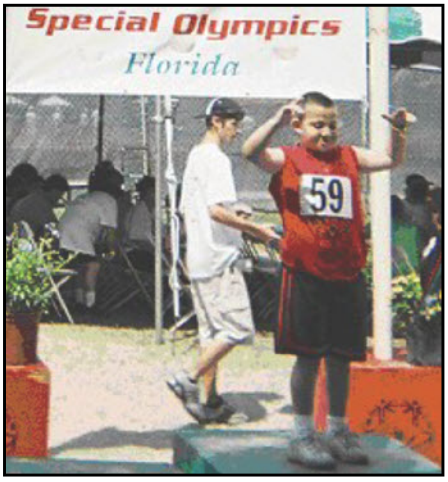
Winter 2007

The Kabuki Journal

Corey Our Little Miracle

By Debra Hixon



Corey on the medal stand January 2006

Corey is our eleven year old son. He was diagnosed with Kabuki syndrome when he was five. Corey was born missing his left ventricle and has had nine open heart surgeries to repair his congenital heart defect. He also had grade 4 bleeds on both sides of his brain. He had a feeding tube until he was five and he still has difficulty swallowing properly. He had cataracts removed from both eyes and has lens implants. His most recent surgery was last year for an undescended testicle.

Although he has gone through so much in his short life, he is a happy and loving young man. He is always concerned about everyone around him and always has a smile on his face. Though he is not very interested in reading or writing, he remembers everything! He is a great speller and he tricks us into thinking he is learning to read because he memorizes the stories we read to him.

Corey has a fourteen year old brother, Tommy, who challenges him to excel in things that he tries. Although they fight like cats and dogs, Tommy really is the best motivator for Corey. Corey wants to do things that Tommy does and usually Tommy takes the time to show him how to do it. Corey is very active in Special Olympics (he is a two-time state medallist in the 25 m dash and the softball throw) and Tommy encourages him to practice and is his biggest cheerleader during the competitions.

Corey is globally delayed in his growth and mental ability and he tends to have a hard time focusing during school. He is on a low dose of Ritalin that seems to help him focus and has reduced some of this frustration. Corey has aspirations of becoming a priest and loves to go to church each week. He has received the sacrament of First Communion and will begin classes for confirmation next year. He has made quite a

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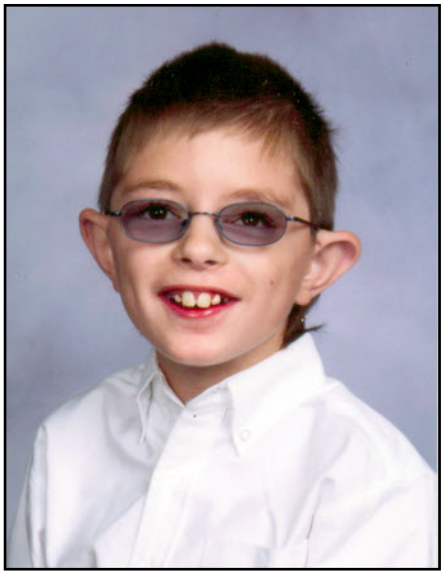
"Open Door"

We hear from...



Dylan (age eight in photo)

Hi my name is Dylan. I am 10 y ears old but I will be 11 in January. I go to school at Central in 4th grade. I did a good job taking my test and got the reward to draw on the overhead projector. I like to write on the overhead but not on the paper. Mrs. Miller and Mrs. Riley help me at school so I can do work. I have to wear my glasses to do my work and they are like sunglasses because last week Last week I sat on my pillow at school because the

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Corey...

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name for himself at our church and has even motivated other "special" students to participate in religious classes.

Corey is also very active in the scouting program and enjoys camping and swimming the best. The other boys in the pack have embraced Corey, even though he drives them crazy sometimes, and always watch out for him during activities. It has been a great experience for him as he has been able to interact with 'normal' children and has learned so much from them about appropriate social interaction.

