

Cleft Lips and Palates;The Roles Of Specialists

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Abstract

Cleft lips and cleft palates are among the most common of birth defects and if left untreated can lead to serious medical and concurrent speech and language problems. However, while the consequences of cleft lips and palates can be severe and long-lasting, these can be averted by medical intervention, especially if it is done as early as possible. This paper explores the various options for surgical, medical, dental, and speech and audiological management of cleft of the secondary palates in children with Apert's syndrome and the ways in which these interventions can help children with these particular birth defects.

INTRODUCTION

We should begin this discussion by establishing what a cleft palate is, in medical terms. Per the Cleft Palate Foundation website (accessed August 17, 2007) a cleft palate occurs when the palatal plates of an individual (which lie in the roof of the mouth) for various reasons fail to come together or "close" during the second month of fetal development. From an early age and up to adulthood, the skull, face, jaws, hands and feet undergo frequent surgical correction. These operations require planning and coordination amongst various specialists, including a neurosurgeon, a craniofacial plastic surgeon, an anesthesiologist, a maxillofacial oral surgeon, an orthodontist, a dentist, an orthopedist and an orthopedic surgeon.

Continuing care throughout the first twelve years of a child with Apert's life (as well as past the first twelve years) is facilitated by a social worker, psychologist, audiologist and speech language pathologist. In addition to the planning and performance of many operations at specialist clinics, each child and his or her family should have regular contact with the craniofacial team. There will often be meetings with several members of the multidisciplinary craniofacial team as well as other medical professionals: the occupational therapist, the pediatrician, the nutritionist, the guidance counselor, the psychologist and the physiotherapist. For the purposes of this research paper there will be a focused discussion of six medical professionals' roles on the craniofacial team in the first twelve years of a child with Apert's life. These professionals are: geneticist, speech language pathologist, audiologist, plastic surgeon, dentist and orthodontist.

ROLE OF THE GENETICIST

Consultation with a geneticist is crucial in order to do a DNA test of the child with Apert's as well as the parents of the child. Per the Mountain States Genetics Regional Collaborative Center's website (accessed August 11, 2007) in Apert's syndrome, there is bicoronal synostosis, midface retrusion, and symmetric syndactyly (webbing of the digits) in both the hands and feet. The craniofacial dysostosis syndromes are inheritable. Per Carinci et al (2005), the gene mutation for each syndrome has been identified. Apert's syndrome is inherited in an autosomal dominant fashion, meaning that there is a 50% likelihood of recurrence of the syndrome in the offspring of the affected individual. Often, the parents of these children are not affected, and the gene mutation arises spontaneously during development. However, the severity of the syndrome may vary from one generation to the next. The researchers state that the gene encoding for FGFR2 is mutated, and this results in a wide spectrum in the expression of phenotype and making anomalous development more complicated. Furthermore, Apert's may not have been detected in a parent, until a child with more remarkable Apert's traits was born. Therefore DNA testing is of the utmost importance to give parents the data to make informed decisions about the odds of bearing another child with Apert's or other genetically inherited disorders. Lastly, it is of great importance to determine the co-occurrence of other congenital and genetic anomalies that may affect the progression of the child's development.

ROLE OF THE SPEECH LANGUAGE PATHOLOGIST

Therapy and follow-up care is coordinated with the speech language pathologist (SLP). Per Shipster et al (2002), a cohort of ten children with Apert's syndrome was studied and a thorough analysis of their speech and language characteristics was done. They often have hyponasal resonance due to an under-developed midface, small nose, and excessively long soft palate. If there is a cleft palate, they may also have hypernasal resonance. Articulation of speech sounds is often distorted due to the malocclusion and high arched palate. Impaired hearing or a general developmental delay will also affect speech and language development. Individuals with Apert's Syndrome often require glasses to correct near- or long-sightedness and thus the Speech Language Pathologist must bear this in mind when doing drills using written media.

The researchers state that there are specific areas requiring therapy based on their examination and standardized testing of these children. Resonance and voice are at issue due to marked nasal obstruction, affecting the nasality of words produced by the children. Diplophonia and wet voice quality were apparent in the cohort's voice and resonance. As a result therapy must focus on nasality and voice therapy as well. Testing of the cohort determined severely impaired receptive and expressive language skills in nearly 50% of the group. This involves an approach to therapy which must address these issues to strengthen receptive and consequently receptive abilities. Attention was delayed by 2;0 to 3;0 chronological years. The cohort displayed single channel attention control that lags behind normal children. This means that therapy must focus on eye contact, pragmatics and posture issues. There were delays in the phonetic and phonological skills of these children. Phonetic errors mainly involved blade production of alveolar consonants as well as some lateralization of alveolar fricative and affricates. While these issues mostly would be helped by alveolar ridge surgery, in the meantime therapy may focus on proper articulation of these phonemes. Phonologically problems involved stopping of fricatives and affricates, final consonant deletion, voicing of voiceless consonants, and fronting of velars and palatoalveolar sounds. These are all major areas for work on the part of the SLP. It is important to note that areas of therapy for the cohort members during earlier years of life were: Portage and Makaton signing (which was relinquished to instead work on spoken language).

