Spring 2011

The Kabuki Journal

Meet Brooklyn Kay Barrett

by Nikki Barrett

Brooklyn Kay Barrett was born at 12:01 am on May 14, 2008. Our dreams came true as she entered her brand new world already surrounded by love and the eager arms of me and my husband Matt. We were a bit on edge before her birth due to an ultrasound that showed a multicystic dysplastic kidney. Although we were completely terrified of this news, Brooklyn progressed normally in utero and we counted down the days to meet her. After a very easy, short labor, our little girl was finally here.

As soon as Brooklyn was delivered, doctors knew instantly there was something "abnormal" about her. She scored fine on the Apgar test, but it was very apparent that both hips were dislocated and her fingers were clinched in a strange position. Her skin was also very loose and lax, but she cried and ate normally. We were absolutely ecstatic with the arrival or our new addition. Daddy's little girl already had melted both our hearts; our world seemed complete on that day.

The following day (just hours after birth) our entire fantasy land was flipped completely upside down. While in the nursery, Brooklyn had spit up while lying on her back and did nothing to "protect" herself. She stopped breathing for a short time and was quickly transferred to a higher level NICU; and so the search for a diagnosis begins!

While in the NICU, we visited with countless specialists and therapists, all trying to figure out what was causing all of Brooklyn "anomalies." Her loose skin, combined with cupped ears, clinched fingers, dislocated hips, wide set eyes, interrupted eyebrows, pointy teeth, a sacral dimple, thick finger and toe pads, a low nose bridge, and one "good" kidney, all had the experts stumped. Brooklyn stayed in the NICU for one week and progressed each day. She was fitted with braces for her hands to straighten her fingers out, as well as a Pavlik harness for her hips. She was eating well and never cried; unless it was bath time!

Brooklyn Barrett

We were discharged from the hospital on May 21st and could not be happier to bring our little girl home! Matt and I were apprehensive about the days to come with all of the follow up appointments that were already scheduled; but we had no idea that the journey ahead would lead to tears, joy, pride, and an intense bond between the three of us as a team.

Brooklyn was starting her ride through mazes of hospital hallways, x-ray rooms, surgery prep areas, and scary blood draw stations. She would endure her first hospital slumber parties with mommy and daddy, something that most children never have to experience in their lifetime. She would endure countless needle pokes as, what seemed like, liters of blood were constantly drawn in an effort to find a diagnosis. She would endure painful hip manipulations from a doctor that we wish we would've never been put into contact with. She would endure all day trips to the hospital for renal scans; all of this in less than three years of life. Our Brooklyn would endure all of this with the strength of a superhero, and the grace of an angel, all while maintaining the innocence and joy of a child.

Continued...page 2

Brooklyn Barrett...

Continued from page 1

Throughout the marathon of appointments in the months ahead, we discovered a plethora of information about Brooklyn's "anomalies" and how to treat them. We spent endless days in waiting rooms and exam rooms anticipating the next step we were to take in each area. We grew more and more frustrated with each appointment. Not because of the time it took to drive an hour and half to each one. And not even because of the days we were forced to take off work so we could be there together, as a team, at each appointment; but simply because no one had the real answer to the one real question. "Why?" Not "Why Brooklyn?" Or "Why Us?" But "Why are all of these things occurring together?" "What is the cause?"

For a year and a half we visited the same geneticist who called it a "connective tissue disorder," possibly Cutis Laxa. After eternal blood draws that were sent all over the country, we got a second opinion at Children's Mercy Hospital in Kansas City where the rest of Brooklyn's specialists were. After just one appointment and one look at Brooklyn, Dr. Artinger concluded that it was Kabuki Syndrome. A year and a half of wondering why no one could figure Brooklyn out, and after just one hour in an exam room with the right geneticist, we finally had our answer to "the" question. But then came the flood of mixed emotions. The thrill of finally having an answer combined with the reality that this is not something Brooklyn will grow out of. This is not something that is curable or something that is going to just go away.

The Barrett Family

After a short pity session for ourselves, my husband Matt and I came to one conclusion, we so desperately wanted an answer and we finally got it. Brooklyn would never think of feeling sorry for herself, ever. So, we move forward with researching what we can about Brooklyn's syndrome; being proactive in the treatments she needs. The pity turned back to the pride we had had all along. We would not only discover so much about her new diagnosis, but we came to realize that we are part of an absolutely amazing and courageous family. We could not be more proud to be parents of a child with Kabuki. We are honored that we were chosen as Brooklyn's parents.

