Cleft Lips and Palates ;The Roles of Specialists

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Abstract:
Apert syndrome is a congenital disorder characterized by craniosynostosis, maxillary hypoplasia, mental retardation, mid-facial malformations, and syndactyly. The developmental disorder is inherited in an autosomal dominant manner, though most cases are sporadic. Approximately 75 percent of Apert's patients suffering from dental anomalies suffer from cleft palate or bifid uvula. Cleft palates are common congenital disorders of the upper mouth, and occur when the palatal plates fails to join together during the second month of fetal development. This research paper focuses on the roles played by the craniofacial team in the management of secondary palates in children with Apert's syndrome.
The consequences of Apert's disorders can be severe, long lasting, and sometimes incomprehensible even to the sufferers. But this study has aptly shown that the suffering can be averted, especially if proper corrective strategies are taken as early as possible. However, these strategies must fundamentally revolve around a multidisciplinary approach to sufferers of Apert's syndrome. This study looks into the roles played by the craniofacial team in the management of Apert-related complications such as Cleft lips and palates. The team is made up of the geneticist, speech language pathologist, audiologist, plastic surgeon, dentist, and orthodontist.
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Tremendous medical achievements have been made over the past few decades in the prevention and management of critical developmental disorders affecting millions of people worldwide. The improved ability to manage these anomalies have been stimulated by a complete metamorphosis in the way medical professionals have been able to conceptualize the developmental deformities. However, developmental anomalies such as Apert's syndrome and Jackson-Weiss syndrome continue to present challenges due to the complexities they present. Apert's syndrome, also known in the medical field as acrocephalosyndactyly, is an extremely rare congenital disorder that was first described in 1906 by Eugene Apert.

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The disorder is characterized by craniosynostosis, maxillary hypoplasia, mental retardation, mid-facial malformations, and syndactyly. Although most cases are sporadic, estimated at one in 200,000 live births, victims continue to inherit the syndrome in an autosomal dominant way.[1]

Many Apert patients have been found with oral cavity deformations exhibited by a diminution in the size of maxilla, especially in the anterioposterior direction. According to Verma & Draznin[2], this reduction has been associated with many dental anomalies such as tooth crowding, impacted teeth, ectopic or delayed eruption of teeth, supernumerary teeth, and thick gingiva. Approximately 75 percent of Apert's patients suffering from dental anomalies suffer from cleft palate or bifid uvula. Indeed, cleft lips and palates are among the most common of birth defects that often leads to serious medical and concurrent speech and language problems if left untreated. However, while the consequences of

cleft lips and palates can be severe and long-lasting, they 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, 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.

Brief Overview of Cleft Palates and Management

According to Fraser[3], cleft palates are common congenital disorders of the upper mouth that have continued to attract the attention of medical professional worldwide. A cleft palate occurs when the palatal plates fails to join together during the second month of fetal development. Separate areas of the face are known to develop individually during the first months of pregnancy. Due to interplay of both genetics and environmental influences, the right and left sections of the palate may fail to join properly, resulting to the formation of cleft palate. Cleft palates come in different variations, from a mere opening situated at the rear of the soft palate to a critical severe separation of the upper mouth. According to Hutchinson[4], cleft palates are known to affect partial or whole sections of the uvula, alveolus, soft palate, and hard palate.

The treatment and management of cleft lips and palates may prove challenging due to their complex and inconsistent nature. However, treatment usually involves performing multi-stage surgeries to close the palate. Most children heal superbly well if the surgeries and accompanying treatment are administered early in life. However, these operations require a multifaceted approach. Proper planning and coordination among health professionals, including the neurosurgeon, craniofacial plastic surgeon, anesthesiologist, maxillofacial oral surgeon, orthodontist, dentist, orthopedist, and an orthopedic surgeon is fundamentally needed to achieve success. In addition, the social worker, psychologist, audiologist and speech language pathologist comes in handy to provide much needed care and support to children with this kind of birth defect, especially during the first twelve years of life. For the purposes of this research paper, there will be a focused discussion on the roles played by the craniofacial team in the management of secondary palates in children with Apert's syndrome. The professionals that will be discussed include: geneticist, speech language pathologist, audiologist, plastic surgeon, dentist and orthodontist.

