

From Russia with Love

INSIDE THIS ISSUE	
President's Message	2
Board Spotlight	3
Princess in Cowboy Boots	5
Fun Race Car!	8
PRISMS Support	9
Research Call	11
SuperKid Jacqueline	14

Last spring, PRISMS was contacted by Vera Tzetlina from Moscow, Russia. Vera is a sibling of a young lady, Sonia, a person with SMS. Vera wrote to us about her sister, Sonia, and also about the book that her mother, Bela Kafengauz, had recently written. In the book, Bela chronicles their family's life with Sonia, and the road to a correct diagnosis. The book also includes lovely family photos. Bela founded the Russian SMS Society (www.smsru.ucoz.ru) which helps families within Russia to understand Smith-Magenis Syndrome, provides information and support, and helps to connect families with one another.

We are so honored that Bela has written a very personal article for the PRISMS newsletter. We think you will see how devoted Bela and her family are to Sonia. We are so fortunate to meet our families from all parts of the world, and it reminds us that, though we are a very small community, we can stay connected and be a part of each other's lives. It also reminds us that worldwide awareness of Smith-Magenis Syndrome is critical to the ongoing research of SMS, and better understanding and management of the disorder.

Welcome Sonia, Vera, Bela and Alexander!

My husband and I live in Moscow. We have been married for 36 years. Alexander is a professor in the Biology Department of Lomonosov Moscow State University. He is also the director of a biological research station on the White Sea. My own initial training was also as a biologist, but after a long break to take care of our young children, I decided not to return to science. For ten years I have been working in the field of education at the Moscow State University of Psychology and Education. I am the founder of the Russian Smith-Magenis Syndrome Society.

I was encouraged by Maggie Miller to write this paper. We got to know each other a few months ago, and she invited me to write an article for the newsletter. I was inspired by knowing that these words would be read by people who

could understand me like nobody else. The article has been translated by our younger daughter, Vera, and edited by Maggie. I am very pleased to thank both of them.

The first and the eldest girl in Russia with Smith-Magenis Syndrome.

Sophia is our eldest daughter. She was born in 1984 in Moscow, just four years after genetic counselor Ann C. M. Smith and pediatrician and medical geneticist R. Ellen Magenis in the USA first described a group of children with 17p11.2 deletions. *Sonia*, as we call her, was diagnosed only at the age of thirteen in Canada by FISH analysis. At that time, none of the physicians we had consulted in our

...Continued on page 12



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A Message from PRISMS President *Randy Beall*

It's very quiet around my house all of the sudden. For the past 26 years, if things got too quiet, we knew we'd better go see what Laura, our daughter with SMS, was up to.... but that's all changed now. Laura has moved into a "group home" or, as we sometimes describe it, "a very nice private home with 3 other young ladies." Over eight and a half years ago, we put Laura's name on the Texas Medicaid Home and Community-based Services (HCS) waiting list. Once we were told that she would start receiving HCS benefits, we chose a local provider but had to wait almost a year later until the provider had a home available for her. She loves it. I've gone from feeling like each busy day is too long, to the realization that time goes by quickly.

Our 2012 PRISMS conference will be here before we know it. It will be held June 28-July 1 in Denver, Colorado. Our dedicated conference planning committee has been working hard to make this conference the best ever! Since my last column, we have secured author, professor and psychiatrist, Dr. Ross W. Green and Kelly Dorfman, M.S., L.N.D., a health program planner and nutritionist. I hope you are planning to come. It will be a great time to connect with other families and learn. If you are going to need financial help, please begin to ask your local agencies, school districts, and other charitable groups for support. We try to keep the costs as low as we can, but it's expensive to host a conference. Registration fees don't come close to covering the cost of putting on the event.

Due to the current economic environment, we have had to postpone the camp we had planned in partnership with Camp Breakaway Australia. Some of the foundations we approached for funding told us that they would like to fund our project, but this year all funds were going to rescue local programs that had lost their funding. We are continuing to look for funding sources and plan to have the camp in the future.

As you all know, we are a member-supported organization. We depend upon our members to host our conferences and continue our many programs. As always, the PRISMS board and volunteers are here to serve you. If you have suggestions, concerns, or comments, we would love to hear from you!!

Best regards,

Randy Beall



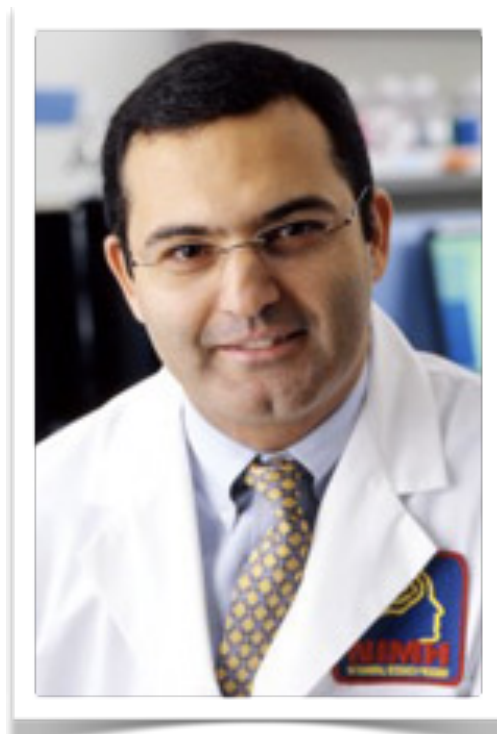
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PRISMS Smith-Magenis

Professional Advisory Board Spotlight

Gonzalo Laje, M.D.