ROLE OF THE AUDIOLOGIST

Per Rajenderkumar, Bamiou and Simimanna (2005), the

major concern related to audiological treatment of Apert's is the risk of hearing impairment caused by repeated infections in the middle ear. However, the researchers point to the significant debate regarding the efficacy of repeated pressure equalization tube insertion vs. the efficacy of amplification to ensure hearing ability. Otolaryngologists will handle the middle ear infections by inserting pressure equalization tubes into the eardrum to equalize middle-ear air pressure and drain liquid. The audiologist will check the ears for placement of the tubes as they often fall out of place.

Per the researchers, some individuals may require hearing aids, as a hearing impairment will affect speech and language development. They argue based on their data from a cohort of seventy cases that hearing aids are more effective than tube insertion in the long run. Hearing impairments caused by sensorineural damage (the inability of the nervous system to mediate sound impulses) are uncommon. Further impairments of hearing that are monitored and managed by the audiologist (with consultation with a Otolaryngologist) are: low-set ears, microtia, macrotia, posteriorly rotated external ears, ossicular fixation, wide cochlear aqueduct, and abnormal surface configuration of the pinna. Regular audiological testing is done according to schedule in order to assess ongoing changes in the child's hearing.

ROLE OF THE PLASTIC SURGEON

Per the Children's Craniofacial Association website (accessed August 19, 2007) the physical characteristics of Apert's include defects of the skull, eyes, and face. The skull is: short from back to front, wide on the sides. The eyes are: bulging, eyelids tilt downward abnormally at the sides. The face is: mid-face has a sunken-in appearance, the upper jaw slopes backward, lower teeth project in front of the upper teeth. Early surgery relieves the pressure on the brain and eyes by allowing the bone plates of the skull to be detached from one another. Even in severe cases of Apert's syndrome a significant cosmetic and functional improvement is possible and a decreased risk of optic difficulties or blindness secondary to orbital hypoplasia can be achieved.

Per Paravatty et al (1999) plastic surgery procedures include release of the prematurely fused sutures; the traditional surgery involves advancing the frontal bones, correcting the bulging eyes and upper facial deformities including the retrusion or hypoplasia of the midface. Depending on the severity of Apert's syndrome and the associated congenital abnormalities, other operations such as rhinoplasty - plastic surgery of the nose, genioplasty - plastic surgery of the chin,

eye muscle surgery to correct strabismus or eyelid surgery to correct the abnormal downward tilt, and surgical separation of the fingers and/or toes are performed according to a staged treatment plan. To give the brain space to grow and to improve the shape of the head, the fused bones are subjected to early surgery, often when the child is six months old. Corrections of the midface and jaws are currently not undertaken until adolescence, when all the permanent teeth are in place. It is also important to note that plastic surgery of the hand in Apert's syndrome often has to be started early, to allow the child to develop a grip.

Per the Mountain States Genetics Regional Collaborative Center's website (accessed August 11, 2007), the fingers are all separated and shaped, using skin and possibly also bone transplants. This may enable the child to have three to five fingers on each hand. After such surgery, the child will require training under the supervision of an occupational therapist to develop grip and coordination; the Speech Language Pathologist will work on similar issues of tone and articulation related to muscle strength in the speech mechanism.

Per Sadove, Van Aalst, and Culp (2004) the plastic surgeon is involved in early repair of the cleft lip in the first few months of life and works with the dentist and orthodontist to manage appliances to close the cleft in the secondary palate. The surgeon also is involved with surgery to the hands and feet to create digits. Regarding the secondary palate surgery, there are a number of approaches. These include the von Langenbeck repair, the Veau-Wardill-Kilner palatoplasty, two-flap palatoplasty, vomer flap surgery, Z-furlow (Z-plasty), and four-flap palatoplasty surgeries. It is important to note that the researchers report the discussion between two schools of thought on repair timing. One is to repair early in life to accommodate the onset of speech at 1 year of age, vs. delaying repairs to allow for maxillo-facial growth with a complete transverse facial growth at 5 years of age. The current approach stated by the researchers, is to do the soft palate repair at 0;3 to 0;6 and the secondary hard palate by 1;6 CA.

