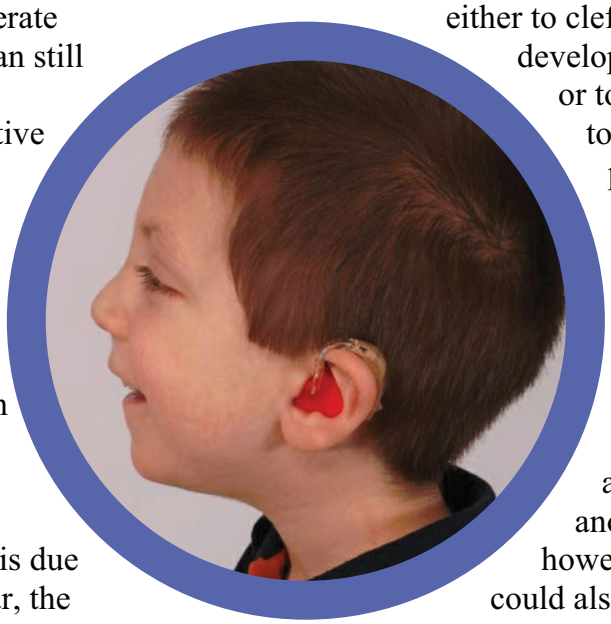
Hearing

Hearing loss is a common finding in Kabuki syndrome and can be of three basic types: conductive, sensorineural or mixed.

Conductive hearing loss occurs when sound is not conducted properly through the outer ear, middle ear, or both, such as in ear canal obstruction or in acute otitis media (ear infection). It is generally a mild to moderate impairment because sound can still be detected by the inner ear. Generally, with pure conductive hearing loss, the quality of hearing (speech discrimination) is good, as long as the sound is amplified loud enough to be easily heard. This type of hearing impairment can often be medically or surgically treated.



Sensorineural hearing loss is due to the damage of the inner ear, the cochlea, or to the impairment of the auditory nerve. It can be mild, moderate, severe, or profound, to the point of total deafness. It is a permanent loss and it doesn't only affect sound intensity such as the ability to hear faint sounds but also makes it more difficult for you to recognize complex sounds, to understand speech and to hear clearly.

Mixed hearing loss - In some cases, such as in complication of recurrent/chronic otitis media, a conductive hearing loss occurs in combination with damage of the inner ear or of the auditory nerve. When this occurs the hearing loss is referred to as a mixed hearing loss.

Conductive hearing loss, mainly due to recurrent otitis media, is reported with a frequency ranging from 24% to 82%. In fact chronic otitis media is extremely frequent in individuals with Kabuki syndrome during childhood. It is probably related either to cleft palate and abnormal development of the Eustachian tube or to immune deficiency. It has to be cured in order to limit permanent hearing loss sequelae (mixed hearing loss).

Sensorineural hearing loss is very rare in Kabuki syndrome. Only a few cases are reported in the literature and are mainly caused by anomalies of the inner ear, however this low prevalence could also be due to incomplete neuro-radiological investigations (CT brain) reported up to now in the medical literature.

Some children may utilize a personal or classroom soundfield FM system, either in conjunction with aids or without. The FM system enhances the distance to noise ratio, in the typical classroom, so that environmental/background noise is decreased while the voice of the speaker is amplified.

Dr. Stefania Barozzi

Dr. Di Berardino

Audiological and Vestibular Findings in Kabuki Syndrome

By S. Barozzi [1], F. Di Berardino [1], A. Selicorni [2]

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Note: *For complete study results see: Audiological and vestibular findings in the Kabuki syndrome - Am J Med Genet A Jan15;149A(2):171-6 2009 Author: Barozzi S, Di Berardino F, Atzeri F, Filipponi E, Cerutti M, Selicorni A, Cesarani A*

In Kabuki syndrome there is a high prevalence of otolaryngologic problems such as ear diseases (92%), hearing loss (82%) and airways problems (58%) only partially due to the prevalence of cleft palate. *See Fig. 1*

Concerning the external ear, which consists of pinna and external auditory meatus, minor anomalies are typical of this syndrome and have been described by most authors. Prominent large and cup-shaped ears are the most common findings (85-100%) and one of the diagnostic criteria of the Kabuki facies. However aural atresia (absence of the pinna), small external ears or preauricular fistula can also be present along with accessory auricular appendages (preauricular pits).

Hearing loss is also a common finding in Kabuki syndrome and can be of three basic types: conductive, sensorineural or mixed.