Corey keeps us very busy going to doctor appointments, sporting events, church activities and school functions, but we don't mind as we know he is our miracle. The doctors didn't give us much hope when he was born. They kept telling us he would be brain damaged and not able to function on his own. Well, as you can tell, they were very wrong! He has been a true blessing to us because he has taught us so much about love and tolerance.

What a great place the world would be if everyone could be as open and loving as Corey!

Message from the Editor

Happy New Year One and All!

2007 brings a time of transition to The Kabuki Journal. Since our first issue in 1999, Mike and Grace Garrett - owners of Best Impression Printing in Merced, CA - have donated all materials and services for the production of our newsletter. Dean Schmiedge's company, SGI (Saskatchewan Government Insurance) has generously volunteered to take up the reins and has jumped right in with this issue. At last, the 'kabuki-journal-retirement' that Mike and Grace so richly deserve! We thank them, from the bottom of our hearts, for their long-standing generosity and dedication. And to Dean and SGI, thanks so much for stepping up! Your contribution, and commitment, to KSN is greatly appreciated.

Hats off to Alana and Dylan for contributing to the "Open Door" column. We're thrilled that our kids, especially, have taken to this idea and get so excited about seeing their thoughts in print. Keep 'em coming. Feel free to include a picture, it's great to see your faces!

Thank you to all our authors for providing such terrific content! In addition to the Journal 'favorites' we all look forward to, this edition includes comprehensive information about hypoglycemia and a study on Central Auditory Processing Disorder featuring Liz S., one of our very own! A very special thank you to Daria Akers for her incredible strength and generosity. Daria, her family, and little Cassie will forever be in our thoughts and our hearts. Be sure to read Daria's very important message, page seven, regarding hypoglycemia.

Until next time, Heather

The Kabuki Journal *is the newsletter of the Kabuki Syndrome Network. The purpose of this newsletter is to provide information and support to individuals with Kabuki syndrome and their families. We will not knowingly print inaccurate or libelous material. We do not promote or recommend any treatment, or professional. Consult with your private physicians / professionals for information and advice regarding medical and therapeutic treatments.*

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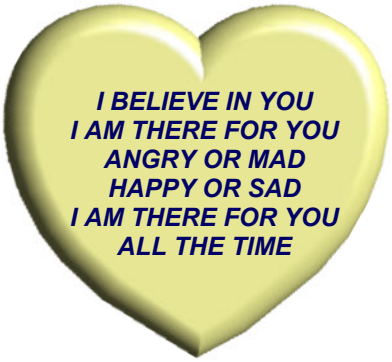
'Open Door'...

Continued from page 1

doctor said I hurt my tail bone when I fell down outside. Mom said I am 4 feet tall and I'm 47 pounds but I'm a big boy. The kids at school play ball with me and I got to play t-ball before school started and I hit the ball and got a medal. The Denver weatherman said it might snow and I want to play in the snow tomorrow. I like to ride my bike and make snowman's. I like Christmas because I have a blow up Frosty the snowman. He goes in the front yard. You can have lots of lights at Christmas and I put lights on my playhouse. I do calendar at school everyday but not on the weekend 'cause there is no school. I like to cuddle with my mom and I sleep at night. On the t.v. I like to watch the Disney Channel, Nickelodeon, and Home and Garden Television. I help vacuum when we clean. I like the little blue vacuum. I have an area to play video games, watch t.v., and get on the computer. I like to type except my little finger is to small to get the last buttons. Duste' is my sister and Drew is my brother. Their dogs died and can't go for a walk anymore. We have a fence so I can't play by the street. The rule is stay in the yard and don't go past the sidewalk. My dog is named Santana but I don't like furry so she needs to lay down I don't like dog kisses. I tell her go sit down over by the kitchen. At school there is a bunny, I did pet him one time. Grandma watches me in the morning and takes me to school. I go to her house when my mom goes to the college to do work in class. My mom works at the Head Start with the little kids. I like to play there. I want to go to school there but mom said I am too big. I like to go into the shed. I have Christmas decorations in there. The Santa is in there and the Christmas tree is in the box. The Doctor in Scottsbluff said I have the Kabuki Syndrome and I am grandmas' little Kabuki boy and so handsome. I listen to the XM radio and like to hear the music. I play on the piano and I want to play on the key board. I got to ride on the float for the parade downtown. I live in my house in Sidney, Nebraska. No kids live here with Kabuki Syndrome. I'm special because I'm the only one here. I have to go now. My lunch box needs fixed with my food and I have to take a bath to wash my body and hair. Mom said have to be clean and not dirty.