ROLE OF THE DENTIST

Dental treatment is necessary in the case of Apert's syndrome. Per Kaloust, Ishii, and Vargervik (1997) the oral cavity of these children is characterized by supernumary teeth, missing teeth, impaction and crowding, and delayed eruption. The maxilla is affected and the mandible has an abnormal shape and size. The dentist works closely with the

orthodontist to time adjustments to the oral cavity and dentition. The researchers point to two significant findings: a delay of 0.96 years vs. a normal timeline of dentition, as well as a marked slowdown in dental maturity that slows more notably with age. This means that the dentist must time their involvement with the patient along these parameters. They will work on extraction of supernumary teeth and impaction and crowding, but the work is done at different ages as normal children. The dentist also works closely with the orthodontist and plastic surgeon to assess dentition all the while that processes are worked on for closing the alveolar ridge after age 5;0 as well as palatal closure and velopharyngeal port surgeries.

Per Shipster et al (2002), there are a remarkable number of cases that present with Class III malocclusion, specifically Class III incisor relationship, anterior bite and bilateral posterior crossbite. The degree of incisor crowding and irregularity is variable among cases. The dentist considers how difficult it is for the child to maintain good oral hygiene owing both to crowded teeth and to restrictions in fine motor skills. An electric toothbrush may be a useful aid. Frequent appointments with the dentist and/or a dental hygienist are important and, as there is an increased risk of caries, preventive fluoride treatment should be given.

ROLE OF THE ORTHODONTIST

Orthodontists play a significant role in the treatment of a child with Apert's syndrome. Per Kuijpers-Jagtman (2006) there are four distinct processes that the orthodontist will participate in up to and beyond the child's twelfth year of life. The orthodontist will confer with the dentist and plastic surgeon that care for a child with Apert's. This is to determine proper timing for the implementation of orthodontic treatment.

The first process begins from 0;0 to 7;0 years, after the initial treatment plan is devised with the craniofacial team. This is the period during which the orthodontist constructs neonatal maxillary orthopedics for the child ages infant through elementary school age. The purpose of this device is to bring the maxilla in alignment with the rest of the head, with consideration of the mandible and dentition in the process of the orthopedic treatment. There is debate as to the proper timing for orthopedics and the efficacy of using extra-oral pin-retained appliances versus passive appliances. Per the researchers 54% of craniofacial centers use neonatal maxillary orthopedics.

The second process involves orthodontic treatment of the

deciduous dentition stage, which the researchers state has a direct correlation with the patency of circummaxillary sutures. This occurs in the latter period of 5;0 to 7;0 years. It is significant to reiterate the research of Kaloust, Ishii, and Vargervik (1997), bearing in mind once again that there is a 0.96 year delay in dentition of Apert's vs. normal children's dentition. Treatments are needed for the lack of deciduous dentition in the area of the alveolar cleft, and these treatments may include a face mask to protract growth. Treatment to manage crossbite includes equilibration for occlusal interference.

The third process is in the mixed dentition period in the 9;0 to 11;0 years age range. This is concurrent with alveolar bone grafting, 6 months prior to graft insertion with fixed appliances placed on the maxillary arch. The researchers explain that this eliminates crossbite and other unfavorable consequences of malpositioned incisors, and helps with dental aesthetics.

Eruption of the canine adjacent to the cleft of the secondary palate is of importance as this will control the timing for further orthodontic treatment. The researchers point to evidence that the canines erupt in synchronicity with bone graft placement.

The fourth process is in the permanent dentition stage, anywhere from 10;0 to 13;0 years of age or even older. In this period there is a determination whether orthognathic surgery is indicated. The researchers state that there is a high percentage of patients that require this surgery vs. the general population, but that it is not needed in more than 10% of the Apert's and cleft palate patients. This is particularly relevant in Apert's patients as they have a high incidence of Class III skeletal issues and thus the orthodontist carefully exams the other evidence from the craniofacial team in order to determine candidacy for treatment.

CONCLUSION

There is clearly a need for further and more controlled research on the disciplines involved in the craniofacial teams. There is a need for larger cohorts to gather more data specific to Apert's syndrome as this will give better evidence about treatment efficacy and treatment outcomes. The craniofacial teams should be advised to produce studies that have to do strictly with Apert's syndrome so that the body of research regarding genetics in particular so that models for future treatment can be perfected even more for the benefit of these patients.

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