Of course every parent dreams of having "healthy, typical" children. But parents never think about the blessing of having a child that is unlike most. No one thinks about the thrill that a child with special needs can bring. Our view on life instantly changed after Brooklyn was born, it's amazing what you can take for granted in life. After Brooklyn we realized what really matters in life. It's not fulfilling the usual dream that everyone else wants, the usual family, in the usual home, doing the usual things. It's making magic happen with the unusual. It's making dreams come true with unusual circumstances and wrapping your heart around the people who look up to you. It's

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Continued...page 3

loving your place in life and being proud that you have been chosen to cherish a child with special needs.

Among the dozens of appointments Brooklyn has had in her few years of life, we have come to just as many conclusions. All of Brooklyn's major organs are in good condition except for a slight heart murmur. Her multicystic kidney is dissolving like it should be; what a relief! Her one good kidney continues to function well, which we are truly thankful for!

Brooklyn's most troublesome issue thus far has been her bilateral hip dysplasia. She was born with both hips dislocated and, although the Pavlik Harness has a 90% success rate, it failed to work for Brooklyn. (This came as no surprise to us. It seemed every appointment led to a doctor saying "I've never seen this before. This treatment works for most kids.")

We then switched to a Rhino Harness which also failed to correct Brooklyn's hips. At just 6 months old, Brooklyn underwent her first hip surgery on her right hip. She was put in a plaster body cast for 6 weeks, then switched to a fiberglass cast for an additional 6 weeks. The surgery was minimally invasive and involved slight tweaking of her ligaments. To our dismay, the surgery was not successful and at 15 months**,** Brooklyn had a capsular tightening done on her right hip and was put into another body cast for 6 weeks. After immediate bracing following the cast removal, again, her hips continued to pop in and out of the sockets regularly. In July of 2010 at 26 months, Brooklyn had her first surgery on her left hip; a reconstruction with insertion of a rod and pins.

Brooklyn in her hip harness

So far, her left hip has held. On April 19, 2011 Brooklyn is scheduled for her 4th surgery, a reconstruction on the right side. We are keeping our fingers crossed that her hips cooperate this time around. Her surgeon has done all that she can with her hips up to this point. We're not sure where we'd go from here if the hip gods fail us yet again!

Brooklyn has her quirks, her textures she hates, and her foods she refuses. But she also has a smile that is absolutely contagious, a toughness that is incomprehensible, and an excitement and joy that absolutely radiates through her eyes. Like every other child with Kabuki, Brooklyn continues to amaze us each and every day. She continues to reach her own milestones and is always so proud of her accomplishments. Her heart melting smile is completely infectious, and her spunky personality (although exhausting at times!) is absolutely magnetic. She is a true inspiration. She is our angel. She is our superhero.

Update from Margot

After a long and snowy winter, we are finally seeing signs of spring here in Saskatchewan. With spring comes thoughts of renewal, and that is exactly what's happening at Kabuki Syndrome Network! KSN will launch its new website sometime this spring. It has a brand new fresh look and will include many new features that modern sites are now able to display. We will continue to add to the site's capability over the next year or so.

One of the site's capabilities will be to generate E-newsletters. We have given it much consideration and believe this is a cost-effective choice. As a result, this newsletter will be our last hard-copy version. With our membership's support, we will continue to work hard to produce quality newsletters. We look forward to this new endeavor!

Although KSN has come a long way since its beginnings in 1997, there is still much to be accomplished If anyone is interested in setting up a foundation to aid in research or organizing a national conference, please contact us. We would love to hear your ideas!

My Kabuki Story

by
Crystal Daley

My name is Crystal Daley. I was born on May 12, 1992. I have Kabuki Syndrome and I also have a moderate hearing loss. When I was born, I had a cleft palate. I have had lots of different surgeries growing up. Most of my surgeries that I had were at B.C. Children's Hospital and I'm happy to say that, I've had my last surgery in September of 2010 at Vancouver General Hospital and I'm hoping that I don't have to have anymore. After all, I've had twenty different surgeries. Like my mom always says to me now I could teach those nurses a few things.