Role of the Geneticist

Consultation with a geneticist is crucial in order to perform a DNA test for the child suffering from the Apert's complications together with his or her parents. According to Robin, Falk, and Haldeman-Englert[5], the role of a geneticist in the management of the syndrome cannot be underestimated since it is primarily a genetic disorder triggered by the premature fusion of some skull bones, a condition known as craniosynostosis. The skull is prevented from growing normally due to the early fusion, resulting to malformations such as the cleft palate. Scientists believe the Apert syndrome is caused by counterproductive mutations in the FGFR2 gene. A protein released by the gene (fibroblast growth factor receptor 2) causes undeveloped cells to mutate to bone cells during the early phases of embryonic development. Mutations occurring in a specific area of the FGFR2 gene ignite an alteration of the protein, promoting the untimely blending of the bones in the skull, hands, and feet due to prolonged signaling. 3 This is known to cause cleft palate, among other complications.

The role of geneticists in the management of cleft palates becomes clear since all craniofacial dysostosis syndromes, including cleft palates and cleft lips, are inheritable. According to Robin, Falk, and Haldeman-Englert[5], Apert's syndrome is known to be inherited in autosomal dominant fashion, meaning that a copy of the malformed gene in each cell has the ability to cause the disorder. New mutations in the FGFR2 gene in individuals with no known history of the genetic disorder in their families has been blamed for causing almost all the documented cases of Apert's syndrome. However, the condition can be inherited from one generation to the other. Furthermore, Apert's may not have

been detected in a parent until an offspring with more dominant traits of the syndrome is born. It is therefore imperative to conduct DNA testing to equip the parents having such genetic predispositions with vital information that will keep them in the know about the chances of getting a child with Apert-related complications such as cleft palate or webbed feet. 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. Such roles can only be performed by a geneticist.

Role of the Speech Language Pathologist

Therapy and follow-up care of children with cleft palates and other developmental disorders must include the invaluable input of speech language pathologist (SLP). A study conducted by Shipster et al. 6 to analyze the speech and language characteristics on a cohort of ten children with Apert's syndrome came up with useful insights about the disorder. The children were characteristically found with hyponasal resonance due to an under-developed midface, small nose, and excessively long soft palate. As a matter of fact, hypernasal resonance is found in children with cleft palates. In nearly all cases of cleft palates, articulation of speech sounds is often distorted due to the malocclusion and high arched palate. Impaired hearing or a general developmental delay is also known to affect speech and language development. Individuals with Apert's syndrome often require glasses to correct their sight deficiencies – either short or long sightedness.[6]

Based on their examination and standardized testing of the cohort, Shipster et al. [6] came up with specific areas that required therapy. Resonance and voice came up as major issues due to noticeable 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. Testing conducted on the cohort revealed severely impaired receptive and expressive language skills in nearly 50% of the study group. Attention was delayed by 2 to 3 sequential years. The cohort displayed single channel attention control that lagged behind normal children. 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. Phonological problems involved stopping of fricatives and affricates, final consonant deletion, voicing of voiceless consonants, and fronting of velars and palatoalveolar sounds.

All the above shortcomings on children with Apert-related complications such as cleft palates call for major intervention on the part of the SLP. Professionals in this field must administer nasality and voice therapy to children with these defects before and after corrective surgery is made. The SLP must also actively assist victims to strengthen their receptive abilities. Due to phonetics and phonological difficulties, therapy must always focus on eye contact, pragmatics and posture issues. The therapist must also be in the forefront in teaching the children how to strengthen their muscles after surgery of the palate. To be successful, individualized plan of intervention must be sought.

Role of the Audiologist

According to Rajenderkumar, Bamiou and Simimanna[7], 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 make reference 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 since they often fall out of place.

