia are characterized as having an asymmetrical orientation of their facial features. During embryologic development,

there is a disruption of some sort having it's impact on the proper development arising from the first and second branchial arches. As a reminder, the first or mandibular pair of branchial arches are the primitive formations of both the upper (maxilla) and lower (mandibular) jaws. The second branchial arch is responsible for the formation of the third and smallest ossicle of the ear, the stapes, and contributes to the development of the other ear ossicles, the malleus and the incus. The number of children who are born with Hemifacial Microsomia ranges from 1:3000, to 1:5000.

> "Unilateral or bilateral cleft lip and/or palate occurs in an estimated 7% to 15% of patients with Hemifacial Microsomia, , with cleft palate occurring about two times as often as cleft lip. In addition, these patients may exhibit non-cleft velopharyngeal incompetency, resulting from asymmetric movement of the velum and/or pharyngeal musculature." (peterson, Jones, Kamell, 2001, p. 41).

Most of the children who are born with this syndrome are affected on both sides of their face, usually with one side more deformed than the other. Actually, this syndrome is characterized as belonging to a family of deformities, which can occur alone, or in association with other syndromes of malformation. Children with this syndrome usually have deformities associated with the ears, and both the upper (maxilla) and lower (mandible) jaws. Because a child with this syndrome may have many deformities associated with his or her facial features, it is extremely important for them to undergo a full spectrum of evaluations. If the child has an anomaly of the ear(s), the parents should take their child for a full audio logic evaluation, to find out if the he/she has a hearing loss.

There are different degrees of severity associated with the facial deformities. Most commonly, a child born with greater malformation to their mandible, will have a greater chance of more severe deformities to their ears. For example, the deformity of the ear(s), can range from only slight differences in the auricle (visible external ear), to an entire absence of the external ear, and ear canal. These deformities can also stretch into,

and affect the middle ear cavity and it's ossicles. The deformities of the middle ear can also range from mild to severe. The facial nerve has it's trek up into the brain through the middle ear cavity, so any deformity in it's pathway poses a threat to potential damage.

Most children with outer and middle ear deformities are reported to have a mild to severe conductive hearing loss. If the deformities are severe enough, they may effect the functioning of the cochlea, leading to a sensorineural hearing loss.

A child's ears may also be displaced, meaning that they are too high set, too low, too far forward, or too far back.

If the child has deformities involving both of the jaws, they can also manifest as mild to severe. An example of a mild jaw deformity is an underbite or overbite, compared to a severe deformity, where the child has a jaw with no functioning capability of opening, closing, or articulation.

These children may also have deformities to their eyes, again ranging from mild to severe. In addition to the deformities of facial features, it is extremely important that these children have an evaluation of the functioning of their heart, spine, and kidneys.

All of these deformities that can manifest themselves in all areas of the child's body can pose potential detrimental threats to their physical development, and to their communicative abilities. The child may have a hearing loss, creating a possible loss of speech and language comprehension, development, acquisition, and communication. The child's speech capabilities may also be restricted, due to physical obstructions of the oral and facial articulators, in association with asymmetric musculature, resulting in unintelligible speech.

> "Normal interaction between parent and child, and other environmental aspects is impaired. Stimulation for the infant is limited by the hearing loss, until the loss is assessed, and appropriate amplification is provided. The infant's stimulation is also limited by the inability of adults to

recognize and reinforce early speech attempts." (Peterson, Jones, Kamell, 2001, p. 47).

Early intervention is key. The earlier the child can begin their normal development of speech and language, the better off they will be with their communication and comprehension capabilities. An evaluation with an audiologist and speech-language pathologist is highly suggested for the proper development of the child. The child may even undergo a wide variety of reconstructive surgeries, where the repositioning of the oral and facial features is common.

Mandibulofacial Dysostosis

The second syndrome to be discussed is known as Mandibulofacial Dysostosis. Mandibulofacial Dysostosis, just like Hemifacial Microsomia, is a syndrome that manifests itself as multiple deformities of the face. It's primary defects are seen in the eyes, the ears, and both the lower (mandible) and upper (maxilla) jaws. Again, as with Hemifacial Microsomia, the deformities associated with this syndrome are limited to the structures of the first and second branchial arches.