Growing up was always hard for me. I have had many struggles while growing up. School was always hard for me, but I've always tried very hard at everything in school whether I was good at it or not. When I got to high school things got better for me. My teachers made the school work easier, so that it was at my level which made school better for me. I've always enjoyed learning in school.

I got to say high school made me look at life different. There were days where I didn't want to be me because I was ashamed of my Kabuki Syndrome and I didn't want other people to know about it. I'd rather just let them know that I have a hearing loss.

I'm very thankful that I have so many family and friends who are always very supportive of me. There are so many different things that I really enjoy like horseback riding, curling, photography, writing and many other things. I've been horseback riding since I was four years old. I stopped horseback riding to try new things but I didn't enjoy those other stuff as much as I enjoyed horseback riding so I went back to horseback riding a few years later. I ride at Valley Therapeutic Equestrian Association. I'm also in Special Olympics I do curling in the fall and winter. I also do bocce in the spring and summer.

Other things that I like to do in my spare time are photography and writing. I took a photography class in high school for grade 11 and grade 12. I learned so much from it and I love taking pictures with my camera all the time. I also like to try and take some different photographs with my camera.

Writing has always been one of my favorite things that I do in my spare time or whenever I feel like sitting down and writing something or going onto my computer and typing something. One of my goals was to write a book start to finish and I did achieve that goal about a year ago now because I've written tons of different things over the years, but I've never been able to keep going on it and finishing it. I would just forget about it and just start something new. I've also started writing my next book, but this time I'm going to take my time on it, because I would really like to try and get my first book published. Writing has always been a big part of me. I'm always writing something whether it's about how I'm feeling or if I have a thought that I can't forget.

Crystal Daley

For me I want to get out there to other people by writing stuff that other people can relate to. That's really important to me if there's people out there that don't like what I have to say or write about that's fine with me. I just want to get out there so people know who I am and hopefully I might inspire someone to get themselves out there just like what I'm trying to do. If I'm not able to get my book out there that's fine with me no one can stop me, I'll just keep writing even if I don't get myself out to other people.

Just because I have a disability that doesn't stop me. I would love to show other people who do have a disability that we can also try to achieve our goals and set new ones even if we have a harder time then other people do. For me just because I have a disability, that doesn't mean that I'm going to give up or stop what I love doing. It just means that I'm going to work hard and not give up. It's going to make me work harder until, I can reach my goal and then start my next goal.

There are days where I feel like I'm not getting anywhere with what I'm doing or with anyone, but deep down inside I know that things will happen for me but maybe not yet. That's what keeps me going and I also have a lot of a amazing people that have helped me along the way. I also have people who are always there for me when I need someone. If I didn't have people like that, then I wouldn't be where I am today and I wouldn't be very successful.

Music Soothes the Soul

As appeared in the Charlotte Observer on Feb 26, 2011

(Note: Cooper is a young boy with Kabuki Syndrome)

Hailey's songs help special-needs children learn, remember and engage.

Hailey Douse is teaching a 5-year-old girl to spell her name, but instead of talking to her, Hailey strums her guitar and sings.

For 30 minutes, Hailey sings her way through a lesson plan, using music to reach out to the child, who doesn't talk much, and to another student who doesn't talk at all. Hailey is a music therapist and uses her voice, guitar, piano and other instruments to help children with autism and learning disabilities.

Music therapy is also used to treat people with Alzheimer's disease and brain injuries. Though it's not as well-known as other therapies, its popularity is growing. The rehabilitation center where Arizona Rep. Gabrielle Gifford's is being treated uses music therapy to stimulate patients' brains. In the new film "The Music Never Stopped," Julia Ormond plays a music therapist who discovers that a young man re-engages with the world when he listens to rock music. And in Jodi Picoult's newest book, "Sing You Home," the main character is a music therapist.

"Some kids with learning disabilities and developmental disabilities can't remember all the steps, such as going to the bathroom, brushing their teeth, yet they'll remember them if there's a song," Hailey said. "It helps them remember the order of things. We learn our ABCs through songs. I do color songs, counting songs."

The healing power of music was recognized long ago by the early philosophers. During World War I and II hospitals hired musicians to play for wounded soldiers, and the first music therapy degree program opened in 1944 at Michigan State University.