PRISMS is pleased to announce that Gonzalo Laje, M.D., has joined the **PRISMS Professional Advisory Board** as of January 2012. Dr. Laje is new to our **PAB**, but has been a friend to our families for several years, as well as a dedicated researcher and physician. Dr. Laje has focused much of his attention on the pharmacological treatments of behaviors associated with Smith-Magenis Syndrome. He continues to drive the research for pinpointing better medications to treat the behavioral issues of SMS, while championing the need for more awareness, especially in the mental health professional community. Dr. Laje has published research papers regarding his study of SMS, and is a member of the SMS Research Protocol at the National Institutes of Health. Dr. Laje has also been an invited speaker at the past two PRISMS conferences, and was part of a panel presentation on Smith-Magenis Syndrome at NIH for visiting researchers from Australia this past summer. Dr. Laje's kind and soft-spoken demeanor has been a welcome relief to many of our families who might be in crisis and need guidance. He is always happy to answer phone calls and questions, and deeply understands the complexity of the syndrome and its effects on the entire family support system.

Dr. Laje is a psychiatrist and also has a Master of Health Sciences in Clinical Research. He maintains a private practice as well, specializing in psychopharmacological treatment and consultation of mood and anxiety disorders, pervasive developmental disorders (autism spectrum), attention deficit hyperactivity disorder (ADHD), and psychiatric management of genetic disorders. Dr. Laje's training began in Buenos Aires, Argentina, where he is originally from, and continued in the United States. He completed his training in general psychiatry at New York University/Bellevue Hospital in New York City, and his training in child and adolescent psychiatry through the combined program NIMH/Children's National Medical Center in Washington, DC. Dr. Laje has been the recipient of multiple awards, including: American Academy of Child and Adolescent Psychiatry (AACAP) Outstanding Child and Adolescent Psychiatry Resident; the AACAP Outstanding General Psychiatry Resident; the International Medical Graduate Mentorship Program in Psychiatry by the American Association of Directors of Psychiatric Residency Training (AADPRT); the American Psychiatric Association – New York District Branch – Resident Research Award; the APA – Janssen Research Scholar on Severe Mental Illness and the NCDEU-NIMH New Investigator Award.



We are honored to have Dr. Laje join the board and further strengthen the depth of expertise that is maintained on the **PRISMS PAB**. Welcome, Dr. Laje!

Recent Publications:

Pharmacological Treatment of Disruptive Behavior in Smith-Magenis Syndrome

Am J Med Genet C Semin Med Genet Nov 2010

Gonzalo Laje, * Rebecca Bernert, Rebecca Morse, Maryland Pao, Ann C.M. Smith.

Autism spectrum features in Smith-Magenis syndrome.

Am J Med Genet C Semin Med Genet Nov 2010

Gonzalo Laje, Rebecca Morse, William Richter, Jonathan Ball, Maryland Pao, Ann C.M. Smith

The Advocacy Corner

There is a new Website Guide to Disability Services. The ARC created this website with funding from the U.S. Department of Health and Human Services. The site, dubbed the Medicaid Reference Desk, offers a breakdown of various Medicaid benefits including medical and social services offered to those with disabilities based on where they live. The benefits available from one location to another and eligibility requirements for programs can vary wildly. Although the federal government mandates that Medicaid programs in each state meet certain requirements, states have significant leeway.

The reference desk offers basic information about each state's offerings, and also features a glossary of common terms and a selection of frequently asked questions. It helps families wade through the bureaucracy.

Peter Berns, CEO of the ARC, stated, "This website helps individuals with intellectual and developmental disabilities and others access and gain knowledge about Medicaid, which is an essential lifeline for millions of individuals with intellectual and developmental disabilities and their families." Once you access The Medical Reference Desk you can access information on Medicaid, Medicaid info by state, and download Person-Centered Planning Tool Kit.

DisabilitiesScoop.com is another website that claims to be the Premier Source for Developmental Disabilities News. They cover the latest updates on disabilities in several areas: education, health and behavior, politics, education, living and money, to name a few.

Take a look at some of the latest headlines:
Accessible Taxis Would Lead to Injuries and Lawsuits, Mayor Says
PBS Documentary to Chronicle Disability Rights Movement
SSI Payments to Increase
MTV to Focus on Students with Disabilities.

Volunteer Opportunities

Newsletter editor still sought! PRISMS is seeking interested individuals who have organizational skills, computer skills, and an eye for design. There is a well-developed newsletter committee to support the editor. The current editor would like to work with someone for several issues during a transitional time. If you are interested and would like to talk to the editor, Julia Hetherington, please contact her at editor@prisms.org. Please give your contact information plus the best time to call, and Julia will gladly call you to answer all your questions.

Parent-to-Parent

Did you know that PRISMS sponsors a parent-to-parent program? If you need someone to talk to who REALLY UNDERSTANDS what your life is like, try another SMS parent. You may want to find another parent in your area, or perhaps one who has a child the same age as yours. Tell us what you need. PRISMS keeps a list of parents willing to be contacted. We'll send you addresses and phone numbers, and then you can talk all you want. Contact:
Mary Beall
Phone: 972-231-0035
mary.beall@tx.rr.com

Important note:
PRISMS parent-to-parent program will only give out information on parents who have agreed to be contacted. If you would like to be added to the list of contact parents, please email Mary at the above address.

What is Smith-Magenis Syndrome?



Smith-Magenis Syndrome (SMS) is a chromosomal disorder characterized by a specific pattern of physical, behavioral and developmental features. It is caused by a missing piece of genetic material from chromosome 17, referred to as deletion 17p11.2. The first group of children with SMS was described in the 1980s by Ann C.M. Smith, M.A., a genetic counselor, and Ellen Magenis, M.D., a physician and cytogeneticist. Although the exact incidence is not known, it is estimated that SMS occurs in 1 out of 15,000 births. SMS is underdiagnosed, but as awareness of it increases, the number of people identified grows every year.

