



Parents and Researchers
Interested in
Smith-Magenis Syndrome

Spectrum

Volume 7, Issue 1

Spectrum - The Newsletter of PRISMS

Winter • 2003

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 1980's by Ann CM Smith, MA, a genetic counselor, and Ellen Magenis, MD, a physician and cytogeneticist. Although the exact incidence is not known, it is estimated that SMS occurs in 1 out of 25,000 births. SMS is underdiagnosed, but as awareness of it increases, the number of people identified grows every year.

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Questions?

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SMS Research Roundtable

July 3, 2002 – Denver, CO

By:

*Ann C.M. Smith, M.A., D.Sc. (hon)
Chair, PRISMS Professional Advisory Board*

With funding from the National Institutes of Health (NIH) Bench-to-Bedside award, the SMS Research Unit in the National Human Genome Research Institute and PRISMS' Professional Advisory Board (PAB) convened the 2nd SMS Research Roundtable on July 3, 2002 in conjunction with PRISMS national education conference in Denver, Colorado. The roundtable, held exclusively for the exchange of scientific data and treatment approaches, brought together an invited group of basic science and clinical researchers to discuss their current research efforts and foster future collaborations. A special morning SMS Sleep Symposia concentrated on research efforts to understand the chronic sleep disturbance in SMS in the context of current knowledge about the biology of sleep. The afternoon session provided time for researchers to summarize their current research efforts and findings to understand the:

- Clinical and natural history of SMS
- Neurobehavioral aspects of SMS
- SMS critical region and potential gene(s) causing SMS
- Potential and/or existing animal model(s)
- Optimal management and/or treatment intervention plans

SMS Research Roundtable July 3, 2002 - Participants (alpha order)

- Judith Allanson, M.D. Children's Hospital of Eastern Ontario, Canada
- Weimin Bi, Ph.D. Baylor College of Medicine, TX
- Sarah H. Elsea, Ph.D. Michigan State University, Detroit, MI
- Brenda Finucane, M.S. Elwyn Inc. , Elwyn, PA
- Ellen Conroy, D.O. Family Practice, Wallingford, PA
- Hélène De Leersnyder, M.D. Hospital Necker, Paris, France
- Wallace Duncan, Ph.D. SMS Research Team, Biological Rhythms, NIMH/NIH
- Ellen Elias, M.D. Children's Hosp, Denver, CO
- Barbara Haas-Givler M.Ed. Special Educator, Coopersburg, PA

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(Continued from page 1) SMS Research Roundtable

· Peter Hammond (Prof.)	Biomedical Informatics/Eastman Dental Inst. for Oral Health Care Sci., London
· Hanna Hildenbrand	SMS Research Team, Rehabilitation Medicine, CC/NIH
· Kyle P. Johnson, M.D.	Oregon Health & Science University, Portland, OR
· Liqun Luo	Associate Professor, Dept. of Biological Sciences, Stanford University, CA
· R. Ellen Magenis, M.D.	Oregon Health & Sciences University, Portland, OR
· Staci Martin, Ph.D.	SMS Research Team, NCI/MICC/NIH, Bethesda, MD
· Rebecca Morse	SMS Research Team –NIH Summer Intern, Bethesda, MD
· Lorraine Potocki, M.D.	Baylor College of Medicine/Texas Children's Hospital, Houston, TX
· Christine Shaw	Baylor College of Medicine Houston, TX
· Beth Soloman, M.S., CC-SLP	SMS Research Team, Speech Pathology Section, CC/NIH, Bethesda, MD
· Ann C.M. Smith, M.A., D.Sc.(Hon)	Head, SMS Research Unit, NHGRI/NIH; Chair PRISMS Prof. Advisory Board
· Michaele Smith, PT	SMS Research Team , CC-RMD/NIH, Bethesda, MD
· Pawel Stankiewicz, M.D., Ph.D.	Baylor College of Medicine, Houston, TX
· Mary Anne Tamula, M.A.	SMS Research Team, NCI/MICC/NIH, Bethesda, MD
· Jian Yan, M.D.	Baylor college of Medicine, Houston, TX
· Katherina Walz, Ph.D.	Baylor College of Medicine, Houston, TX
· Colanthe Wilson, MD	Oregon Health & Sciences University, Portland, OR

Reprinted SMS Research Unit January 2003 Research Update Newsletter

Sleep Disturbance in SMS

An Exception to the Rule for Melatonin



Wallace Duncan, Ph.D., NIMH/NIH/HHS

Ann C.M. Smith, M.A., NHGRI/NIH/HHS

Behaviorally, some of the key features of Smith-Magenis syndrome (SMS) include sleep-disturbances, hyperactivity and sudden changes in mood. Poor sleep has an enormous impact on daily living, both for the child with SMS and for his/her parents or primary caretakers. Past research in the study of behavioral aspects of the syndrome (Dykens and Smith, 1998) show that sleep is the most significant factor in predicting the difficult behaviors that occur in SMS. Efforts to understand sleep patterns & habits in SMS have led to the confirmation of an unusual inverted circadian rhythm of melatonin. Determining the underlying cause(s) for this disrupted sleep cycle have only just begun. While exciting, the early findings raise many more questions that warrant further research study.

What is already known?