"I define music therapy as a health profession that uses music as a treatment tool to address nonmusical goals," said Al Bumanis of the American Music Therapy Association in Maryland. "The trend has been fairly steady that music therapy is being recognized more and more, and more and more third-party insurance reimbursement is occurring."

Hailey enrolled at Winthrop University, intending to get a degree in music education. But she quickly realized she wanted something different. A teacher told her about music therapy.

"It sounded like something that I would love to do," Hailey said. "I've always loved music, and I just really like working with people. I was thinking about switching to psychology, and I saw that this did both things, the music and the mind."

She got her degree in music therapy from Charleston Southern University in May 2009. During a college internship, she worked with CarePartners Hospice and Palliative Care in Asheville and sang hymns to calm a patient in distress because of pain. She said she coaxed a 101-year-old woman who hadn't played piano in years to play a keyboard.

"Sometimes I would visit people who were non responsive," she said. "If they were breathing really hard, I would bring my guitar and play at the same pace they were breathing. Gradually I would work down to a more relaxed state and breathing would calm down."

Whether someone is young or old, Hailey said, music reaches them in ways more traditional therapy cannot and through the senses - hearing, seeing, touching, feeling. Studies show music can reduce blood pressure, heart rate and respiratory rate.

Hailey now works for The Cyzner Institute, a therapeutic school for children through sixth grade. She said she coordinates her therapy plans with the school's speech and occupational therapists.

"It really gives the children a different kind of opportunity to work on the same skills they're working on in the classroom, but in a much more creative way," said Lisa Cyzner, director of the school. "Many of our children are very calmed by the music. There are children who will do things in music who won't do them in any other venue."

Hailey sits on a multicolored rug, strumming her guitar and singing to two boys.

"I'm feeling good. I'm feeling bad. I'm feeling happy. I'm feeling sad." "Good, bad, happy, sad." She smiles when she sings "happy," and frowns at "sad." "Cooper, how do you feel?" she sings to one boy. "Good!"

Continued on page 6

Next she sings a song about February. And then she gets the boys to clap and stomp while she sings about clapping and stomping.

Hailey tailors her techniques to the children. "If a class comes in raucously, I'm not going to immediately play calming music," she said. "I will meet them at their boisterous level with an ... activity or song where it's okay to be loud, and gradually bring them down to a 'calmer' state of mind."

Hailey has a beautiful smile and a beautiful voice, and the children obviously adore her. One boy has a tough time not wrapping his arms around her every few minutes in a big hug.

"The kids ask for her,"Cyzner said. "She's the superstar. Music is the universal language. It creates meaning for these children that we don't even know."

Thirty minutes passes quickly and class is over. Another teacher comes to get Cooper. He is reluctant to leave. Hailey hasn't sung 'good-bye' yet. She picks up her guitar.

"It's time to go. Oh, oh, oh", he sings. "Music is over for today. What do we say?" "Good-bye!" Cooper shouts and happily walks out and onto his next class.

Gidgets n Gadgets

AMTA Logo Boogie Bot

A fun way to capture a young child's attention. This wind up toy dances and beats the drum with the AMTA* logo as it spins on his chest. Made of plastic; no batteries required. ***Price $6.00 each; AMTA Member Discounted Price $3.00;***

Quackler

The Quackler is back - by popular demand! This fun noisemaker "quacks" when you shake it. ***Price $5.00 each; AMTA Member Discounted Price $3.00***

Tangle Toys

These little toys are addictive! Unwind and change the multi-colored tangle into many different shapes and back again. Imprinted with the phrase, "Unwind with Music Therapy". Great gifts. ***Price $3.00 each; AMTA Member Discounted Price $2.00;***

Sound-Eaze CD

Written by an occupational therapist and a music therapist as a fun way for parents, teachers and therapists to help children with auditory sensitivity and fear of loud noises. It combines vocals and rhythm with sounds that can be upsetting to many children. The music and rhythm are eased out until only the sound remains. Allowing your child to adjust the volume may give the feeling that he/she is in control of the sound. Sounds included are: baby cry, vacuum cleaner, fireworks, dog bark, siren, phone ring, thunder, fire alarm, blender, hairdryer, lawnmower, toilet flush.

Item # 029634 Catalog Price $17.49 www.abilitations.com

Craniosacral Therapy

By Josh Chappell

Josh Chappell

Hi, my name is Josh Chappell. I love massage therapy and have been a massage therapist for ten years. For my work, I am privileged to work with people who have different abilities (disabilities). It is so very rewarding being able to help somebody to be able to function and feel better.