Bye – See you.
Dylan



HI MY NAME IS ALANA. I'M A SISTER OF A KABOKI KID NAMED NICHOLAS. NICHOLAS IS 11. I THINK ITS COOL TO HAVE A BROTHER WITH KABOKIE BECAUSE I DEFEND HIM WHEN OTHER KIDS BE MEAN TO HIM. I DID PE TUTORING, AND SO DID NICK, WITH DEVELOPMENTLY DISABLED KIDS. I LOVE KABOKI KIDS, I THINK THAY ARE COOL. MY BROTHER CAN'T HEAR VERY WELL.

Nicholas and Alana

FROM:
ALANA L EDGECOMB
KENT, WASHINGTON - AGE 9

The CAPD Model and Kabuki Syndrome

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 com prehension, 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

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CAPD...

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(Katz and Fletcher, 1981), a measure of Liz's sound blending skills, Liz's progress was as 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.

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

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Theresa Cinotti, Liz S. and Dr. Kim Tillery (left to right)

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. Be- sides her teaching and research Dr. Tillery maintains a private practice, has served as the Co-President of the Speech-Language and Hearing Assoc. of Western NY (SHAWNY) for two-years, received the 2003 SHAWNY Award for her dedication and service to the communicatively disabled of WYN, and serves on other Professional Advisory Boards and Committees.

The KSN logo in our nameplate is the Japanese character 'K' representing the word 'Kabuki'. Its beautiful form is especially appropriate as it also illustrates two out-stretched arms, symbolic of the invitation to network with one another.

(Logo Design by Jos Vergouwen, of the Dutch Netwerk)

HYPOGLYCEMIA:

The Pressing Need for Increased Awareness and Proper Diagnosis

Information-sharing among Kabuki families, physicians and professionals is a vital key to better understanding and management of KS and its many associated conditions. Daria Akers, a member of KSN, tragically lost her daughter Cassandra Anne following a routine surgery in 2005. It has been determined that Cassandra's death was due to complications of hypoglycemia. Hoping to prevent similar tragedies, Daria has recounted the events surrounding Cassie's death for KSN. The Akers family has our deepest sympathy for the loss of their precious daughter, and our gratitude for their desire to help us all. To address the immediate concerns raised by this issue, Dr. Mark Hannibal has provided information about hypoglycemia and its specific relation to Kabuki. His article contains the varied, often subtle, symptoms of hypoglycemia and outlines the components of an adequate evaluation for individuals with Kabuki syndrome. ~KSN

Daria Akers ~ "My daughter died from undiagnosed hypoglycemia, and this is our story."

On March 28, 2005 my nineteen month old daughter with Kabuki syndrome, Cassandra Anne Akers, went in for routine surgery. She never regained consciousness and eight days later she passed away. Seven months later, at the meeting to discuss her autopsy results, the head of the Pediatric Intensive Care Unit asked us to share our story with all Kabuki Parents. My daughter died from undiagnosed hypoglycemia and this is our story.

Cassie was diagnosed with KS, when she was eleven months old, based on her medical history and distinctive facial features. She had several of the symptoms of KS including immune syndrome problems; the inability to coordinate suck, breathe, and swallow; finger pads; and persistent ear infections. Because Cassie aspirated when feeding and had severe reflux, the doctors decided when she was nine months old that she needed a Nissen Fundoplication and a g-tube for feeding. She was fed on a schedule and throughout the night so that she could get the calories she needed.

