Evan
By Linda Pollard

Kabuki Syndrome?? What is it and how can my child have it? Those are the questions I asked the geneticist when Evan was diagnosed with Kabuki seven years ago at two years of age. I recall leaving the hospital with three pieces of paper she had copied from her large “syndrome” book that described Kabuki. It was the only information she could offer me. My son had a rare syndrome. I was consumed with feelings of fear, sadness, helplessness and denial. My only comfort was that Evan finally had a diagnosis.

Yearning to know more about Evan’s condition, I began searching the web. There was no Kabuki website at the time, but I did find Margot’s email address. I was relieved, and yet anxious, that I had found another Kabuki parent. I wanted to hear good things, but was fearful of what I might find. I am very thankful that the Kabuki Network was formed and that we, as parents of Kabuki Kids, have the opportunity to share our experiences and support each other. Even though we are from diverse lands and cultures, I feel we are connected through the unique challenges and gifts this rare syndrome brings. We are fortunate that we all share one thing in common – we have very special children.

I cannot envision my child’s future like my neighbour. I cannot sit on the side of the baseball field and cheer as he hits a home run. His abilities are much different from most children, and I have learned through time that what Evan can achieve and bestow upon others is extremely fulfilling and rewarding. Evan has something very special – he has a gift, a gift of pure happiness and love. He has heart of gold and an incredible (some say “old”) soul. This is Evan’s story.

Update from Margot
By Margot Schmiedge

Hello Fellow Members!

Thank you, again, to Jon and Rose Jacobi for hosting their 5th Annual Barbecue this past summer. This was the second time that Dean and I, along with a few kids, were able to attend. It’s so wonderful to meet so many of you! An afternoon is never enough time, but I’m grateful for at least that — and very grateful to the Jacobis for opening their home to so many of us.

In regards to other get-togethers that I’m aware of:

- Kerrin Windsor-Male is hosting a get-together in Australia on November 6th. You can contact her at 08 8338 2520.
- Sally Harren from California has organized several get-togethers. For more information you can contact her at 619-479-9369 or randsharren@cox.net.
- The Dutch Kabuki
Growth, behavior, and clinical findings in 27 patients with Kabuki (Niikawa-Kuroki) syndrome


Summary:
By Margot Schmidge

This study documented the growth patterns, behavior and relationship between intellectual level and head circumference of 27 individuals with Kabuki from Australia and New Zealand. There were 14 female and 13 male clients ranging in age from 7 months to 36 years. Of the 27 subjects: 21 were under 10 years of age, 4 were 10-16 years of age, and 2 were over 16 years of age (one 17 years, one 36 years). In regards to family history, all cases were sporadic.

All children/adults were evaluated and two clinical geneticists confirmed diagnosis. A questionnaire was completed by parents and individuals with Kabuki. Intellectual ability was assessed according to psychomotor development, school performance, ability to read and write, and formal developmental assessment where available. Based on the results, the degree of intellectual disability for each client was classified as mild, moderate, or severe. Body mass index (BMI) was calculated on all patients over the age of 5, using BMI measures standardized for age, to diagnose overweight or obese clients. Results were compared to those typical of the general population, matching both age and sex.

RESULTS

Birth Prevalence: It was calculated that the birth prevalence of children with Kabuki in the state of Victoria in Australia is 1 in 86,000. The authors felt, though, that the lower birth prevalence found in Australia compared to the 1 in 32,000 found in Japan most likely reflects under-recognition of the syndrome in Australia rather than a true difference in prevalence between the two countries.

Pregnancy History: Five

See Growth page 3

Update...
continued from page 1

Kabuki Syndrome Network hosts an Annual Family Day.
You can contact Els Vergouwen at 043-3650207 or martijn95@tip.nl.

In the last newsletter I thanked various people for their contributions to KSN. It wasn’t until it came to print (and not because he made any mention of it) that I realized I completely forgot to thank my own husband. That shows you how much we take each other for granted! Dean is KSN’s accountant, looking after all financial aspects – preparing income statements, balance sheets, etc., etc. Thank you Dean! 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, therapy, institution or professional. Consult with your private physicians/professionals for information and advice regarding medical and therapeutic treatments.