I also have a son, Bryce, who was diagnosed with Kabuki Syndrome when he was nine months old. As his father, I have been blessed to be able to work with Bryce and observe first-hand the effects of Craniosacral Therapy (CST) for somebody with Kabuki Syndrome. He has made such great progress and I attribute much of that progress to his receiving CST. This article will detail more of what CST is and how it can help you, your child, or the person you care for with Kabuki Syndrome.

In a nutshell, Craniosacral Therapy is a light touch therapy that releases restrictions and adhesions in the Craniosacral System. The Craniosacral System is intimately related to many systems of the body, including: endocrine, neuromuscular, nervous, immune, lymphatic, and musculoskeletal systems. Because of this relationship, by benefiting the Craniosacral System, these systems are also benefited.

When I first went to school, we students practiced on each other. I distinctly remember learning Craniosacral Therapy and feeling my partner's cheekbones move. This was so cool to me! From this moment, I became extremely intrigued and wanted to know all I could about Craniosacral Therapy. I have since taken and re-taken classes in Craniosacral Therapy from John Upledger, an osteopathic physician who developed this great therapy.

It can be called many different things: CST, Cranial, Craniosacral -- It is a light-touch therapy in which very light pressure is applied to the bones of the skull, spine, sacrum and surrounding tissues, releasing restrictions or adhesions that inhibit the flow of cerebrospinal fluid (CSF). Abnormal pressure is also removed or lessened from the brain. By enhancing flow and releasing adhesions, a better flow of CSF is achieved.

CSF is responsible for cushioning and providing nutrients to our nervous system.

This therapy is very beneficial for: mental disorders (autism, ADD/ADHD, hyperactivity, etc.), seizure disorders, neuromuscular disorders (cerebral palsy, etc.), sensory integration disorders, and head injury. It helps the whole nervous system (brain, spinal cord, sacrum) relax and by so doing, also benefits related systems (emotional, neurological, neuromuscular, immune, endocrine, etc.)

My son Bryce is now 5 and has been given a combination of problems common to Kabuki that most of us are not used to dealing with ourselves. He was born with a cleft palate and a heart defect. He had difficulty nursing or eating by mouth so has eaten formula through a feeding tube all of his life. He has a G-tube in his stomach where we feed him using a feeding pump. He has severe developmental delays and sensory issues (akin to sensory integration disorder), and for most of his life, he has had an aversion to any food or liquids(even candy!).

Bryce Chappell

Bryce has been fun to work with, and one of the most memorable experiences I have working with him is when I did a 10-day Craniosacral Therapy trial on him. I worked on him every day for 10 days in a row, using basic Craniosacral techniques. Imagine my wife, Heather's, and my joy when he began tasting, chewing, and swallowing food!

Little or no gagging, hardly any hesitation. This was after years of not eating anything and always spitting out the food we offered him. It was more than coincidence. It was a miracle.

Through continued perseverance, we are now able to get him to eat enough calories by mouth on most days that we are able to drop one of his four-daily tube feeds and he's still maintaining his weight. (not only that, but he's also getting some of his nutrition from whole foods, rather than scientifically-formulated "semi-foods")

Needless to say, we are very excited and continue to work with him. I haven't been able to do CST as

Continued on page 8

regularly as I would have liked, but the improvement in his eating is still there. He is chewing and swallowing and tasting so the improvement seems to have been maintained by continually offering him food and keeping his mind active. He is more even-tempered and has less severity in his sensory issues. He is also able to focus better and expresses himself better through vocalizing his needs.

Because consistency really helps to "re-pattern" the whole craniosacral system and help the body become used to the improvement as it becomes more normalized, it is best to find a therapist who can work with the person you care for regularly.

Now, I could stop here and you would have a pretty good idea of what CST can do, but I must tell you about another experience we've had with him. Bryce is constantly on the go. He doesn't stay on task for very long, and is always looking for some sort of stimulation or excitement. He would not sit still very easily, but one Sunday before church, I worked on him for about 20 minutes and his behavior improved so much! He seemed much more aware and alert, he sat on our lap through church, and we were absolutely amazed by his progress.