- *Conductive hearing loss* occurs when sound is not conducted properly through the outer ear, middle ear, or both, such as in ear canal obstruction or in acute otitis media. It is generally a mild to moderate impairment because sound can still be detected by the inner ear. Generally, with pure conductive hearing loss, the quality of hearing (speech discrimination) is good, as long as the sound is amplified loud enough to be easily heard. This type of hearing impairment can often be medically or surgically treated.

- *Sensorineural hearing loss* is due to the damage of the inner ear, the cochlea, or to the impairment of the auditory nerve. It can be mild, moderate, severe, or profound, to the point of total deafness. It is a permanent loss and it doesn't only affect sound intensity such as the ability to hear faint sounds but also makes it more difficult for you to recognise complex sounds, to understand speech and to hear clearly.

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Conductive hearing loss, mainly due to recurrent otitis media, is reported with a frequency ranging from 24% to 82%. In fact chronic otitis media is extremely frequent in patients with Kabuki syndrome during childhood. It is probably related either to cleft palate and abnormal development of the Eustachian tube or to immune deficiency. It has to be cured in order to limit permanent hearing loss sequelae (mixed hearing loss).

Sensorineural hearing loss is very rare in Kabuki syndrome. Only a few cases are reported in the literature and are mainly caused by anomalies of the inner ear, such as bilateral absence of cochlea with dilated dysplastic vestibule and unilateral enlarged vestibule. This low prevalence could also be due to incomplete neuro-radiological investigations (CT brain) reported up to now in the medical literature.

In our study of ten patients affected by Kabuki syndrome, seven males and three females, with ages ranging from 10 to 25 years, only three showed normal hearing. We found that a slight mild or moderate hearing loss was extremely frequent since it was evident in 70% of the affected ears.

In this group of ten subjects, all hearing losses were conductive or mixed. We didn't find any sensorineural hearing loss, thus confirming that it is a rare disorder in Kabuki patients.

Otomicroscopy was mandatory to study the condition of tympanic membrane and chronic otitis media complications. Pure tone audiometry was easily performed in seven patients, while three non-cooperative individuals required behavioural audiometry (audiometry used in young children).

In the ears with hearing loss the most frequent finding was otitis media and its consequences (otitis media with effusion, serous adhesive otitis media, antroatticotomy and tympanomastoidectomy).

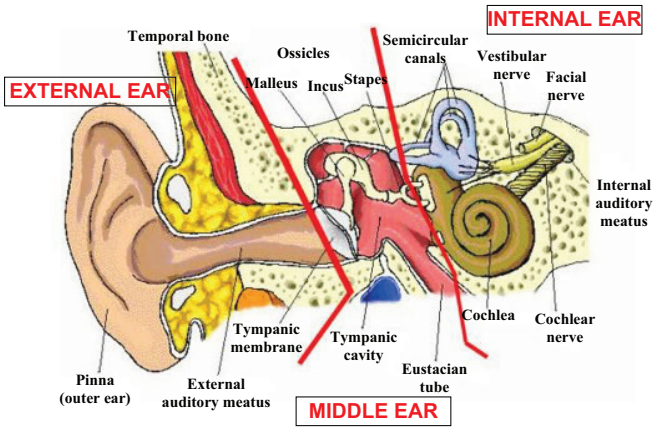


Fig 1

ln Kabuki patients, the frequency of otitis media is likely related both to the high incidence of infections and to the Eustachian tube impairment. In our study, none of the seven patients affected by otitis media and its complications had had a cleft palate. These findings support Peterson-Falzone et al. (1977) who indicated that the prevalence of ear disease in Kabuki syndrome cannot be explained solely on cleft palate and suggested that hearing loss in Kabuki syndrome requires the diagnosis and treatment expertise of audiologist and otolaryngologists.

The hearing loss in the other impaired ears was related to aural atresia in one ear and, in 5 ears, it was associated with a normal otomicroscopy and, in immittance audiometry tympanometry, with the absence of stapedial reflexes suggesting a possible ossicular fixation. As reported in literature, the skeletal anomalies frequently observed in Kabuki patients might also involve the middle ear ossicles with a fixation of the joints. Therefore we suggest to always perform a complete hearing test that includes pure tone and immittance (tympanometry and stapedial reflex determination) audiometry.

In our experience the vestibular evaluation was difficult in the Kabuki subjects since they cannot offer the cooperation needed for caloric examination. Caloric test is used to evaluate the peripheral vestibular function through the irrigation of cold and warm water into the external auditory canal. This test can be carried out exclusively in cooperative patients with no anomalies of the external ears, tympanic membrane perforation, or oto-surgical outcomes. In our patients, caloric tests have been possible only in six subjects.