My Princess in Cowboy Boots

By Kaye Saxton

My grandkids call me Grandma Kaye. The story I am about to share with you is true. All grandparents have something in common with each other; their grandchildren. To each grandparent, we can share personal emotions, excitement, laughter, and frustration bringing the realization you are not alone in the world of Smith-Magenis. I hope this special story "My Princess in Cowboy Boots" will touch the heart of each grandparent who has a grandchild diagnosed with Smith-Magenis Syndrome (SMS) in their family. Before I tell my story, I want to give you an introduction to McKenna's world before being diagnosed with SMS.

McKenna (Grandma Kaye calls her Kenna) was born February 17, 2000. Kenna seemed to be a quiet, content baby hardly fussing at all. After Mom went back to work, I was the one to spend the days with her playing and tending to her needs till her parents came home from work. Kenna and I cuddled during the day sneaking restful hours napping together morning and afternoon. I was diagnosed with Multiple Sclerosis in 1997, with fatigue being a big problem that I had to deal with. My son would scold me, telling me to keep her awake during the day so she would sleep at night. Kenna had become a "Night Owl" and daddy was the one staying up with her for countless hours trying to get her to sleep. Little did we know a SMS child's circadian rhythm is backwards, meaning days are like nights and nights become days. Crawling or scooting did not seem to interest my granddaughter. Kenna would sit for hours making it perfect for Grandma Kaye to weed the strawberry patch with her at my side. A little dirt on her clothes and hands lead to a quick bath and a clean outfit, never leaving tattletale signs of our adventures during each day.

Being a nurse, I knew crawling was an important marker for a healthy baby. Many times with my daughter, April, we "patterned" her on the floor. Evening hours CeeCee, Kenna's mom, joined in helping Kenna become the "perfect crawler." We all agreed it was better late than never as she did learn to crawl. Still, I thought my grandbaby was "OK." Little fits of rage became visible as Kenna struck out pulling hair, scratching arms and faces, to the point of biting her cousins. Thinking it was a stage she was going through, I thought this, too, shall pass. When Kenna started biting herself, I knew this was going overboard a bit much. Her other cousins were not this aggressive towards each other; why her??? Because of the love a grandmother has for her grandchildren, it was easily reasoned everything was fine. This was not fine, and something was wrong; but what??? Mom and I discussed Kenna having her own type of "speech lingo" and noticed communication was something she could not do. What did she want? Do we not



understand her needs and desires? I suggested teaching her sign language; her mother agreed. My good friend, Karen, loaned me several VHS videos from Signing Time. Together, Kenna and I watched these magnificent videos and we started signing (talking) simple words. Water, milk, more, momma, daddy, and her vocabulary started to grow. Her mom and I took a sign language class at Eastern Idaho Vocational College. An unexpected transformation took place. McKenna showed less frustration and I knew what she wanted. Very interesting comments were made by family members; both pro and con. Teaching Kenna sign language" opened the door for her speech therapists to teach her to "talk" using real words.

By this time, mom and dad had taken McKenna to numerous doctors searching for reasons why McKenna was the way she was. Doctors labeled her as being deaf, autistic, ADHD, having a stroke at birth, and seizure disorders, among many others. The label most interesting to me was right occipital subpenymal nodular heterotopia; quite a mouthful of words. I still don't understand how it pertained to Kenna. It came to a point that McKenna's mom asked her pediatrician to do genetic testing on her daughter. This particular physician stated, "Why would it do any good? McKenna is what she is. It sure won't change her life because we can't fix it." I learned very quickly parents have to be their child's best advocate. Mom continued searching for an answer. What was wrong with McKenna? Then, at the University of Utah Hospital, Dr. Rope, Geneticist, found enough evidence warranting genetic testing. Dr. Rope had mentioned the

possibility of Smith-Magenis Syndrome; something he had never seen, only read about. I searched the internet, finding the best resource for SMS: www.prisms.org. I made a 3-ring binder for both Kenna's mom and myself with the precious information doctors did not understand. February 2007, we all sat in a room together listening to Dr. Rope give the concluding diagnosis, Smith-Magenis Syndrome. He presented us with 5 pages of information and mom proudly held her binder full of the information I copied from PRISMS. Learning about SMS has made a huge difference in the care and well-being of Kenna. We both attended PRISMS 5th Annual Smith-Magenis Conference that very year meeting parents worldwide who were in attendance. I will never forget the realization of knowing we were not the only family with a child acting the way Kenna did. The big treat for me was meeting Ann C.M. Smith, M.A., D.Sc.(hon) and Ellen Magenis, M.D.

And my story begins.

Once upon a time there was a little blue-eyed blond-haired girl sent from heaven. Her life was colored with curiosity, a touch of amazement, and splashed with giggles and laughter. Kenna had the softness of an angel's touch and a heart filled with the never ending love of a true princess. She was sweet and innocent, with the mischief of pixies. Her character is jam-packed with spunk and stubbornness, laced with total meltdowns, often sparked by the unknown. Definitely a "bombshell dressed in pink" with her Grandma Kaye wrapped around her little finger.

In Kenna's world, Disney princess' come to life. Watching Cinderella dance with Prince Charming, she sways to the music in perfect rhythm holding her jeans out as if she was dressed in the most elegant gown a princess could wear. When the music stops, my Kenna curtsies with a bow, ending the royal dancing event. She recognizes each princess and often plays their part as she watches their individual DVD. My heart aches as it is impossible to find her princess T-shirts in size 14. Kenna's body is growing up; but her mind will live forever in her princess world.