Hearing impairment is known to affect speech and language development. It is therefore imperative to provide individuals suffering from this kind of impairment with hearing aids to avoid further deterioration. From a cohort of seventy cases, Rajenderkumar, Bamiou, and Simimanna[7] found out 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. Hearing impairments that can be successfully monitored and managed by the audiologist (in consultation with the 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 should be done according to schedule in order to assess ongoing changes in the child's hearing process.

Role of the Plastic Surgeon

According to Xiao et al.[1], the physical characteristics of Apert-related disorders include defects of the skull, eyes, and face. The skull may be short from back to front, and wide on the sides. The eyes may bulge outwards, with eyelids tilting abnormally downwards at the sides. The mid-face has a characteristic sunken-in appearance, with the upper jaw sloping backwards and the lower teeth projecting precariously in front of the upper teeth. The plastic surgeon comes in handy in such situations as early corrective surgery will definitely relieve 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 cleft palates and other Apert-related syndromes, a significant cosmetic and functional improvement is possible with the help of a plastic surgeon. Such an improvement decreases the risk of optic difficulties or blindness caused by orbital hypoplasia.

Paravatty et al. [8] states that plastic surgery procedures include release of the prematurely fused sutures. The conventional 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 can be performed in accordance to a staged treatment plan. To give the brain some room for growth 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. In the case of webbed fingers, the plastic surgery has to be initiated early in life to allow the child develop a grip.

According to Sadove, Van Aalst, and Culp [9] the plastic surgeon is involved in early repair of the cleft palate in the first few months of life. The surgeon should work in conjunction with the dentist and orthodontist to manage the appliances used to close the cleft in the secondary palate. There are a number of surgical approaches that can be used to correct secondary palate. 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. After such surgery, the child will require training under the supervision of a speech language pathologist to assist in the development of tone and articulation related to muscle strength in the speech mechanism.

The researchers compared two schools of thoughts on the timing of cleft palate repair. One seeks to repair the deformity early in life before the onset of speech development which usually occurs within the first year of life. The other school of thought revolves around delaying palate repairs to allow for maxillo-facial growth and complete transverse growth, which normally occurs at the age of 5 years. According to the researchers, the current surgical approach for soft palate repair is usually performed when the child is between the age of three and six months, with the secondary hard palate following at one-and-half years of age.

Role of the Dentist

Dental treatment is necessary in the case of Apert-related malformations such as cleft lip and palate. Kaloust, Ishii, and Vargervik [10] stated that the oral cavity of children suffering from these disorders 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 must work 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, impaction and crowding, but only at the right age in the normal development of children. The dentist should also work in conjunction with the orthodontist and plastic surgeon to assess dentition and other processes administered to close the alveolar ridge, usually at the age of five years. Such processes include palatal closure and velopharngeal port surgeries.

Shipster et al.[6] postulates that 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 varies among cases. The dentist should consider how difficult it is for the child to maintain good oral hygiene due to both crowded teeth and 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. Also, preventive fluoride treatment should be administered due to the risk posed by caries.

Role of the Orthodontist

Orthodontists play a significant role in the treatment of a child with Apert's syndrome. Kuijpers-Jagtman[11] stated the four distinct processes that the orthodontist will participate in up to and beyond the child's twelfth year of life. The orthodontist must work in conjunction with the dentist and plastic surgeon to provide care for a child with Apert's. This is aimed at determining the proper timing for the implementation of orthodontic treatment.

The first process begins from birth up to seven years of age, and follows the initial treatment plan devised by the craniofacial team. This is the period during which the orthodontist constructs neonatal maxillary orthopedics for the infant child through to elementary school age. The purpose of this device is to align the maxilla with the rest of the head, while duly considering 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. Available data reveals that that 54% of craniofacial centers use neonatal maxillary orthopedics.