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Information available in Dutch

Thank you Dean!
pregnancies had polyhydramnios (too much amniotic fluid), two had oligohydramnios (too little amniotic fluid), one had increased nuchal translucency (fluid under the fetus’s skin), and two infants had a 2-vessel umbilical cord (vs. the normal 3) – all indicators that there could be something amiss with the fetus/infant.

Facial Phenotype: The Kabuki facial appearance was apparent from infancy and did not change with age.

Weight: Data on 25 individuals showed no birth weight less than the 3rd centile. Failure to thrive in the first year was apparent in 18/27 babies (which was defined by a weight of less than the 3rd centile) and was often associated with feeding problems. A diagnosis of gastro-oesophageal reflux was made in 10/27 babies.

Eight of the 14 children (57%) over the age of 5 had an elevated BMI – 7 in the overweight range and 1 in the obese range. Six out of 8 of these children had failure to thrive in the first year of life. Interestingly, parents reported normal calorie intake and no increase of hunger or eating when full.

The proportion of individuals age 5-10 and 10-17 years who are overweight or obese in the Australian population is 20% and 20-25%, respectively. Comparatively, this study documented a significantly greater (57%) incidence of overweight/obesity among the Kabuki patients.

Data on the rates of obesity in Australian and New Zealand adolescents with intellectual disability was not available. Further studies using age-standardized BMI will help define if elevated BMI and obesity are a distinctive part of the phenotype in middle-childhood/adolescent Kabuki individuals. There was no relationship between degree of intellectual disability and BMI.

Height: Eight of 26 individuals had a height less than the 3rd centile and none had a head circumference above the 50th centile. True short stature and microcephaly are less common than previously reported. The authors felt that it may be more helpful in diagnosing Kabuki to note that height or head circumference over the 50th centile is atypical for the syndrome.

Head Circumference: Four of 27 individuals had a head circumference below the 3rd centile, and as mentioned, none had a circumference above the 50th centile. Head size is widely used in the pediatric setting as a marker of brain growth and development, but its use as a predictor of intellectual ability in the context of a syndrome is unknown. In this study no correlation was found between head circumference and severity of intellectual disability.

Behavior: Parents reported distinctive behaviors. Avoidance of eye contact was reported in 12/27 children, musical ability outside that of the family experience in 6/27, and outstanding long-term memory in 15 of the 22 individuals over the age of 2. In several children music was found to be an effective tool to teach new skills or concepts. The researchers noted that a love of music is also reported in other individuals with intellectual disability and may not be specific for Kabuki syndrome. Parents reported their child with Kabuki had excellent long-term memories for faces, music lyrics, events, and dates. The authors speculated that this raises the possibility of splinter or scattered skills in some individuals with Kabuki. The general picture that emerged was that of an affable, affectionate child who had poor eye contact, repetitive behaviors, musical ability, and outstanding memory. Abnormal socialization was not found in their participants, but it was noted that a previous study of cognition and behavior of 4 individuals, showed 3 out of 4 had some autistic-like behaviors and one met the diagnostic criteria for autism [Ho and Eaves, 1997].

Morbidity: Parents reported that the greatest time of increased infections and other health problems was in the first year of life. Commonly reported conditions were failure to thrive, feeding problems, recurrent ear infections, and surgeries for congenital abnormalities. Medical intervention in the middle and later childhood was generally infrequent. The authors reported insulin-dependent diabetes mellitus as a new association with Kabuki syndrome.

Mortality: Three of the 27 patients died from the long-term complications of congenital abnormalities. Aside from the effects of congenital abnormalities there has been no evidence that an individual with Kabuki has a shortened life span.