In the restricted group of patients examined for the vestibular function, 92% showed normal results. In particular, all the ears studied with caloric tests were normoreflective. As the vestibular caloric stimulation was impossible in the patient with aural atresia, the implementation of bone VEMPs was useful, revealing a saccular impairment on the side of the abnormal ear. Vestibular Evoked Myogenic potentials can be used to investigate saccular function, measured from the tonically contracted sternocleidomastoid muscles in response to bone conducted sound stimuli at 70 dB SPL. The saccule is a small labyrinthic sac situated between the cochlea and the semicircular canals. Also this test requires patient cooperation to keep the head elevated in order to contract the sternocleidomastoid muscles.

In view of these findings, it would be advisable to study each patient affected by Kabuki syndrome through audiological examinations and reserve the vestibular assessment for selected patients with vestibular symptoms, with sensorineural hearing loss or inner ear abnormalities.

In conclusion we recommend audiological evaluation in all patients with Kabuki syndrome and vestibular assessment in selected individuals.

Speech

Delay in speech and language acquisition is very common, exacerbated by craniofacial anomalies, hypotonia, and poor coordination.

Articulation errors are common and are likely due to oral-motor hypotonia and general poor coordination. It is not felt that structural abnormalities such as velopharyngeal insufficiency, dental malocclusion and cleft palate are major contributors.

Also common is abnormal oral resonance, again likely due to oral-motor hypotonia and not structural abnormalities. Resonance is the quality of the voice as the result of sound vibrations in the pharnyx (throat), oral cavity (mouth) and nasal cavity (nose).

Abnormal prosody, defined as the the rhythm, stress and intonation of speech, is evident in many children. One study showed that the prosody and articulation errors became more pronounced when spontaneous speech increased in length and complexity. It also found that pitch, loudness and prosody did not mature significantly over time, despite ongoing speech services, resulting in inappropriate and difficult to understand speech production by adolescence. More long term follow-up studies of the distinctive speech patterns of Kabuki Syndrome are needed. This will hopefully lead to better tailoring of speech-language therapies, specific for Kabuki.

Theresa Cinotti

Kim Tillery

The CAPD Model and Kabuki Syndrome

By Theresa Cinotti, M.A., CCC-SLP & Kim Tillery, Ph.D., CCC-A

What is Central Auditory Processing Disorder (CAPD)?

CAPD is not how one hears, but rather "what one does with what they hear". Clients with a CAPD display a wide range of functional behavioral limitations: difficulty understanding or remembering auditory information, weak phonemic skills, intolerance to noise, difficulty understanding speech in background noise, frequently require directions to be repeated, substitute improper speech sounds, display weak reading, spelling, organization and comprehension skills, and often act as if they have a hearing loss.

There are different types of CAPD that dictate specific therapy regimens. Decoding type of CAPD involves a breakdown at the phonemic level where the client struggles in understanding each sound, displays weak reading and spelling skills and requires a long time to respond. A second type of CAPD is known as Tolerance-Fading Memory (TFM) which involves weak short-term memory resulting in poor reading comprehension and weak expressive language skills. Often those with TFM forget the first set of information verses the final set. A third type of CAPD is known as Organization, as weak sequencing and organization abilities are characteristic of this type. A fourth CAPD subtype is Integration, involving poor language and phonemic ability and severe reading and spelling delays.

While an audiologist is the professional who diagnosis the types of CAPD, it is usually the speech-language pathologist who provides therapy and who also evaluates language skills. Most individuals with a CAPD exhibit normal hearing. The etiology is unknown although it is speculated that a history of ear infections and genetic links may be related.

LIZ

Liz was first diagnosed at 14 years of age with Kabuki Syndrome. Currently at age 22 years, she presents with several characteristics related to the syndrome, such as a submucous cleft of the palate, hypotonia, visual perceptual difficulty and mild-to-moderate cognitive challenges. Hearing problems include sensorineural (inner ear) and conductive (middle ear) hearing impairment with recurrent bouts of ear infections. In addition she exhibits speech-language delays and increased nasality of speech.

Liz was first referred for a CAPD evaluation at 16 years of age by a reading specialist as Liz could write the grapheme (letters), but was unable to make the sound-symbol relationship. For example, Liz was able to write her name, but did not understand the relationship of the sounds to the letters, an essential precursor to reading, rhyming and spelling. The reading teacher reported a lack of understanding of left to right scanning of words across the page and also noted that Liz was unable to perform on preschool literacy testing.