Because I work with many children and adults with different disabilities and varying degrees of severity, I have had the opportunity to use Craniosacral Therapy on many of them. Unanimously between all their parents, they noticed more alertness, better communication, as well as them being more aware of their surroundings as a result of the CST. One teacher even said it helped her student become more "cognitively minded."

From my experience, Craniosacral Therapy has been nothing short of a miracle therapy in helping my son with the symptoms of Kabuki Syndrome. He still has many of them, but with CST as a complimentary therapy, he is learning to overcome many of them.

The symptoms also do not seem as severe. I recommend CST to anybody with a child with Kabuki Syndrome or any other kind of disability.

For questions or comments, Josh can be reached at jc4massage@yahoo.com. Or Go to http://www.upledger.com and click on the "find a practitioner" tab at the top to find a craniosacral therapist in their area.

Mom Receives Award!!

Michele Westmaas, from Pittsfield, IL was recognized March 11 (*Early Hearing Detection and Intervention Day*) for her commitment to serving families with children who have a hearing loss. She received the *Guide By Your Side Parent Achievement Award*, designed to recognize parents of children with hearing loss who go beyond "just being a parent." This year marked the first time an award was given to a parent.

"As they read off the list of parents nominated, many that I know that are really busy involved moms from all around the state, it really hit me this was a big deal. To be recognized among them was really nice," Michele said. "All moms are always trying to do the best for the kids. We don't usually get a pat on the back, so to be recognized like that was really, really nice."

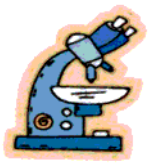
Michele Westmaas

Michele is a trainer/consultant with the Illinois School for the Deaf Outreach and Hearing and Vision Connections. Her daughter Aubrie, 13, was diagnosed at birth with CHARGE Syndrome, a recognizable genetic pattern of birth defects including hearing and vision impairment, poor speech and motor issues.

"For people with disabilities in general, it is so important to be a voice for them, to help parents connect," she said. "It just needs to be done. If we've got energy and the ability, we really need to be a part of doing things."

As reported by; Deborah Gertz Husar for the Herald-Whig

FOOTNOTE: Michele's daughter, Aubrie was originally diagnosed as having CHARGE Syndrome and just recently the diagnosis was change to Kabuki Syndrome.



Update on Genetic Testing For Kabuki Syndrome

In August 2010, researchers at the University of Washington (Ng et. al., 2010) discovered the MLL2 gene mutations responsible for approximately 2/3rd's of the cases of Kabuki Syndrome. As a result of this finding, many laboratories are now offering blood tests to confirm if an individual carries this gene mutation. Some of these labs are also offering parental and prenatal testing as well. The number of labs offering these tests continues to grow. For an updated list of labs offering the test, check The National Center for Biotechnology's listing at: http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/216289?db=genetests

If you are interested getting tested, you can ask your geneticist or another specialist to order the tests, perform the blood work (about 5 cc's of blood draw) and have it sent to one of these labs. Turnaround time to receive results is approximately 6-8 weeks. The test is called MLL2 Full Gene Sequencing and the test code is SMLL2. If applicable, it is advisable to call your insurance company to see if the test will be covered. All insurance companies are different, but many insurers cover this test because it is diagnostic. CPT codes are: 83891 (x 1), 83894 (x 1), 83898 (x 76), 83904 (x 152), 83909 (x 152), 83912 (x 1) and cost is approximately $2,000 to $3,000 US dollars (if not covered by insurance) for the first individual. Once a mutation is identified, other family members can be tested and prenatal testing can be done. Cost for testing another individual for a particular mutation is $390 and cost for prenatal testing for this mutation is $590. In Canada and other countries, the testing may not yet be available, but there have been situations where a geneticist may send the samples to the U.S. for testing.

In considering Full Gene Sequencing testing, it is important to remember that the area of exome micro array analysis and gene sequencing is brand new and therefore information is continually coming available. For instance, while the University of Washington study indicated that MLL2 gene mutations are responsible for approximately 70% of cases, a new study by Milunsky et. al. at Boston University School of Medicine, suggests that the MLL2 mutation may be responsible for a much smaller amount of cases and additional gene(s) yet to be discovered may account for remaining cases.