The article concluded with a detailed table of the various conditions and measurements found with each of the 27 individuals.
Katrina's Story

By Cathy Miller

My story starts eight years ago when I gave birth to a beautiful baby girl prematurely due to abuse. She had multiple problems - some from the abuse and some genetic. She was tested for Down's syndrome due to distinct facial features. The results were negative, but showed that she and I had an abnormal 14th chromosome with extra material. That was enough to get Katrina started in the early intervention program. She was given a g-tube to feed her because she could not latch on to my breasts or a bottle. Although she had a sub mucous cleft palate, this was not found until age five. She had heart problems as well. Those, too, were misdiagnosed. I requested to have her transferred to another hospital on the other side of the state. It was not until the day before her scheduled open heart surgery that we discovered the correct diagnosis of PAPVR and VSD. Changing hospitals was a major blessing in disguise as the surgeon on the case was one of only two in the country at the time that could perform the surgery. Two hospitals tested Katrina for genetic problems and she was not diagnosed until eighteen months of age and three major hospitals later. We went to the third geneticist who diagnosed her in fifteen minutes by looking at her. I laughed at him and then he showed me pictures of other children with the same syndrome. I was shocked at how accurate he was. I researched on the net and library and discovered as much as I could to help me get Katrina the help she needed as early in her life as possible.

I had been told by the NICU doctors that Katrina would not live and if she did she would be severely retarded and not walk, talk, or eat. Because of my faith and love, and perhaps even denial, I could not give up on her and became very persistent. I quit work for three years and went to every therapist I could find. She received PT, OT, ST, dysphagia, vision, and music therapies. I performed Healing Touch and Reiki on her as well as the traditional sensory integration therapy, oral stimulation exercises, and gross motor exercises. I went back to school to become an RN and had a private duty nurse work with Katrina while I was in school. She would stimulate her through reading, music, sign language, TV, games, brushing therapy, etc. I also had all my friends and family pray for her. There were people in fifteen states and three countries saying prayers. I truly believe that all that love and prayer helped tremendously in guiding us to the next state at the right time to help Katrina reach her potential. She is now integrated into a regular classroom with an aid to help her write and do some of her ADL's (adaptive daily living skills) - going to the bathroom, etc. - as she still has some difficulty reaching the faucets, and snapping pants. She has a computer and can use a keyboard. She is going into second grade and tests at third grade for comprehension. She is catching up quickly to the others in her class.

I have remarried a wonderful man, John. We had a healthy boy together, Jonathon. Jonathon shows no signs of the 14th chromosome abnormality or of Kabuki syndrome. He is a healthy active boy. Jonathon and Katrina love each other and help each other learn new things everyday. John also has three children: Christopher 18, Patrick 17, and Shannon 15 who love Jonathon and Katrina dearly. We are truly blessed.
I am Evan’s Mom. My name is Linda and Evan is my only child. We live in Vancouver, BC Canada with Evan’s stepfather, Jim. Evan was born on March 9, 1995 weighing 8 lbs. Although my pregnancy and delivery were normal, Evan was born blue and struggling for his first breath. I remember feeling so helpless in my delivery room. Time seemed to stand still while I waited for the doctor to return with news ... which of course, wasn’t good. Evan had a number of complications. Once Evan’s breathing was regulated in an incubator, he was transported to BC Children’s Hospital for more advanced care. He spent the next month in BC Children’s Hospital in their special care nursery where he was dubbed the “giant” as all the other babies in the nursery were premature. Evan was tested from top to bottom, and inside and out, for every possible anomaly. There began a journey that included several surgeries and what seemed like an endless number of tests. Seeing Evan in so much pain and discomfort seemed to slow time to a crawl.

Evan required analplasty to remove the skin covering his anus to allow him to go “number two.” It became immediately apparent that Evan was not digesting his food because blood was not reaching his lower body. At two weeks of age, he underwent corrective surgery to repair a coarctation of the heart. I’m sure most of us have been thankful for advanced medical technology! Along with the aforementioned operations, Evan’s “head to toe” inspection brought to light the following: one kidney is smaller and lower than the other; his spinal cord ends at L2 (lower than normal); he has extremely low muscle tone; his hips were dislocated at birth; he has a high arched palate with a partial sub-mucous cleft; and a double urinary tract.

Recently, we found out that Evan has a mild hearing loss (20%) in his left ear.