Let me tell you about the time Kenna put a dead mouse on her mother's side of the bed. Actually, she placed its tiny body gently on Mom's pillow covering it with a warm blanket. She proceeded to the fridge for cheese because Cinderella's friend (you know, Jaq') was hungry. Mom came unglued! All bedding got washed! Cheese was thrown away! Kenna stood by the bed sobbing, "My friend, my friend, daddy throw way my friend."

Kenna's search for happiness is found in small, simple moments of life adults overlook as foolish or silly. Did I mention Kenna is adventurous? Not the normal girl playing dolls or having a tea party. She struts out into the pasture with one thing in mind: catching grasshoppers. Quick, fast and on target, she catches one or two in her hands cupped tight. Sometimes she lets them go; sometimes she does surgical procedures on them. Amputating a leg or dissecting their abdomen, she gets "yuck" on her hands and

carefully wipes it on her jeans. She loves showing her trophies to Grandma Kaye. I am very cautious when she comes wearing a big smile on her face, giggling her silly giggle, and hands clasped tight together telling me, "I surprise for you!" Grabbing my hand, we go in search of "rolly pollys" (potato bugs). Turning over rocks and finding these little guys is another favorite pastime of Kenna's. Have you ever taught tricks to lady bugs? Grandma Kaye has, leaving Kenna mesmerized until they fly away.

When Kenna and I go shopping and she spies something she would like, two little hands go under her chin with a big smile and she asks, "Peese, grandma." Melting my heart, it is hard to say no as she doesn't ask for a lot of toys or candy. This outing a princess book was picked out to take home to read. When we arrived, she jumped out of my car with book under her arm and straightaway skipped to the horse corral. Grabbing my camera, I knew what she had in mind. Sure enough, as I came around the corner, Kenna was in the corral reading her book to a horse named Bert. Walking the perimeter of the corral, Bert was two steps behind her with his head hanging over her shoulder "looking at the pictures" in her princess book. This brought about a solution to her school homework with Kenna refusing to read a book to mom and dad. Mom wrote a note explaining to her teacher to have Kenna read her book to Bert, the horse, after school. Mom listens with a careful ear as she finishes evening chores; her teacher is pleased with task accomplishment, and Kenna loves spending time with Bert. We scored another "win-win" situation.

Looking back through time, I remember as a toddler she was drawn to the pet horse my son had retired from the race track years before. Arthritis had set in April's joints, slowing down her step. Kenna played under, around, and behind April daily. I honestly believe there was a heartfelt connection between April and Kenna. Both knew of each other's limitations, April keeping Kenna safe from harm. In time, Kenna learned to climb the fence calling April over to pet her. I was watching Kenna as mom was busy in the garden, realizing she wanted to ride April. Talking her through the necessary steps in reaching for her mane, mom hollered to me, "Do not put Kenna on April's back." I assured her I was not physically touching her, just carefully instructing on grabbing the horse's mane. As I whispered "Jump," Kenna looked at me with a grin from ear to ear. Yup, my little "princess cowgirl" made the "leap of her life," holding tight to the horse's mane, landing across the back of the horse. Not letting go of her tight grip, Kenna wiggled her legs in place, giggling out loud and proud of her most recent accomplishment.

Kenna's neighbor calls her the "Little Horse Whisperer." She mimics her dad, Jesse, as he trains horses to lead. Showing no fear, she reaches for the lead rope with stick in hand, and puts the horse through the required paces, including backing them up. Kenna has accompanied her dad to the horse auction, and he allowed her to walk the horse round the ring "showing off her skills" as people bid a price to buy.

The love for horses is tucked in her heart next to her feelings of love for the “princesses.” Mom and dad found a riding horse therapy class for children with special needs. North Fork Therapeutic Riding has become a “getaway time” from schools (Kenna attends schools year around), OT, PT, and speech therapies. Again, fun time has turned into benefiting Kenna’s struggles with SMS. A horse’s gait, similar to the human walk, helps strengthen spine and pelvic muscles, improving posture and coordination, increasing joint mobility. Children with cognitive disabilities not only benefit from the exercise, but also improve sensory integration, concentration skills, and motor planning skills. The special bond developed between horse and child helps increase confidence, self-esteem, and promotes a sense of accomplishment.

Now I can fit the cowboy boots into my story. You guessed it, Grandma Kaye took Kenna to her favorite western store, and boots without tags were worn home. Kenna danced and skipped with her face aglow making me her new found hero. She knew what the cowboy boots were for; riding her horse! A pink cowgirl t-shirt was also tucked into a shopping bag to take home. Kenna has officially become Grandma Kaye’s “princess in cowboy boots” opening another chapter in her life. Her best friend, Daisy, has become a permanent part of my granddaughter’s life. Who is Daisy? Kenna’s own little horse she rides almost every day in her “cowboy boots” with a smile upon her precious face.

Kenna, like most SMS children, hates loud noises, fireworks included. Celebrating the 4th of July, Kenna would hide indoors covering her ears from the sound of fireworks. This last year I saw an amazing accomplishment with Kenna sitting on her horse for over an hour as the grand display of fireworks were shot off above her head, enjoying something she always had feared. This was another first in her life attributed to the love of her horse, as she patted Daisy’s head reassuring her best friend, “It’s OK.”

Kenna is now 11 years old, and I have been part of her good times and bad. My mother taught me, “without any rain, how can we have rainbows?” Keeping this in mind, I bite my lip when Kenna suddenly outbursts, “Grandma Kaye go home!” “No like Grandma Kaye,” hits my stomach hard, but I have the realization she has never said, “I don’t love you.” I thrill when she waves both hands signing “I love you” over and over making everything all right, running into my arms and giving me a great big hug!