This syndrome has been extensively studied by geneticists, where they have discovered the presence of an autosomal dominant gene, being traced to chromosome five. According to Jones (1997), the frequency of occurrence of Mandibulofacial Dysostosis is extremely rare, occurring in one out of every 50,000 live births. It is known that around 65-80% of these patients also exhibit some form of clefting. (p. 10).

Although the facial deformities are described as being in symmetry with each other, the children born with this syndrome are characterized as having a "downward slant to the comers of their eyes; lower eyelid defects, which include protrusions or notches, or a total absence of lower eyelashes; defects to the iris of the eyes; and also defects to the lower (mandible) and upper (maxilla) jaws." (peterson, Jones, Kamell, 2001, p.43-44). For example, the mandible in these children is characterized as being

extremely small, with extra protrusions and notches, contributing to some form of dental malocclusion. It is also highly predicted that these anomalies will become even more deformed with age. These children also experience pharyngeal deformities, where the diameter of their pharynx is unusually small.

Because of such complex oral and pharyngeal deformities, these children are at high risk of experiencing extreme respiratory complications during infancy. The abnormal small diameter size of the pharynx causes a restricted airway space, which may not allow the child to breathe normally. These respiratory problems may become progressively worse as the child grows and develops, due to the effects of the other deformities of the mandible and maxilla.

Children with Mandibulofacial Dysostosis may also suffer from ear deformities, which again range from mild to severe. The outer ear may be slightly deformed, to highly deformed, to completely absent. The external auditory canal (ear canal) leading to the middle ear may have a slight deformity, also ranging from mild to a complete absence. Deformities of the middle ear are also characterized as mild to severe, with their most devastating affects on the ossicular chain.

Some children may be born with an absence of the ossicles of the middle ear. Typically, children with deformities to their ears experience a mild to moderate bilateral, conductive hearing loss.

> "The hazards to normal early childhood development are multiple and potentially severe, with specific threats to communication development. The need for, and evaluation of neonatal hearing evaluation and assessment is crucial for potential successful communicative abilities. As a vast majority of these patients are of normal intelligence, the early recognition of deafness and it's correction with hearing aids or surgery (if necessary and if possible), are of great importance to development.". (peterson, Jones, Karnell, 2001, p. 44-46).

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Honors Capstone

Nager Syndrome

The third syndrome of which will be discussed is known as Nager Syndrome, also known as (Preaxial Acrofacial Dysostosis). When comparing the facial deformities that are characteristic to Nager Syndrome, they are extremely similar to the facial deformities associated with Mandibulofacial Dysostosis and Hemifacial Microsomia. Just like all deformities, the severity tends to vary from one individual to the next. The anomalies associated with the cranial and facial area are expressed compoundly (many occurring at the same time), usually restricting the child of achieving intelligible speech.

> "The cranial-facial features are characterized of downward slanting of the eyes, cheek extensions of scalp hair, lower eyelid defects, and mandibular and maxillary deformations. The lower jaw is even smaller and more deformed than in Mandibulofacial Dysostosis, causing severe problems in the opening and closure of the jaw. The inability to open the mouth creates problems in oral hygiene and dental care, and may prohibit surgical or prosthetic treatment of velopharyngeal abnormalities." (Peterson, Jones, Kamell, 2001, p. 46-48).

In fact, the functioning of the velopharyngeal structure is considered to be the worst when compared to Mandibulofacial Dysostosis, particularly due to the complete absence of the soft palate.

These children not only have severe deformities associated with their facial and cranial regions, but they also have extreme deformities of their hands and arms. Some form of malformation associated with the upper limbs is always present, and the severity varies from one individual to the next. In turn, the deformities of their hands and arms most often prevent them from learning and using sign language as an alternate means of communication.

For infants, the facial deformities pose deadly threats to their survival. "The deformities of the mandible and maxilla, as well as the displacement of the tongue- in

conjunction with the limitations of the opening of the jaw, cause airway obstruction." (Peterson, , Jones, Kamell, 2001, p. 47).

Toddlers are restricted in their learning experiences, for their deformities of their limbs prevent them from exploring and interacting with the environment. . "Each child with this syndrome requires early assessment, and continued individualized habilitative and educational planning to maximize communication skills." (peterson, , Jones, Kamell, 2001, p. 47).