I believe he had “failure to thrive” syndrome during his first year and I’m sure some of you can relate when I say it wasn’t easy. Evan was admitted to the hospital for a week when he was six months old with a kidney infection, and again for ten days with a rare blood infection at the age of one. Just when I was really struggling for hope, I met a pediatrician who would change everything for the better. He put Evan on iron supplements and prescribed him weight gain powder for his formula. He also told me about a liquid called lactulose, which solved his severe constipation problem. I felt an angel had been sent from heaven to give me direction. This was a significant turning point for Evan. He began to gain weight and strength and started to crawl. Life for Evan and me became much easier.

The next few years of Evan’s life, up to the age of four were quite stable. He always has been, and still is, below the charts for weight and height — but he maintains steady growth on his own line. As a nine-year-old today, he is very slender and is finally 48 inches tall. Yahoo! I say that because he is now the required height to go on the big roller coasters rides, which he loves.

Evan’s next surgery was at four and a half years old. He underwent a Salter Osteotomy on his right hip and wore a full body cast for two months. He was a real trooper. In fact, he didn’t want it taken off. I’m sure it was because he enjoyed sitting in his recliner chair like a little prince, giving me polite orders all day long, while watching videos! Although he was tough as nails, I know he was a little anxious when the cast came off. One thing that Evan does when he’s nervous or worried is chew on the sleeves of his shirts or the material just under his neck. I had to change his shirts regularly the week that the cast came off. Although his shirt chewing habit is not as frequent, he still wets those arm sleeves every now and then.

In June of this year, Evan became very sick and was admitted back into the hospital with a kidney infection. He had a large 1-cm stone lodged in his ureter and another in his kidney. After controlling the infection, the doctors performed lithotripsy to break up the stones. The procedure failed due to the location and size of the
stones and he went under the knife to have them removed. He is doing very well today and we are waiting for the results of his stone biochemical test to determine what caused his body to create the stones. It is possible that they may be calcium stones, which apparently may have formed while Evan was in the body cast.

Educationally, Evan requires help from a Special Education assistant at school. He is in grade three this year for the second time. We held him back a year to give him some more development time. He is very popular with his classmates and he is treated with respect, which is real nice to see. He loves to use the computer and tries very hard to excel at everything he does. We have recently discovered a teaching method which has helped Evan progress significantly. Evan works with an Orton Gillingham (OG) tutor three hours a week. I'm very pleased to report that he is starting to read for the first time!!! Evan has made more progress with six months of OG tutoring than he did in three years of school. This teaching method is best known to help dyslexic children, but can work very well with developmentally challenged children. With his low muscle tone, he struggles with printing and colouring – but he's making progress with occupational therapy.

Evan has a very good memory for people and places. He often speaks of his great grandma, who passed away over five years ago, and he'll always remember who gave him what for Christmas three years later! While in the car, he regularly points out landmarks he remembers even if we haven't seen them for years. I've starting asking him to remind me of things I need to do in the morning and he never forgets. He's like a personal day planner!

If I could choose one thing to say about Evan it would be: to know Evan is to love Evan. He senses when people are lonely or depressed and he reaches out to them — showering them with love, affection and humour, which inevitably draws smiles where there were none before. Evan has a bubbly, loving, positive personality. It is hard to sum up in so few words how special he is.

Evan has this innate sense of rhythm and he loves music. His favourite is opera, classical and of all things, Tuvan throat singing!! It is apparently very therapeutic. Strangers are always approaching us, wanting to meet Evan as they watch him respond to music with passion or interact with people with such enthusiasm. Evan loves to laugh and definitely loves to be in the spotlight. He certainly didn't inherit my stage fright! Evan is also a huge video fan. His favourites are the oldies like Chitty Chitty Bang Bang, Sound of Music, Singing in the Rain, and many other old musicals, which he'll watch over and over – he even memorizes the lines!

One of his other favourite pastimes is looking through hundreds of photos we have taken on the computer. He likes to see himself in action! We think he is going to be an entertainer of some kind.

No matter what kind of day I am having, I am guaranteed that Evan will bring a smile to my face and make me or “Jimmy” laugh. A few of Evan's “feel good” lines are: “Mommy, you look so pretty”, “Mommy, your hair looks so nice”, “Mommy, you're the best!” and “I love Grandma and Papa.” He'll even notice and comment if I am wearing a new shirt! He always has something nice to say. He is just so sweet. We barely go a day without having to buy a card or flowers for one of the many special people in his life. He loves to give.