The CAPD evaluation indicated two subtypes of CAPD: TFM and Decoding and Liz was referred for CAPD therapy. She received two 50-minute therapy sessions, per week for one and half years, targeting the Decoding CAPD subtype. Therapy consisted of Phonemic Synthesis Training Program (Katz and Fletcher, 1982), Visual-Rhyming Therapy, and general auditory training exercises.

Phonemic Synthesis Training Program consists of 15 lessons to expose the client to the concept of sounds in words by auditorily presenting one sound at a time for which the client is instructed to properly blend the sounds into the target word. For example, the client hears: "b-oa-t" and should respond "boat" without any form of delay or struggle. The goal of this program is to enhance the client's ability to properly perceive sounds in words and utilize that skill in higher level of comprehension, reading and spelling tasks.

Visual-Rhyming Therapy is a technique derived from Soundabet, a training activity in the Processing Power program (Ferre, 1997), which assists the client to recognize sounds and sound patterns represented by all graphemes (letters), thus enhancing rhyming skill. For example the

client is presented a target pattern such as “at” and must rhyme this provided word or nonsense word using all probable consonant sounds. The client would respond with, “bat, dat, fat, gat, hat, jat, kat, lat, mat”, etc. with the visual cue provided in left-to-right format.

b d f g h j k l m

n p r s t v w y z

Upon the success of accurately blending the above consonants with the target pattern, the chart is expanded to include consonant blends, such as br, bl, dr, fl, fr, and st, etc.
This therapy enhances knowledge of left-to-right reading, phonemic and phonological awareness, rhyming, and sound-symbol awareness, again all skills needed for comprehension, reading and spelling. General auditory training exercises were used to supplement the above therapies. Therapies utilized would be considered aural rehabilitation (AR) therapies, although the impact is often seen in language and written language development.

After completing the above therapies, Liz demonstrated progress in the areas of focus. On the Phonemic Synthesis Test (Katz and Fletcher, 1981), a measure of Liz’s sound blending skills, Liz’s progress was follows:

Pre Therapy:
4 accurate responses
1 year later:
19 accurate responses

Lesson 12 of the Phonemic Synthesis Training Program was administered as a baseline measure prior to beginning the entire Phonemic Synthesis Program (lessons 1 through 15). On Lesson 12 Liz performed as follows:

Pre Therapy:
2 accurate responses
3 months later:
21 proper responses
1 year later:
39 proper responses

In August of 2002, at 19 years of age, Liz entered a therapy program which focused on further enhancing auditory decoding and phonological awareness skills while concurrently fostering language abilities, in essence combining aural rehabilitation and language therapy techniques for functional generalization of skills learned.

With this new therapy program, sound blending was a continued focus with sound segmentation added to the challenge. Sound segmenting tasks involve an individual hearing a word, perceiving the sounds in the word, and then being able to identify the sounds individually and in sequence, the inverse of a blending task. For example, if asked to segment the word “tent” the individual would be verbally presented with the word and then required to say the sounds “t-e-n-t”.

Being able to perceive the sounds in a word is a precursor to actual spelling abilities and an aid to fluent reading. As segmenting skills develop, an individual is then challenged to represent sounds with symbols. At first arbitrary symbols such as blocks may be used and, later, the actual graphemes (letters) will be added. As an example, when segmenting the word “ten” an individual may verbally respond “t-e-n” and place three different colored blocks on the table, representing the three different sounds heard. They then could assign letters to correspond to the blocks to actually spell the word. As segmenting skills and sound symbol association skills increase, an individual’s spelling as well as reading skills should subsequently improve. The aforementioned methodology is similar to that advocated in programs such as the Lindamood Phoneme Sequencing Program (Lindamood and Lindamood, 1998), the Phonological Awareness Kit (Robertson and Salter, 1997), and the Ortan Gillingham Program (Institute for Multi-sensory Education), to name a few.

When Liz first began attempting segmenting tasks she required maximal support to separate the sounds in two phoneme (sound) words (ie. no = n-o). As therapy progressed, she was able to consistently identify the sounds in two sound words and also represent the number of sounds heard using arbitrary symbols (colored blocks). Liz continued to progress in segmenting and is currently able to segment four phoneme words using colored blocks and match blocks to appropriate letters with some consistency. Liz is able to match sounds to corresponding consonants approximately 90% of the time with less consistency with matching vowel sounds to letters. However, using this structured system with a speech-language pathologist to guide her through the process, Liz is able to spell two, three, and four sound words with minimal error. Some carryover is seen in spontaneous spelling of words outside of the clinic setting, however, Liz has not fully generalized her skills and continues to work toward independence in this area.