Given the newness of this research, we should keep in mind that while genetic blood testing is a valuable tool in diagnosis, a clinical diagnosis from a geneticist based on the cardinal features of Kabuki Syndrome (listed in sidebar) should still be a family's first step. Even if an individual blood test comes back negative for an MLL2 mutation, professionals and families might still have reason to believe that the child has Kabuki Syndrome. Also, by opting into a research portion of these tests, individuals are providing samples to help further learning about the syndrome that will benefit the broader Kabuki population.

Finally, while receiving an accurate diagnosis is important, focusing on proactively addressing medical and developmental issues through early intervention and consultation with specialists is of utmost importance in making sure the individual achieves their own best health and highest potential.

CARDINAL FEATURES OF KABUKI SYNDROME:

1. Facial features: long palpebral fissures with eversion of outer third, arched eyebrows with sparse outer half, prominent eyelashes, prominent and/or misshapen ears, and depressed nasal tip
2. Skeletal abnormalities: may include brachydactyly, brachymesophalangy, and clindactyly of the fifth finger, and vertebral anomalies including scoliosis.
3. Dermatoglyphic abnormalities: including persistent finger fetal pads
4. Intellectual disability (mild to moderate)
5. Postnatal short stature

NOTE: In addition to the cardinal features, doctors also use some co-existing medical conditions to support a diagnosis. For a list of some of the more common co-existing traits associated with a diagnosis, please visit the Kabuki Syndrome Network website at www.kabukisyndrome.com.

Medical Journal Articles

Updated November - April 2011

Published Articles for May 2011 Newsletter

1. **Strabismus and Poor Stereoacuity Associated with Kabuki Syndrome** – Korean J of Opthamology 2011 Apr;25(2):136-8. Epub 2011 Mar 11 **Authors**: Kim NG, Kim HJ, Hwang JM.
Kabuki syndrome is characterized by long palpebral fissures, large ears, a depressed nasal tip, and skeletal anomalies associated with postnatal dwarfism and mental retardation. There have been few prior detailed descriptions of strabismus or stereopsis in these patients. We report a patient with Kabuki syndrome who showed small-angle strabismus and poor stereopsis. This case illustrates the need for patients with a diagnosis of Kabuki syndrome to have an ophthalmologic evaluation. Strabismus associated with Kabuki syndrome may have a small angle that can be easily overlooked.

2. **Congenital Lymphatic Dysplasia in Kabuki Syndrome: First Report of an Unusual Association** – Lymphology. 2010 Dec;43(4):188-91 **Authors**: Morcaldi G, Boccardo F, Campisi C, Bellini T, Massocco D, Bonioli E.
Kabuki syndrome was first described in Japan in 1981 as a rare disorder of unknown cause. Its main features include characteristic facies, postnatal growth retardation, and mental delay. To date, there is no molecular marker for Kabuki syndrome, which is considered genetically heterogeneous and still is a clinically-based diagnosis. Here we describe the first case of a patient affected by Kabuki syndrome associated with lymphatic dysplasia. We suggest accurate evaluation of all Kabuki patients as early as possible in order to diagnose lymphedema or other clinical manifestations of lymphatic system involvement. Early identification of lymphatic system maldevelopment provides the best chance for reducing the risk of developing progressive lymphedema with associated tissue changes (fibrosis, sclerosis, and fat deposition).

3. **Revisiting Mendelian Disorders Through Exome Sequencing** – Human Genetics 2011 Apr;129(4):351-70. Epub 2011 Feb 18 **Authors**: Ku CS, Naidoo N, Pawitan Y
Over the past several years, more focus has been placed on dissecting the genetic basis of complex diseases and traits through genome-wide association studies. In contrast, Mendelian disorders have received little attention mainly due to the lack of newer and more powerful methods to study these disorders. Linkage studies have previously been the main tool to elucidate the genetics of Mendelian disorders; however, extremely rare disorders or sporadic cases caused by de novo variants are not amendable to this study design. Exome sequencing has now become technically feasible and more cost-effective due to the recent advances in high-throughput sequence capture methods and next-generation sequencing technologies which have offered new opportunities for Mendelian disorder research. Exome sequencing has been swiftly applied to the discovery of new causal variants and candidate genes for a number of Mendelian disorders such as Kabuki syndrome, Miller syndrome and Fowler syndrome. In addition, de novo variants were also identified for sporadic cases, which would have not been possible without exome sequencing. Although exome sequencing has been proven to be a promising approach to study Mendelian disorders, several shortcomings of this method must be noted, such as the inability to capture regulatory or evolutionary conserved sequences in non-coding regions and the incomplete capturing of all exons.