Personal Observations

When parents first find out that their dream of having a perfect child has been taken away from them,, they may feel extreme guilt and pain because they think that they are the ones to blame. They were expecting their child to be normal and healthy, but the presence of their child's anomaly has shattered that possibility. As a speech-language pathologist, it is extremely important to not only act as a speech therapist, but to also be a counselor, by providing undivided attention,, compassion, and support to the parents and family members. It is important for the speech-language pathologist and other clinicians working with the child, to offer a great deal of opportunities where the family can discuss their fears as a new parent of a child with a congenital anomaly.

The speech-pathologist and the Cranial-facial team will work hard in educating the parents in letting them know important information about the child's growth and development. . As discussed in a previous section of the literature review, family education consists of letting the parents know what to expect in regards to the child's speech and language development, motor and mental development, , socio-communicative competence and development, and the possible negative potential reactions of others. Parents will usually ask the clinician a question when they are ready to accept the reality of the answer.

During this time of parent education, the speech-language pathologist and other clinicians working with the family, should encourage the parents to get involved as much as possible. Parents who feel that they are adequate and competent to care for their child, are the parents who are going to benefit their child the most. Peterson, Jones, and Kamen (2001, p.334), have utilized a number of approaches to be able to explain the experience of the initial feelings and fears of parents, who have just given birth to a child with a congenital facial or cranial deformity. Approaches which were the most effective included questionnaires, structured interviews, and anecdotal information.

> "Many reports, including first-person accounts provided by parents, point to the importance of the baby being shown to the parents immediately after birth, or as soon as possible. Delays in seeing the baby heighten anxiety, whereas being able to hold the infant, and keep him or her nearby, allows the parents to realize that he/she is more like a normal child, and can partake in bathing, cuddling, and comforting." (Peterson, Jones, Kamen, 2001, p. 334).

Pope (1999, p.36) voiced the fear that "if parents are not able to make the emotional adjustment at this neonatal time, their unresolved feelings are likely to reappear and negatively influence their parenting." (Cited in Peterson, Jones, Kamen, 2001, p.334).

There are many other concerns that the parents will continue to have. One of the biggest concerns is about the severity of the facial disfigurement. In this day and age, so much emphasis is placed on trying to be the prettiest or skinniest, which has created havoc among young women and even younger girls who have not yet reached their teens. In fact, many people believe that physical attractiveness may foster one's trek in achieving success, in both their professional and personal lives. The physical attractiveness of someone also contributes to other's interpretations. The physical attractiveness of the face is often felt to be extremely important, for some feel that it

allows an individual to communicate and express oneself to the world around them. Hughes and Ashgate (1998, p. 16) have reported that "the patient's self-perceptions, emotional stability, personality characteristics, and social circumstances appear to be the salient factors in dealing with maxillo-facial disorders and the rehabilitation process."

In addition to dealing with the facial deformity, when the child arrives at the preschool years, the parents and the child are often times faced with a whole new array of challenges. For one, children who are between the ages of four and six years, begin to become more aware of who they are and how they look. If the child has some form of a facial disfigurement, the child may have to put up with teasing, name calling, and harassing questions at school, related to why they look or sound a particular way. These negative reactions by the child's peers, in turn, causes the child to feel like an outcast, leading to a poor self image, and low self-esteem.

In addition to these problems at school, there seems to be much stereotyping going on in the classroom, for sometimes children will perceive someone to be less intelligent because of the differences in his/her looks. In addition to the bullying and stereotyping, a child with a cranial-facial anomaly may be absent from school many times for the necessary on-going therapy, medical evaluations, and surgical interventions, leading to interference in the child's social development with his/her peers. Peterson, Jones, and Kamell (2001, p.191), note that children born with a cranial-facial anomaly show decreased socio-communicative competence. Peterson, Jones, and Kamell (2001, p. 191) stated, "that a lack of assertiveness in this population could reflect a reluctance to communicate, because of either present or past problems with speech intelligibility, and negative listener reactions."

Although these children born with cranial-facial anomalies are faced with a wide variety of challenges throughout their development, these children show extreme courage and are extremely strong willed. As a student in the field of speech-language pathology, being accepting and open to the diversity seen throughout the world is an extremely

important trait. All children deserve to lead normal and healthy lives without experiencing negative comments and remarks. The Cleft Team can become a piece of the child's world, aiding in hislher proper development and management of speech and language, self-esteem, education, and social competence.

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