We had no reason to believe that Cassie had hypoglycemia. Cassandra had even fasted for surgeries before. This was Cassandra's fourth surgery. The first was for the insertion of her g-tube and the Nissen to stop her from refluxing. Then, shortly after she was diagnosed with KS, she had ear tubes inserted to stop the persistent ear infections. In the January before she passed away she had spinal surgery to release her tethered spinal cord. For each of these surgeries she had at least a five hour fast, without any problems, so we so we never had any idea that she might be hypoglycemic. Cassie's fourth and fatal surgery was to repair a hernia that had developed when her Nissen Fundoplication ruptured. Also, to insert a mediport to make her IVIG treatments, for her low level of IgG, easier.

Cassie's surgery was scheduled for early afternoon. She hadn't had formula since 6:00 p.m. the night before and her last liquid was Pedialyte at 5:00 a.m.. Cassandra's surgery was pushed to later in the afternoon because of an emergency case. Cassie was acting cranky and sleepy but we thought that was because she had missed her morning feeds and she was use to her schedule. They finally took her in at almost 3:00 p.m.. Cassie was transferred to post-op to recover at around 6:00 p.m..

With every other surgery, Cassie would wake up as soon as she heard my voice, but this time it was different. She seemed to be having problems breathing. She kept holding her breath and in order to keep her oxygen levels up I had to hold her head so that her airway was open. Because I had heard of other children with KS having difficulty with their airway after surgery, I requested an anesthesiologist to evaluate her condition.

The doctors and nurses were convinced that Cassie was having a reaction to the narcotics used during her surgery. I tried to explain that she'd had this narcotic before and had never had a reaction like this. As the doctors discussed using a drug that would counteract the narcotic, I looked over to see that Cassie was on an IV drip without any sugar in it. I asked the doctors to take Cassie's blood sugar. At 8:00 p.m., when the first reading was taken, the machine just read "LO". A second reading was taken and it was the same. A blood sample was sent to the lab for a more sensitive reading and Cassie's blood sugar read 28 (normal is 70-100). The doctor began giving her sugar though her IV and when her levels read 78 she began having uncontrollable seizures.

At 3:20 in the morning, after she had been transferred to the Pediatric Intensive Care Unit, a CAT scan showed severe brain swelling. It was decided that Cassandra needed to be put on a respirator, and sedated, in order to stop the seizures. Unfortunately, Cassie never regained consciousness and passed away eight days later.

Why is our story so important that the head of the Pediatric Intensive Care Unit asked us to share what happened to us? Because a simple test could have saved Cassie's life. If Cassie's blood sugar had been monitored during her surgery, the hypoglycemia could have been caught.

Cassie's pediatrician, who sees five other children with KS in her practice, has had all of their files labeled so that their blood sugar will be monitored before and during surgery. This is a simple test and it could save your child's life.

Read the following article, By Dr. Mark Hannibal, for medical information about hypoglycemia and Kabuki syndrome ~ page 8

KS and Hypoglycemia: Symptoms and Diagnosis

By Mark C. Hannibal, M.D.,

Hypoglycemia, or the presence of low blood sugar, usually measured as glucose, is most often reported as a short-lived problem in newborns with Kabuki syndrome. Glucose is the primary fuel source for the brain. If the glucose levels remain low, below an accepted "safe" level of about 60 mg/dl, it may expose a child to the risk of brain injury. Some individuals with Kabuki syndrome, however have persistent hypoglycemia throughout infancy and childhood. The underlying cause has not been found in most cases—most likely because it has not been studied well and reported in the medical literature. There are case reports suggesting some may release too much insulin—a hormone made in the pancreas that helps to lower and regulate blood sugar levels. There have also been cases reported of other hormone deficiencies that to some degree regulate blood sugar levels. These include growth hormone, adrenocorticotropic hormone (ACTH) and cortisol. There may also be other undescribed biochemical or metabolic reasons for some children with Kabuki syndrome to have hypoglycemia.