Given that the development of decoding and phonological awareness skills begins in infancy and continues through a child’s school years, LS has made remarkable progress in “catching up” over the last six years of her life to reach a level of phonological processing consistent with early readers. Her most recent testing, using the Phonological Awareness Test (Roberson and Salter, 1997) revealed rhyming skills to be at a 5 year 2 month level and segmentation skills to be at a 5 year, 4 month level. Liz’s ability to isolate sounds in words (determining what sound

was heard at the beginning, middle, or end of the word) was found to be at a 6 year, 0 month level, and her deletion skills(ability to determine what the remaining sounds in a word are when a sound or set of sounds are deleted – say "bat", say "bat" again without the "b") were found to be at a 5 year, 10 month level.

As Liz continues on her journey toward enhanced skills it is a goal to have her consistently make sound symbol associations for functional vocabulary that she will encounter in her environment or during her daily routine. In addition to using decoding therapies to enhance spelling and reading ability, sight word reading is also a focus to enhance comprehension and use of written words pertinent to Liz's vocational, academic, and personal life. Visualization, association, and first letter cuing strategies are currently utilized to develop Liz's recognition of words. Although Liz requires several weeks for the establishment of each new set of sight word vocabulary, this practice has allowed Liz to use, recognize, and read words too complex at this point in her development to sound out independently. Recently, in addition to sight word recognition, common phrases have been targeted for recognition. The goal is to have Liz recognize common phrases from a list of phrases that she will use as a cue to independently create appropriate written language, particularly targeting e-mail communication with friends and relatives.

(L to R)Theresa Cinotti, Liz S, and Dr. Kim Tillery

In addition to written language (spelling, reading, and writing), Liz's understanding and use of language has been targeted through the years. Particularly, Liz has made outstanding progress in compensating for auditory comprehension issues resulting from language delay and hearing loss and compounded by her auditory recall difficulty and perception related to her auditory processing disorder. Liz has developed and frequently utilizes strategies such as attending to visual cues (body language and lip reading), recognizing comprehension breakdowns, and repairing breakdowns through asking for repetition or clarification.

Liz's expressive language has continued to blossom with therapy targeting expansion of simple utterances to form complex. In addition, pragmatic skills, which are interaction abilities have flourished as Liz's practice and maturity have resulted in improved conversational abilities. As language and auditory processing skills have developed, Liz has been able partake in functional activities geared to enhance daily living through improved organization and problem solving. For example, medication recognition and organization, calendar planning, event planning, and situational problem solving and role-playing have contributed to enhancement of Liz's overall independence. Liz has made outstanding progress through the years in all aspects of her communication and overall development. Liz's successes are likely a function of her positive attitude and the outstanding support that she receives from each of her family members. Liz consistently attends and participates in scheduled sessions, and carryover of skills is facilitated by family as her mother regularly attends sessions and continually communicates with Liz's speech-pathologist, audiologist, and ENT to optimize care. Continued success is projected for Liz's future. Liz's story has been shared at numerous conferences and serves as an inspiration to professionals, conveying the message that those with multiple challenges can achieve amazing feats with the appropriate therapies and supports. Central auditory processing therapies have been integral in Liz's skill development, particularly related to her comprehension skills and her reading, writing, and spelling development. The first step in proper treatment planning is appropriate evaluation. Those suspecting an auditory processing disorder, should consult a qualified audiologist with verbal and written language skills assessed by a speech-language pathologist. It has been an honor to work with Liz and her family. They are truly an inspiration to all.

About The Authors

Theresa M. Cinotti, M.A., CCC-SLP
Clinical Assistant Professor at the University at Buffalo

Theresa is currently the Speech-Language Coordinator and one of the clinical supervisors at the University at Buffalo Speech-Language and Hearing Clinic, a training clinic for graduate students pursuing their master's degree in speech-

language pathology. Theresa runs the Intensive Language and Auditory Processing Program at the University, an intensive summer program which addresses the language and auditory processing needs of children ages 5 years and older. In addition, Theresa coordinates the adult language and auditory processing program at the University, a program which focuses on optimizing processing skills and functional communication for adults with auditory processing and related issues.

Kim L. Tillery, Ph.D., CCC-A
Associate Professor and Chairperson of the Speech Department at SUNY of Fredonia

Dr. Kim Tillery has authored one chapter and co-authored four chapters and several peer-reviewed journal publications regarding Auditory Processing Disorders (APDs) and it's relationship with Attention Deficit Hyperactivity Disorders (ADHD). Invited international, national and state presentations include her research of 1) Ritalin's effects on APD, 2) therapeutic measures for Decoding and Integration types of ADP, 3) the co-morbidity of attention, learning and auditory processing deficits, and 4) how reliable differential diagnosis improves effective management of ADHD, LD and APD. Besides her teaching and research Dr. Tillery maintains a private practice, has served as the Co-President of the Speech-Language and Hearing Association of Western NY (SHAWNY) for two-years, received the 2003 SHAWNY Award for her dedication and service to the communicatively disabled of WNY, and serves on other Professional Advisory Boards and Committees.