Working with mom and Kenna’s therapists, I have found a special way to reward her “good” times and manners. Kenna

becomes my backseat driver as she navigates our way to McDonald’s for an ice cream cone or if she chooses M&M’s, Grandma Kaye is quick to find a package hid away just for her.

When suddenly I miss Kenna in a store, make way for Code Adam. Yes, as quick as I can’t see or hear her, store personnel are notified. I have lost Kenna twice in Walmart, once in Shopko, and the most unusual reaction was in JC Penney’s. They weren’t sure if I was her grandma as she kept dodging away from me. Will I take her shopping again? Of course I will. It keeps my entire senses extra sharp knowing Grandma Kaye can and will handle any crisis Kenna throws my way.

Watching life through my granddaughter’s eyes, I know I am the luckiest grandma having this special angel in my life. I treasure each dandelion she picks just for me. Our silly chats on the telephone paste memories in my heart as she makes kissing sounds of goodbye. I will take the good with the bad, living each day we share together to the fullest. Live, laugh, and love is a great motto to live by. My nickname for Kenna is “Cricket.” I still do not know how she makes this sound so vibrant, but I have tucked this little quirk among the many favorite memories my heart holds dear.



Update: Kenna started her big school August 29th in 5th grade. Her IEP specifies for a personal aid to accompany her throughout the day, meeting her at the school door each morning. First day, no aid. Please note Kenna’s mom called daily for 2 weeks making sure Kenna’s IEP was in place. Not one phone call was returned. Contacting CeeCee, the principal gave a dozen excuses stating, “They had “lost Kenna.” Four adults did a frantic search hoping she was still in the school. Within 30 minutes Kenna was found in an empty classroom. The 2nd day of school there was no personal aid and more excuses. Mom made one more phone call and my granddaughter’s “personal aid” arrived at

school at 11:00 am. I am proud of my granddaughter’s best advocates being her parents. Because of Kenna’s unique needs, mom and dad have learned how to protect their daughter in the school system. Parents are the best advocate for any SMS child. Please don’t let anyone over-look what is needed and what the law states. Last fall, I gave Kenna a t-shirt stating these words, “If I am quiet, you better find me quick.” I have definitely learned these are true words to live by.

Lovingly written by Grandma Kaye and dedicated to the Princess in Cowboy Boots!

Raising Awareness and Having Fun!

By Joe Toussaint

My 14-year-old son Patrick has SMS. We knew from birth that Patrick had serious issues, but it wasn't until we moved to Houston, TX that he was properly diagnosed at the age of 8. We immediately joined PRISMS and became part of the "family." Given the lack of awareness about SMS, I wanted to try to promote (in my little way) some more discussion about SMS by elevating it's profile.

I have a passion for motor racing, and have been incredibly fortunate to be able to participate in the sport personally. While very far from a professional, I do possess "professional credentials" which allow me to enter into and drive in professionally sanctioned races. The biggest event in US road racing is held at the end of January in Daytona, FL. It is known as the Daytona 24 (it is a 24-hour endurance race). While not as big as the 24 Hours of Le Mans (the biggest/most famous sports car endurance race in the world in Le Mans, France), it has become a very big attraction, because the race draws "big time" racers from NASCAR (Jimmy Johnson, Jeff Gordon, etc.), as well as Indy Car (Dario Franchitti, Scott Dixon, etc.), some former Formula 1 drivers and even some Hollywood stars (Patrick Dempsey).

While I do not possess the skill (or financial wherewithal) to race in the Daytona 24 (one can still dream, though), I race in a 2.5-hour support race that happens on the day before the Daytona 24 called the "Grand-Am 200." The racing series (Grand-Am Road Racing) is owned by and affiliated with NASCAR. As such, people are encouraged

to walk around the pits, look at the cars, talk to the drivers and have a great up-close, hands-on experience. This year, I chose to make sure that my car (a 2005 Porsche 997) carried the PRISMS logo proudly on the doors. The car looked great and I can tell you that the logo generated a lot of questions and discussion.

It really was an eye opener for people who had no idea about SMS. I enjoyed talking with everyone I met and it was a great opportunity to spread the word a little.

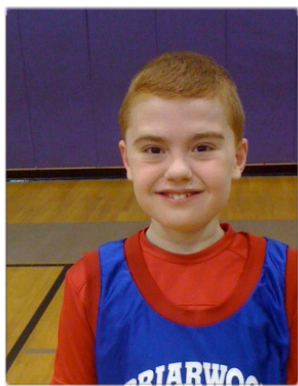


As for the race, we managed to keep the car in one piece and finish the race. Unfortunately, the Porsche is underpowered in its class, so our lap times weren't where we wanted to be (clearly not because of lack of driving skill J). We qualified last (33 out of 33) and finished 27, so at least we moved up the order. During the race we did have some tire issues which cost us too much time in the pits. In fact, during practice at a speed of about 155 mph I had a tire come off the rim. Quite a ride! Those walls look very unforgiving when you see them out of the windshield of a spinning car. Fortunately, I didn't hit anything and managed not to ruin the car.

All-in-all, a fun and successful race weekend – I proudly flew the PRISMS banner, didn't get hurt and managed to bring the car home in one piece. I hope to be able to do it again this coming January!

Anyone interested in looking at the race, the website is www.grand-am.com.

Joe Toussaint



PRISMS Support

Nickels for Nicklas 1st Annual Cornhole Tournament

Students from Dayton High School in Dayton, Kentucky have adopted PRISMS, Inc., as the recipient of an annual fund raiser for the Assisting Community Through Service (ACTS) group. The Dayton High School students have a high population that are recipients of public funding themselves, but they choose to raise money for our Smith- Magenis family for their philanthropy. Sherri Chan, Dayton High School's social worker, encouraged the students to support PRISMS, Inc., because of her friendship with the Weil family (Nicklas "Nico" Weil, a 4-year-old with SMS).