The second process involves orthodontic treatment of the deciduous dentition stage. The researchers believe this stage has a direct correlation to the potency of circummaxillary sutures. This occurs in the latter period of between five and seven years. Kaloust, Ishii, and Vargervik[10], research had shown that there is a 0.96 year delay in dentition of children suffering from Apert's syndrome compared to normal children's dentition. Treatment is needed for the lack of deciduous dentition in the area of the alveolar cleft, and may include a face mask to protract growth. Equilibration for occlusal interference is best administered to manage crossbite incidences.

The third process should be administered during the mixed dentition phase, which mostly comes between the ages of nine and eleven years. This is set to coincide with alveolar bone grafting, and comes [6] months prior to graft insertion with fixed appliances placed on the maxillary arch. This helps in eliminating crossbite and other unfavorable consequences of malpositioned incisors, while at the same time helping 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 should be administered in the permanent dentition stage, coming anywhere from the age of ten to thirteen years, or even older. In this period, a decision on whether to perform orthognathic surgery is made. Researchers state that there is a high percentage of patients requiring this surgery compared to the general population, though 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 tissues. The orthodontist must therefore carefully examine the other evidence presented by the craniofacial team in order to determine the best treatment strategies.[12]

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 in the forefront in documenting studies that deal strictly with Apert's syndrome for the purposes of developing a body of knowledge that will aptly be used to develop future treatment models that will impact positively on the lives of patients

References:

1-Xiao-Xia, W., Wang X., Yi, B., Zi-Li L., Liang C., Lin Y. (2005). "Internal midface distraction in correction of severe maxillary hypoplasia secondary to cleft lip and palate." Plastic and Reconstructive Surgery, 2005; Vol. 116, No. 1, 51-60.

2- Verma S., Draznin M."Apert Syndrome." Dermatology Online Journal, 2005 Vol. 11, No. 1, p. 15. Retrieved October 8 2009; <http://dermatology.cdlib.org/111/case_reports/apert/verma.html

3- Fraser F.C. "The genetics of cleft lip and palate." The American Journal of Human Genetics, 1970;Vol. 22, No. 3, 336-352.

4- Hutchinson, J.M."What was Tad Lincoln's speech problem?" Journal of the Abraham Lincoln Association, 2009 Vol. 30, No. 1. Retrieved October 8 2009; <http://www. historycooperative.org/journals/jala/30.1/hutchinson.html>.

5- Robin N.H., Falk, M.J., Haldeman-Englert C.R. "FGFR-related Craniosynostosis Syndromes" Gene Reviews at Gene Tests: Medical Genetics Information Resource Database.

6- Shipster C., Hearst D., Dockrell J., Kilby E., Hayward R."Speech and language skills and cognitive functioning in children with Apert syndrome: a pilot study." International Journal of Language and Communication Disorders. 2002; 37(3), p. 325-343.

7- Rajenderkumar D., Bamiou D., Sirimanna T. "Management of hearing loss in Apert syndrome." The Journal of Laryngology and Otology, 2005; 119, p. 385-390.

8- Paravatty R., Ahsan A., Sebastian B., Pai K., Dayal P. "Apert syndrome: A case report with discussion of craniofacial features." Quintessence International, 1999; 30(6), p. 423-426.

9- Sadove A., Van Aalst J., Culp J. "Cleft palate repair: art and issues." Clinics in Plastic Surgery, 2004;31, p. 231-241.

10- Kaloust S., Ishii K., Vargervik K. (1997). "Dental development in Apert Syndrome." Cleft Palate-Craniofacial Journal, 1997; 34(2), p. 117-121.

11- Kuijpers-Jagtman A. The orthodontist, an essential partner in CLP treatment. B-ENT, 2006;2(4), p. 57-62.

12- Caranci F., Pezzetti F., Locci P., Becchetti E., Carls F., Avantaggiato A., Becchetti A., Carinci P., Baroni T., Bodo M. "Apert and Crouzon's Syndromes: Clinical Findings, Genes and Extracellular Matrix." The Journal of Craniofacial Surgery, 2005;Vol. 16, No. 3, p. 361-368