Visual

More common ocular conditions can include:

- blue sclerae
- strabismus
- coloboma
- ptosis
- micropthalmia

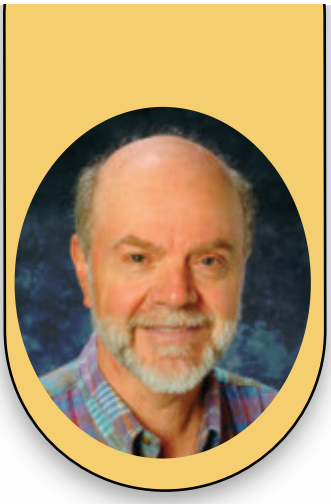
Less common conditions can include:

- nystagmus
- Peters' anomaly
- Marcus Gunn phenomenon
- optic nerve hypoplasia
- obstructed nasolacrimal ducts
- refractive anomalies

Dental

Teeth are often wide spaced, irregularly shaped and/or misaligned. Hypodontia is common, in particular missing upper incisors.

Sensitivity to oral stimulus frequently interferes with proper oral hygiene.



Important Dental and Orthodontic Issues for Children with Kabuki Syndrome

By Bryan J. Williams DDS, MSD, MEd

Pediatric Dentisty and Orthodontics

Oral health is important for all children but is especially important for children with special medical and/or developmental challenges. Children with Kabuki Syndrome have a complex array of special features and functional challenges. Good oral health and proper dental follow up is an important element in the overall care pathway for these children. This paper outlines important issues in dental development, oral health care, facial growth and development and orthodontic care.

Facial Growth Patterns

Children with Kabuki Syndrome have characteristic facial features that have been well documented and described in the literature and this paper will not describe these in detail. As we well know, there is classically some flatness in the cheek areas below the eyes and lack of forward projection of the cheek bones. The lower portion of the face is often disproportionately long compared to typically developing children. This pattern of facial features is rooted in the growth patterns of the jaw structures and the neuromuscular environment. In this paper we will focus on the underlying facial development which has implications for facial pattern, jaw alignment, dental development, and oral health.

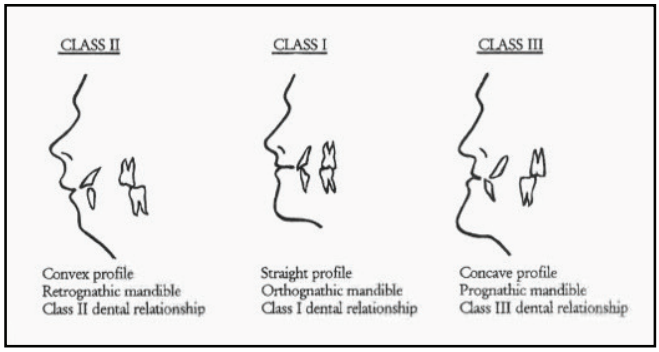


Figure 1: Common Patterns of Facial and Jaw Development

Overall jaw growth and bite relationships are classified into three patterns (Class I, II and III). These patterns result from the relative growth of the two jaws and can be seen in Figure 1. If the upper and lower jaws are in balance this is called a Class I pattern. The bite will be ideal in the molars and the front teeth with the lower front teeth biting slightly behind the upper front teeth. The facial profile will be ideal as well. A Class II pattern occurs when the lower jaw is shorter than the upper which alters the bite on the molars and front teeth. In these children the lower front teeth fit well behind the upper front teeth and commonly these children are said to have an “overbite”. These children have a profile where the chin seems receded and/or the upper front teeth appear to protrude. A Class III pattern occurs when either the upper jaw is too short or the lower jaw is too long or both. In children with a Class III pattern the lower front teeth are in front of the upper front teeth and this is called a crossbite. The facial profile will seem like the chin is protrusive. Jay Leno is a good example of a person with a Class III jaw growth pattern.