Although Dayton High School is an hour away from Louisville, Kentucky, Nico's home, the ACTS students hosted the fund raiser in Louisville. To cut costs and workload, they replaced the PRISMS, Inc., 5K walk they did in 2010 with the first annual cornhole tournament on August 13, 2011.

The cornhole tournament took little preparation compared to other fund raising events. There were five sets of cornhole boards, and almost twenty teams of two players that entered the tournament. The event was at a public park near downtown Louisville with a large playground for children.



James Neumann, from local grocery store, ValuMarket, supplied a large grill and his own grilling expertise for burgers and hot dogs during the event. All of the food was donated by Nico's aunt and uncle, Karen and Bryan Altman, and the drinks were donated by Planning for Life, Inc., a residential provider for disabled adults. With a party atmosphere, the cornhole tournament was a blast! Nico loves cornhole too, but was of course more interested in the food and drinks.

Unfortunately, horrible weather hit suddenly before the tournament was over. Leaving the park there were downed power lines and trees in the road. There were over 125,000 power outages in the Louisville area that night due to the impact of the storm. We SMS families really know how to go out with a bang! For those of you who are concerned, thankfully, our house did not lose power. Thanks again to everyone that donated. A special thanks to the Dayton High School ACTS students for their generous contribution to the Smith-Magenis community.

Buck Creek 5K

Several families met in Springfield, Ohio for the 5th Annual Buck Creek 5K for PRISMS. On Friday, September 23 the rain stopped just in time for runners and walkers to join forces and raise about \$1,000 for PRISMS. A 50/50 drawing and raffle of the World Famous Closson Peanut Butter Pie was a huge success, bringing in another \$200. Two newly diagnosed families joined us on Saturday afternoon for a picnic and time to share our favorite SMS stories. The younger kids played outside with bubbles while the older ones amused us with their one-liners and funny jokes. Make plans to join us next in 2012! Check upcoming issues of the Spectrum for the details.

Charlie and Tina McGrevy

Cooking with the SMS Family

Calling all cooks and their helpers! We would like to put together a cookbook of favorite recipes. We would like to include favorite pictures of your child, preferably helping you to cook or eating their favorite foods. Any comical anecdotes would also be appreciated. Some of you may also have special diets or unique recipes you would like to include. Each contributor can submit as many as three recipes in seven different categories.

Our goal is to have the cookbook ready by conference time. Pre-orders will be sold at a discount. Recipes or hints can be submitted by

email or snail mail. So don your chef's cap, break out Aunt Bell's famous cheesecake recipe, and let's see if we can create the world's most interesting cookbook. Watch for our e-blast with instructions on how to submit recipes online. You may also mail them to me at:

Mary Kate McCauley
24 E. Golfview Rd.
Ardmore, Pa. 19003

Parents Night Out - Houston, Texas

How does a night out on the town sound? Can you imagine going out to dinner with friends while your children are cared for by medical students studying to be pediatricians? The Texas area parents had that great experience on Friday night, Nov. 4. Each November this group of students has thrown a party for children with disabilities, including those with Smith-Magenis Syndrome. The medical students from Texas Children's Hospital in Houston turned out to provide one-on-one help and friendship to the children who attended the party. They are encouraged and supervised by Lorraine Potocki, M.D., clinical researcher and Assistant Professor at Baylor College of Medicine. Dr. Potocki is a member of the PRISMS Professional Advisory Board.



Houston Area Picnic

On Saturday, Nov. 5, PRISMS families from the Texas area gathered for the 8th annual picnic. We met at Bear Creek Park in Houston at one of the pavilions near the playground. Families united to swap stories, eat, play, and laugh. The children gave out lots of those famous SMS hugs. Some of us have been at all eight picnics, and feel very much like family. This year, we were excited to have two new families join us, one who was newly diagnosed and found out about the picnic from the PRISMS website.

Ceci and Gordy Poole planned activities including a bounce house, sensory/book area, and a music station. Games included bingo, a scavenger hunt, a "fishpond" game and cookie walk. Laura (age 26) quickly took responsibility for manning the "fishpond" and music director, with assistance from Coulter (age 5). The most popular songs were those by "Justin Bieber" and "Ernie and Bert." Cory, like most 20-year-old men, was too busy watching sports on his smartphone to play along. Texas is in the middle of a severe drought, resulting in a burn ban in the city of Houston. So the usual grilling couldn't happen. That was solved by cooking the hot dogs in an electric crockpot, and the hamburgers on an electric griddle. At the end of the picnic, we managed to get the whole group together for a picture (35 people) with no meltdowns and only a couple of bribes. We can hardly wait until next year.

*If you would like to host a regional picnic in your area, PRISMS can help by contacting families for you and publishing your event.



Are you online? Check out the PRISMS Facebook page.

Our numbers are growing quickly. Our page is being organized by volunteers, David and Denise Smith. Facebook looks like it will become a new and popular way for families and others to share information and learn about Smith-Magenis Syndrome.



If you have a Facebook account, search for "PRISMS/Smith-Magenis" and become a "friend." If you are new to social media, log onto www.facebook.com, join up, and get online to participate. It's free!

Conference News

The 7th International PRISMS Conference, "Building Bridges of Hope," is less than a year away. Planning is underway, but **we need your help** with fundraising to help defray conference registration fees. To keep costs affordable for our families, PRISMS subsidizes much of the conference costs. In fact, we are projecting that the conference will cost us approx. \$585 per person, yet registration is only \$200 for an adult. In order to charge registration fees of almost a third of the true cost, we need your help with fundraising. Our planning committee and conference helpers are all volunteers and parents of an SMS child, so we need everyone to share the load and assist in various ways.