I think the reason why so many people are continually drawn to Evan is because of his gift of making them feel good. Evan's exuberant personality brings out the playful child in all of us.

A journey into the unknown can be frightful. But when you have a child who rises above every challenge with strength, courage, and the most positive of outlooks; it brings us comfort. And we realize that no matter what happens, everything will be okay as our child is living his life to the fullest with unconditional love and happiness!

Take care. Linda
Resource Links
(non-Internet contact information included, when available)
compiled by Heather Johnson

MEDICINE


Michigan eLibrary ~ Health Information Resources: www.mel.org/index.jsp
Michigan eLibrary - 'Best of the Internet' selections - From general index, select Health Information Resources. Perform a search, browse A-to-Z, or select from 31 subject categories listed. (Useful to read 'Evaluating Health Information on the Internet') - Nicely organized and easy to navigate.

ASSISTIVE TECHNOLOGY / ADAPTED PRODUCTS SERVICES

Access To Recreation: www.AccessTR.com
Adaptive recreation equipment for individuals with physical challenges. Emphasis on innovative design solutions that facilitate participation in recreational/sports activities. Terrain-specific wheelchairs, accessory backpacks, gloves, specialized lifts, exercise and training systems, fishing, hunting, cycles, aids for daily living. Catalog available. 8 Sandra Court, Newbury Park, CA 91320-4302, USA, 800-634-4351

Crestwood Communication Aids: www.communicationaids.com
Products for children and adults - Hearing, visual, verbal/non-verbal aids, picture cards, communication boards, amplifiers, language, educational, adapted toys, and interactive toys. Catalog available — 6625 N. Sidney Place, Milwaukee WI, 53209-3259, USA, 414-352-5678

EDUCATION

PACER Center www.pacer.org
(Parent Advocacy Coalition for Educational Rights) Founded in 1977 - based on the concept of parents helping parents. Mission statement: expand opportunities and enhance the quality of life of children and young adults with disabilities and their families. Working with 18 disability organizations, the PACER center is able to provide assistance to individual families, workshops, and materials for parents and professionals nationwide. Sizeable content is logically organized and easy to utilize. Established organization with quality resources. Paula F. Goldberg, Executive Director, 8161 Normandale Blvd., Minneapolis, Minnesota 55437, USA, 952-838-9000

Dr. Mac's Behavior Management Advice Site www.BehaviorAdvisor.com
Behavior management site devoted to the special education setting. Developed by Tom McIntyre, Ph.D. - professor of special education at Hunter College of the City University of New York. (link provided to view Resume/Vitae) Thousands of behavior management tips, and interventions, with step-by-step directions for implementation. Plus, a bulletin board to post questions and exchange solutions. To see all the topics/information available on site, you need to scroll down the entire first page. Excellent site.
Medical Journal Articles
Update: December 2003 ~ September 2004


Kulkarni ML, Shetty SK, Chandrasekar VK, Kulkarni PM


Niikawa-Kuroki (Kabuki) syndrome and hearing impairment

Variable expressivity in a family with Kabuki make-up (Niikawa-Kuroki) syndrome.

A case report of Kabuki make-up syndrome with cytomegalovirus infection.

Niikawa-Kuroki syndrome: Which characteristics must the otolaryngologist take into consideration?
HNO. March 31 2004; German, Hempel JM, Jager L, Naumann A, Schorn K.

Galan-Gomez E, Carbonell-Perez JM, Cardesa-Garcia JJ, Val-Sanchez de Leon JM, et al.

Long-term follow-up of three individuals with Kabuki syndrome.

New ocular findings in a case of Kabuki syndrome.

Anaesthetic management in a case of Kabuki syndrome.

Hepatic fibrosis in Kabuki syndrome.
Nobili V, Marcellini M, Devito R, Capolino R, Viola L, D’ilio MC.


Temporopontal spinal: a typical EEG finding in a Kabuki syndrome.


Unmasking Kabuki syndrome: chromosome 8p22-8p23.1 duplication revealed by comparative genomic hybridization and BAC-FISH.

Epilepsy and perisylvian polymicrogyria in a patient with Kabuki syndrome.