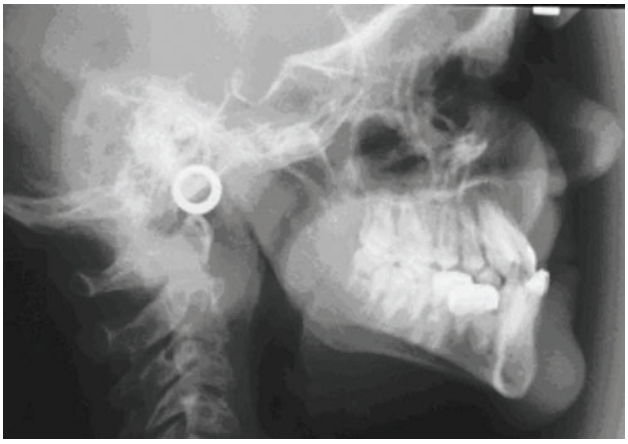


Figure 2: Lateral Jaw Xray of 12 Year Old Male with Kabuki Syndrome

In the Caucasian population Class III patterns are seen in between 1 and 3% of the population. For children with Kabuki Syndrome this is much more common and is the most frequent jaw development pattern. In many children with Kabuki Syndrome this is due to underdevelopment of the upper jaw relative to the lower. Children who have this pattern of jaw growth have less projection in the cheek bone area and the face may appear flatter than ideal in this area. Due to the jaw growth pattern it is also more common to see a crossbite of the front teeth with the lower front teeth in front of the upper. Figures 2 and 3 show a lateral jaw x ray and tracing of a 12 year old male with Kabuki Syndrome. His Class III pattern shows the upper jaw

behind the lower and lower front teeth which are ahead of the upper front teeth. Children with Kabuki Syndrome also often have a tendency to a long lower facial proportion. This relates to a lower jaw which is canted downwards more steeply than ideal. The feeling is that this relates to the neuromuscular pattern where the jaw muscles are more lax allowing the lower jaw to develop at a steeper angle. Figure 4 shows a jaw x ray tracing of a 7 year old girl with Kabuki Syndrome. The lower jaw angle is steep when compared to a tracing of a typically developing child (Figure 5).

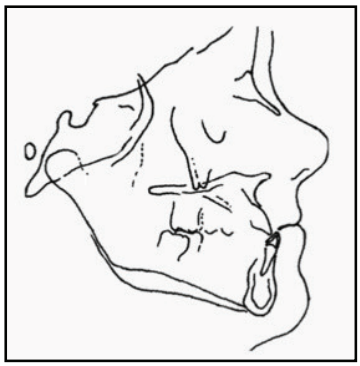


Figure 3: Lateral Jaw X Ray Tracing of 12 Year Old Male with Kabuki Syndrome

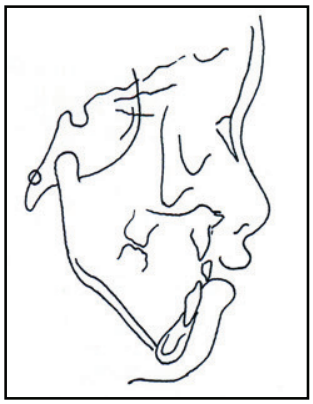


Figure 4: Lateral Jaw X Ray Tracing Showing Steep Angle of Lower Jaw

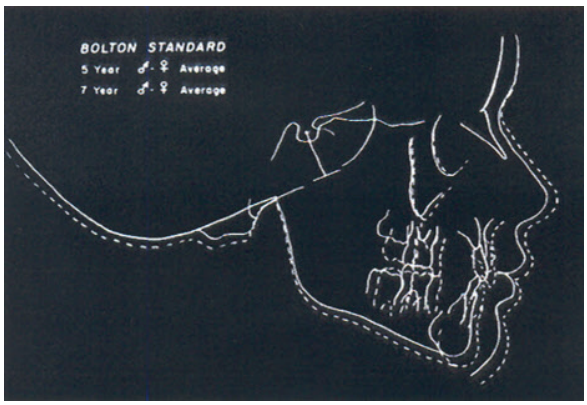


Figure 5: Typical Jaw Development from the Bolton Growth Study - Cleveland OH

The tendencies to have length and angular imbalances in jaw development have significant implications for facial growth, the bite and for potential orthodontic treatment. From the parents perspective it can cause a disconcerting change in the bite as the child matures. Often when the baby teeth are present the bite may look fine to the parent but as the jaw development progresses the bite may become more noticeably irregular. This usually becomes more obvious when the permanent teeth are beginning to erupt. Often this is when the child will be taken for a consultation with an orthodontist. For children with Kabuki syndrome, it is especially important that the orthodontist do a thorough evaluation of the underlying jaw development pattern in advance of initiating any orthodontic treatment. The biting pattern of the teeth is most often a result of the growth pattern of the jaws and not just malposition of the teeth. Treatment options must be carefully assessed in order to optimize outcome. Given the pattern of the neuromuscular environment, children with Kabuki Syndrome may not have the same options for orthodontic treatment as typically developing children where sometimes during growth muscle forces can be harnessed to improve the bite and jaw positions. In children with Kabuki Syndrome with significant jaw length abnormality or vertical jaw imbalance ideal correction may necessitate integrating jaw repositioning surgery into the orthodontic management plan. Obviously careful evaluation of the child's overall medical and developmental status is important prior to considering significant treatment like jaw repositioning surgery.