There are many ways to host a fundraiser or to help organize fundraising events. We also need help with obtaining sponsors for the conference. Do you have contacts that you could approach about being a sponsor? A sponsor form is inserted in this newsletter. This is your conference, so please consider helping in order to keep the costs affordable! If you would like to volunteer or host an event, please contact Tina McGrevy, the PRISMS fundraising chair:

fundraising@prisms.org

Mark your calendars: **June 28th, 2012 – July 1, 2012**, Denver, CO

Registration will open in March. Registration fees are:

Adult (15 & up)	\$200
Child (14 & under)	\$100
SMS Individual	\$75
Professional	\$275
Prof. in training	\$225



Research Survey Regarding Eating Behaviors

Study assessing eating and behavior in SMS

The surveys assessing eating behaviors of children with Smith-Magenis Syndrome are still open! Information gained from these surveys will aid development of therapies and/or treatments of SMS, and there is still time to participate if you have not already. We have extended the survey deadline in order to accommodate parents of adults with SMS, as well. The following surveys are available online:

Parents of children older than 18 months and younger than 6 years old:

http://www.surveymonkey.com/s/SMS18m_5

Parents of children between 6 years and younger than 19 years old:

http://www.surveymonkey.com/s/SMS6_18

Parents of adult children with SMS 19 years of age and older:
http://www.surveymonkey.com/s/over_18years

If you'd prefer a copy of the survey to be mailed to you, or if you have any questions, please contact Dr. Sarah Elsea at selsea@vcu.edu or genetic counseling student Laura Meyer at meyerlv@vcu.edu. You may also reach us at (804) 828-9632, ext. 123 or (804) 628-4081. The surveys will be open through January 1, 2012. Participants have the option of entering in one of three drawings for a \$50 Amazon.com gift card.

country had known anything about Smith-Magenis Syndrome. It is very likely that Sonia was the first person in Russia with this diagnosis, and that she remains the eldest.

Thus, when Sonia was a child, we did not know the name or the characteristics of her condition. We tried to raise her as a normal youngster, and that approach worked well at first. Despite Sonia's many behavioral and developmental problems, she entered kindergarten at the age of five and was enrolled in a group receiving speech therapy. At eight years of age she began to attend a regular primary school with healthy children her age. Primary and secondary school education in Russia normally lasts eleven years. When Sonia was in seventh grade, it became clear that she could not continue in the regular program. During her last four years of secondary school, she studied at a specialized school for children with developmental disabilities.

This was possible because I quit my job and concentrated on helping my daughter. Sonia needed help every day with her schoolwork and daily life activities. Like all people with SMS, Sonia lacks the ability to concentrate. She could not do her homework by herself. But with stimulation and support she ultimately was able to manage it every day! She learned how to type, first on a typewriter and then on a PC, to play the piano, and to read books. It's amazing how many good books we have read together: first she reads a paragraph, then I read one, and we take turns until the end of the book. Sonia studied English as a foreign language, though when she didn't use it she forgot almost everything. But she still remembers a few simple English words, and always tries to use them when necessary.



Sonia is now 27. She can read and write, and she uses her mobile phone, PC, and other devices without problems. Sonia is very sociable and kind, and she likes to talk with people.



Sonia's current, most important medical symptoms include cognitive deficiencies, sleep disturbances, attention deficit, epilepsy, negativity, sudden mood shifts with explosive outbursts or prolonged tantrums, coprolalia, self-injurious behavior, chronic constipation, teeth grinding and tongue sucking, severe conductive hearing loss, and severe myopia. Sonia's condition is monitored by a psychiatrist and an epileptologist. A combination of antiepileptic drugs and neuroleptics prevents seizures and allows her to sleep through the night, as well as to maintain a stable emotional state and keep her behavior on a satisfactory level. These medicines, however, significantly increase the appetite, causing Sonia to gain weight in recent years.

Sonia is unable to live independently. She requires support for everyday life activities. My husband and I both work. Living with us, Sonia is forced to stay home alone during the day. She uses the computer, watches television, and talks on the telephone. She likes to listen to music on an audio player and type the lyrics into a Word file. If we ask her why she is doing that, since all the lyrics can be found on the Internet, she says that she likes to type. She likes rereading old books, which we have previously read together. Sometimes she picks up new ones. Sonia likes to eat, but she is a picky eater. Usually, we prepare food for her so she can heat up her lunch or supper in the microwave, and she eats it with gusto.

Moscow is a very big city. Sonia's condition makes it impossible for her to move about the city alone using public transportation. The only way she can get around is for us to drive her. Twice a week my husband and I take Sonia to the Center for Social Adaptation and Professional Training. These are her favorite days. The trip takes over an hour each way. Sonia has to get up on time and get ready to go. This is just as difficult for her now as it was when she was a

schoolgirl. And we always help her get ready. Sadly, this institution is the only such facility in Moscow, and is also unique in Russia as a whole. Organized as part of Technological College № 21, it is essentially a rehabilitation center for young adults with developmental and mental disabilities. It offers handicraft and creative workshops (ceramics, printing, woodworking, and sewing and textiles), lectures, and guided museum tours. The staff includes artists, teachers, and psychologists. An individual training program is designed for each student. For the last four years Sonia has been working in the sewing and textiles workshop. With the teacher's help, she is able to work on a loom, sewing machine, or by hand, make felt out of wool, and decorate fabrics. Using various combinations of materials and methods, the students create accessories, stylish handbags, ornamental batik, shawls and scarves, amazing cushions, skirts, and even dresses. Twice a year the college holds a special exhibition and fair at which these goods can be purchased. These events have become very popular. Usually the students can't even make enough products to keep up with the demand for them. These festivals give the students an opportunity to earn some money, and are an emotional high point for them.