The behavioral symptoms of hypoglycemia, which overlap with non-specific normal behaviors, can be easily overlooked. Most children experience some of the following symptoms. Infants with low blood glucose may have, low tone or floppiness (hypotonia), poor feeding, seizures, and pauses in breathing (apnea). In older children, symptoms may include sudden irritability, hunger, nervousness, shakiness, perspiration, dizziness, light-headedness, sleepiness, confusion, difficulty speaking, or feeling anxious or weak. Suggestions that low blood sugar may occur at night, while sleeping, include crying out or having nightmares, or finding pajamas or sheets that are damp from sweating. Children may be tired, irritable, or confused when they wake up. As you can see, these symptoms are common childhood behaviors, so there is some intuition required to feel that your child "just isn't right" and request that your physician begin to explore if low blood sugar is a possible cause. A simple test parents can do to confirm their suspicions is to see if the symptom is relieved by providing a source of simple sugars, such as a half cup of fruit juice, sugar candies (~eight lifesavers), a quarter cup of raisins, etc. The symptoms should resolve within ten to twenty minutes after eating if it is due to low blood glucose.

What is done for hypoglycemia? In most cases, a pediatric endocrinologist should help with diagnosis and management. First—how often does it occur? The amount of time that the patient's blood sugar is low is determined—often while a child is hospitalized, but it may be initiated with home blood glucose testing. At home, blood sugar may be monitored with a blood glucose meter identical to the device that an individual with diabetes mellitus would use. Treatment is tailored to the severity of hypoglycemia and its cause. Specific recommendations are impossible to make at this time, because no one common cause has been found for hypoglycemia in Kabuki syndrome. Minimally, simultaneous measurement of blood glucose and insulin levels should be performed. Usually, if a person has low blood glucose, insulin secretion is suppressed and measures very low. Measurement of the amount of glucose needed to keep blood sugars in the normal range should be done (determined by milligrams of glucose needed per kilogram of patient per minute). Additional testing to assist in the diagnosis of a cause of hypoglycemia may include measuring free fatty acids, lactic acid and ketone bodies in the blood as well as ketone bodies in the urine during an episode of hypoglycemia. Beyond these tests, evaluation and interpretation of results becomes much more complex, with measurement of other hormones, organic acids or acylcarnitines. A glucagon stimulation test may be necessary. For unusual cases, the best evaluation is probably achieved by an endocrinologist working in conjunction with a biochemical genetics specialist.

Treatment is directed at making sure sufficient food is provided to prevent low blood glucose. Frequent feedings and avoidance of prolonged fasting may be necessary, but some individuals have required continuous drip feeds through a feeding tube or modified formulas and supplements. Stressful situations, such as illnesses, even minor viral infections, may make management of hypoglycemia difficult. In the case of prolonged inability to take food or to keep food down because of vomiting, other methods of maintaining blood sugar levels are necessary. An intravenous (IV) line, placed in a vein, can provide glucose with IV fluid. The amount of

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Diagnosis/Symptoms...

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glucose and rate of fluid provided will need to be individualized for each patient depending on the assessment of their clinical condition.

There are few reports of treatment of hypoglycemia in patients with Kabuki syndrome beyond the need for frequent feedings. Perhaps some have been treated with medications that are used for some other causes of hypoglycemia, such as producing too much insulin. One such drug, Diazoxide (trade name Hyperstat or Proglycem in the USA), may be used in combination with other drugs to keep the blood sugar in a safe range. Clearly, further reports of management of hypoglycemia in Kabuki syndrome are needed. Since it is a rare complication of a not-so-rare genetic syndrome, no one physician will have much experience. The Kabuki Syndrome Network can serve as a clearinghouse to connect family and physicians so that there is increased awareness of the potential for hypoglycemia.

I would be interested to hear about the experience of families, because this is clearly an area of management that requires more study.

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Update: July 2005 - January 2007

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