4. **Dilated Vein of Galen in Kabuki Syndrome** – Brain Dev. 2011 Feb 15. [Epub ahead of print] **Authors**: Sánchez-Carpintero R, Herranz A, Reynoso C, Zubieta JL
Kabuki syndrome (KS) comprises multiple congenital abnormalities and is characterized by a peculiar facial appearance, dermatoglyphic anomalies, mental retardation, skeletal abnormalities and postnatal growth retardation. We describe the case of a 23-month-old boy with the typical features of KS who had several malformations in the veins of the brain, which had not previously been described in patients with this syndrome. The MRI phlebogram of this patient showed that the vein of Galen was dilated and that it drained anomalously. The sinus rectus was abnormal and the longitudinal inferior venous sinus was absent. In view of this finding, together with the fact that structural brain abnormalities in KS are more frequent than in other congenital syndromes with multiple malformations, we propose that MRI be used in the diagnostic work-up of all patients with KS.

Continued on page 11

5. **MLL2 Mutation Spectrum in 45 Patients with Kabuki Syndrome** – Hum Mutat. 2011 Feb;32(2):E2018-25. doi: 10.1002/humu.21416. Epub 2010 Dec 7 **Authors**: Paulussen AD, Stegmann AP, Blok MJ, Tserpelis D, Posma-Velter C, Detisch Y, Smeets EE, Wagemans A, Schrander JJ, van den Boogaard MJ, van der Smagt J, van Haeringen A, Stolte-Dijkstra I, Kerstjens-Frederikse WS, Mancini GM, Wessels MW, Hennekam RC, Vreeburg M, Geraedts J, de Ravel T, Fryns JP, Smeets HJ, Devriendt K, Schrander-Stumpel CT

Kabuki Syndrome (KS) is a rare syndrome characterized by intellectual disability and multiple congenital abnormalities, in particular a distinct dysmorphic facial appearance. KS is caused by mutations in the MLL2 gene, encoding an H3K4 histone methyl transferase which acts as an epigenetic transcriptional activator during growth and development. Direct sequencing of all 54 exons of the MLL2 gene in 45 clinically well-defined KS patients identified 34 (75.6%) different mutations. One mutation has been described previously, all others are novel. Clinically, all KS patients were sporadic, and mutations were de novo for all 27 families for which both parents were available. We detected nonsense (n=11), frameshift (n=17), splice site (n=4) and missense (n=2) mutations, predicting a high frequency of absent or non-functional MLL2 protein. Interestingly, both missense mutations located in the C-terminal conserved functional domains of the protein. Phenotypically our study indicated a statistically significant difference in the presence of a distinct facial appearance (p=0.0143) and growth retardation (p=0.0040) when comparing KS patients with an MLL2 mutation compared to patients without a mutation. Our data double the number of MLL2 mutations in KS reported so far and widen the spectrum of MLL2 mutations and disease mechanisms in KS.

6. **Esotropia in Kabuki Syndrome** – J Pediatr Opthalmol Strabismus 2010 May 21;47:e1-3. doi: 10.3928/01913913-20100324-05 **Authors**: Sharma P, Dave V

Kabuki syndrome is a mental retardation-malformation syndrome affecting multiple organ systems. The typical facies resembles the make-up worn in Japanese Kabuki theater. Although there are several clinical findings, the ocular findings affecting vision have been underreported. The current patient shows esotropia and previously unreported nummular corneal opacities.

7. **Kabuki Syndrome and Crohn Disease in a Child with Familial Hypocalciuric Hypercalcemia** – J Pediatr Endocrinol Metab. 2010 Sep;23(9):975-9 **Authors**: Ho J, Fox D, Innes AM, McLeod R, Butzner D, Johnson N, Trevenen C, Kendrick V, Cole DE

An association between Kabuki syndrome and autoimmune disease has been described in the literature, which may explain the connection between Kabuki syndrome and Crohn disease. However, it remains unclear if there is a link between FHH, Kabuki syndrome and Crohn disease in this case.

"My doctor told me to avoid any unnecessary stress, so I didn't open his bill."

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