We spend our summer vacations on the White Sea in northern Russia. Alexander works there all summer; Sonia and I join him for two months. It is a special time for all of us. The biology station is a research center, located far away from any towns in a forest on the seashore. The only way to get there is by boat. The community consists of the support staff, students, researchers, and their families. Everybody is friendly and kind to Sonia, so she feels very free and relaxed there. It is a chance to socialize, which is something she very much lacks. She moves around a lot and always takes part in celebrations and trips. She especially likes to go fishing! We live in a wood house. The fine summer climate and the quiet of this place put Sonia in a better, more balanced mood. I wish every person living with SMS could spend time in a place like this!

Sonia's younger sister Vera is 25. She works as a pediatrician and lives on her own. The sisters truly love each other, which makes my husband and me happy and gives us strength.

My book about their childhood came out in 2008 in Moscow. Its title is *A Child with a Hereditary Syndrome: the Story of Her Upbringing, or Faith, Hope, Love, and Sophia*. *The only part of the title that doesn't come across in the English translation is that the Russian word for "faith" is our other daughter's name – Vera. In the book, I tell about the early childhood, kindergarten, and school years of Sonia and Vera, trying to spell out the challenges faced by the families of such children, and discussing educational and social questions that seem important to me. I'll be happy if our experience can help other families manage these problems and move ahead.

* Bela Kafengauz. 2008. Rebenok s nasledstvennym sindromom: opyt vospitaniia ili Vera, nadezhda, liubov' i Sof'ia (Moscow: Prakticheskaya Meditsina, 2008).

★ Jacqueline

Jacqueline was born on Valentine's Day 1991. Although she would have you believe that she is a sweetheart all of the time, this is not always true! But she has certainly been a blessing to all those around her, not the least of which are her mother and father.

Jacqueline loves watching movies and especially loves musicals. She can recite the lines and scenes of almost anything out of such popular musicals as *The Wizard of Oz*, *Hairspray*, *Annie*, and many more. She recently had the pleasure of performing the part of Dorothy in a full-blown play put on by the sheltered workshop where she works, VIP Industries, in Cape Girardeau, Mo. It was truly an amazing experience to watch people from this venue spend months practicing and memorizing lines for a one performance show in front of hundreds at a local theatre. There were many important people who came to support the show, including our local mayor who is a strong supporter of the local workshop and the impact it has on our community. Jacqueline did a great job and really enjoyed the scene where she got to pour water to melt the wicked witch of the west (played by one of her best friends)!

Jacqueline is currently completing her senior year in high school. She attends 1/2 days and works at the sheltered workshop the other 1/2 day. She will continue working at the workshop full time after her graduation next May. Jacqueline loves supporting the sports programs at Central and attending games when possible. Her favorites are the football and basketball games. She is an honorary member of the dance team and participates with them during many halftime activities. This is a big highlight for her!

Favorite pastimes are watching movies, working on workbooks and puzzles, and going to the gym. She goes to the gym at least 5 days a week and almost everyday after school. It is both a social event for her as well as much needed exercise. She walks and works out lightly on weight machines under the instruction of qualified staff at the facility. She knows everyone there, and if anyone new shows up, you can bet it won't be long before she will know everything about them!

Jacqueline also loves pets, especially dogs and cats. She has 3 dogs. Her favorite is Princess who has been a companion of hers for years and sleeps with her every night. As Jacqueline nears the age of 21, her favorite line is, "I am an adult!" Well..... not quite yet but she is certainly growing up quickly!



Definitely our family's SuperKid!

Do you have an SMS SuperKid? We know what amazing things our kids can accomplish, and those big and small moments of success need to be celebrated for all to see. Please consider sharing your moments with us. If you have questions or need help with the story, please contact the editor at: editor@prisms.org.

Sunny Side Up



Two Tales from the Car submitted by Jessica Kirklin

Diagnosed Correctly

One evening, my SMS daughter, Rachel, was with me when we gave a loud and talkative little girl a ride home. After the girl got out of the car, Rachel asked me what kind of syndrome the girl has. I told her that she doesn't have any kind of syndrome. Rachel thought about it for a moment and then said, "I think she has Obnoxious Syndrome."

You Can't Argue with Success

As we approached a red stoplight, our daughter with SMS, Rachel, used her powers of command to change the light by saying, "Verde, por favor!" The light turned green, so Rachel responded with, "Gracias!" Rachel knows only a few words in Spanish, so I thought it was interesting that she chose to talk to the light in Spanish. I asked her what made her think that the light spoke Spanish. She replied, "Because it worked, didn't it?"

Slogan Winners

Congratulations to the winners of the 2011 SMS Awareness Day Slogan Contest!!

Grand prize goes to Jessica Kirklin with **SMS-Less Sleep, More Love**

Jessica wins one free night at the beautiful Denver Renaissance Hotel during the 7th Prisms International Conference next summer, and a \$50 credit to the PRISMS store.

First Runner up and winner of a \$50 credit to the PRISMS store is Denise Smith with her slogan **Meltdown Loading...**

Congratulations to our second runner-up, Yolanda Van Der Schoot with **it'S**

**More
than a Syndrome**

And our third runner-up, Dwanda Daniel with **SMS- Tough to Manage, but Easy to Love.** The second and third runners-up have each won a \$25 gift card to the PRISMS store.

Thanks to everyone who entered. We were overwhelmed with the response and found it difficult to choose our favorites. You can visit our online store at www.cafepress.com/smithmagenis to purchase PRISMS merchandise.



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**Be sure to participate in
the eating behaviors
research study! Info on
page 11.**