Other Important Oral Findings

Once there is an understanding of the overall jaw development pattern there are other important oral and dental development issues.

First it is important to realize that over 50% of children with Kabuki Syndrome have some significant cardiac anomaly. For certain dental procedures and with certain types of cardiac issues it will be necessary to provide prophylactic antibiotic coverage in advance of many dental appointments. The American Heart Association has recently revised the guidelines for antibiotic premedication for children with cardiac irregularities (April 2007). It is important that your dentist be familiar with the new guidelines.

The literature shows that high arched palate is common in children with Kabuki syndrome. Cleft Palate occurs in excess of 50% of the children. Cleft Palate has significant implications for breathing, feeding, speech, jaw development and dental development. Optimum management for children with cleft palate requires a coordinated management plan from the time of birth. The management plan should involve a team of specialists who

will provide well coordinated care for all of these significant issues.

In addition to some degree of laxity of the muscles that position the lower jaw, children with Kabuki Syndrome also have a higher risk of laxity in the ligament and muscular structures that position the temporomandibular joint (TMJ) which is the hinge between the base of the skull and the lower jaw. Although the literature doesn't indicate that a high proportion of the children have problems with dysfunction of the TMJ it is important for the child's dental professional to monitor the function of the joint during routine checkup visits.

Children with Kabuki Syndrome commonly have dental anomalies that can affect the shape, size and number of teeth. The two upper front teeth (central incisors) often have a characteristic shovel shape where the lower edge of the tooth is narrower than the mid portion. This is opposite to the normal shape of this tooth where the lower edge is the widest part of the tooth. This is a dental anomaly that is very rarely found in other children and the presence of shovel shaped central incisors is one diagnostic sign that is helpful in formulation of a diagnosis of Kabuki Syndrome. The dentist can improve the shape of the central incisors with simple cosmetic bonding materials. Children with Kabuki Syndrome often have agenesis or lack of formation of one or more permanent teeth resulting in missing permanent teeth. This most commonly involves the upper lateral incisors which are the teeth next to the big front ones (central incisors). When permanent teeth are missing there are a number of management options that your dentist can consider and discuss with you.

Maintaining Basic Dental Health

For any children with special health needs the maintenance of good dental health is very important. Children with Kabuki Syndrome can have intellectual and behavioral parameters which make dental treatment difficult. For these children it is extremely important to prevent dental disease in order to avoid the need for treatment which could be difficult to accomplish.

In simple terms there are two dental diseases that should concern any parent. One is dental caries or what is commonly referred to as decay or cavities. The second is inflammation or infection of the gum tissue which is periodontal disease. Usually in children severe periodontal disease is uncommon but gingivitis which is an early stage of the disease is much more common. Both cavities and gingivitis have a common cause in that certain types of bacteria in the mouth digest sugar containing foods and secrete acids and toxins which attack the teeth and the gum tissue.

Preventing cavities and gingivitis is therefore relatively straightforward with three key actions by parents and children being important. One is to disrupt the bacteria and food that is left around the teeth by at least twice daily tooth brushing. Also in a child who is cooperative, flossing is of great value in cleaning the areas between the teeth that can't be reached by the toothbrush. Second, teeth can be strengthened by the use of fluoride which hardens the tooth enamel and makes it more resistant to decay. The fluoride can come in many forms including community water fluoridation, fluoride in toothpaste, fluoride supplements by prescription if you live in an area where the water is not fluoridated, or professionally applied fluoride treatments. The third factor in preventing dental disease relates to control of the diet. Sugar containing foods provide food for the bacteria and also if a child snacks frequently (or constantly) there are some natural cavity healing mechanisms in the mouth that don't have a chance to work. Many of the dietary habits that increase a child's risk for cavities also are unhealthy for other concerns like childhood obesity.

Regular ongoing dental care is an important part of good medical care for a child with Kabuki syndrome. The American Academy of Pediatric Dentistry recommends the first dental visit be around one year of age. This provides an opportunity to have a base line evaluation, have your questions addressed and with the dentist develop a long term plan to assure the child will grow with good dental health. In a child with complex long term health and developmental needs it is even more important to get a very solid and early start on good oral health.