As infants, persons with SMS are generally described by their parents as happy, easy going babies who are complacent, sometimes lethargic, and often sleeping more than might be expected (Smith and Gropman, 2000). The sleep disturbance may go unrecognized in infancy. However, usually by age 2-3 years, the sleep pattern shifts, when children with SMS may go to sleep late, sleep for short periods during the night, awaken several times during the night, wake early in the morning (5:30-6:00 AM), and nap frequently during daylight hours. The sleep disturbance in SMS is chronic occurring almost every night, thereby impacting the child's daytime behaviors, as well as family and school life.

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Biology of Melatonin

Melatonin is a hormone secreted by the pineal gland that helps control body rhythms and sleep wake cycles in relation to daylight. In healthy individuals it is often used as a marker for the hands of the internal circadian biologic clock. The secretion of melatonin is controlled by the internal circadian “clock” that is located in a part of the brain called the suprachiasmatic nucleus, or SCN, located inside the hypothalamus and the pineal gland..

The term “circadian rhythm” comes from the Latin words *circa diem*, that means “about a day”. Under normal circumstances in a 24-hour period, light stimulation of the retina at the back of the eye relays a message to the pineal gland via the SCN about the light signal. Light at night interrupts the SCN signal to the pineal gland, thereby blocking the secretion of melatonin. Otherwise, in darkness, the SCN signals the pineal to make melatonin. Throughout the plant and animal kingdom, melatonin levels rise in the evening, peak during the night, and then fall to very low values during the day. But this does not appear to be the case in persons with SMS.

Based on exciting research and work, first by Dr. Lori Potocki at Baylor College of Medicine (Potocki et al., 2000) and confirmed later in France by Dr. Helene DeLeersnyder (2001), we now know that SMS is an exception to the rule that melatonin is produced at night! Both groups reported compelling evidence of a reversed daytime (diurnal) variation in melatonin, i.e., increased in the day and decreased during the night. This finding is in sharp contrast to all other living things in the plant and animal kingdom with nighttime highs and daytime lows of melatonin. In addition, melatonin levels should be suppressed by daylight. However, this may not be the case in SMS. Future research to understand this conundrum must answer two critical questions:

- Why is SMS an exception to the circadian rule for melatonin?
- Why doesn't daytime light suppress melatonin levels in the daytime like it does in other species?

National Institutes of Health - SMS-Research Team **Home Assessment of Sleep**

Sleep disturbance in SMS has a significant impact on child's daily living and behaviors at home and in school setting. Any management strategies that can improve sleep have potential to improve behaviors.

Innovative approaches are needed to study and fully understand the disrupted sleep patterns

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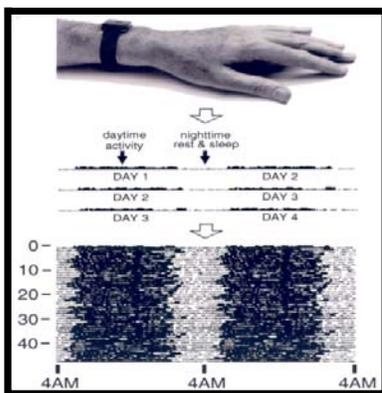
in SMS that are associated with elevated daytime highs of melatonin. In September 2002, the SMS Research Unit at the National Institutes of Health began a Home Assessment of Sleep (SMS-HAS). This new study includes three non-invasive measures to study the sleep-wake cycle in SMS in the home setting.

Monthly Sleep Diary

Parents are asked to chart their child's sleep/wake patterns using a standard monthly sleep diary. The form permits the primary caretaker/parent to note the times when their child is sleeping or napping by coloring in grid spaces and leaving them open for awake times. At the end of each month the completed diary is returned to the SMS Research Unit, where it is scanned and then transferred into a digital format. Statistical analysis using special research software permits Dr. Wallace Duncan to calculate the average amount of sleep and awake time for the month. By collecting sleep diaries for several consecutive months, a quantitative picture of a child's sleep pattern emerges that can be related to different drug or school schedules.

Any families interested in charting their child's sleep are welcome to a copy of the Monthly Sleep Diary form (contact Ann Smith at NIH or the PRISMS office). Families considering a trial of a new medication may find the sleep log useful to monitor how a medication impacts their child's sleep. When a new medication trial is considered, keeping the sleep log for the 1-2 weeks prior to trying the new medication and then continuing to track sleep patterns over the course of the trial and/or when medication is discontinued, may be useful in determining if the new medication is having any impact on sleep patterns. Noting the medication dosage and time given are critical to understanding drug effects.

Fig. 1 Actiwatch



Home Assessment of Mood & Behavior

In conjunction with the sleep diary, the primary caretaker is also asked to rate their child's mood/behavior twice daily, using a simple visual line scale.

Wrist Activity Monitor Actiwatch

Another tool used for the SMS-HAS is the wrist activity monitor (Actiwatch). This monitor is a small wristwatch like device used to record the child's movements. Worn on the child's non-dominant wrist, the Actiwatch measures activity and rest patterns during each 24-hour day.

Individuals with SMS are asked to wear the activity monitor for up to 2 months or longer as tolerated. Inside the watch is a microchip that records the activity. When the watch is returned to NIH, Dr. Duncan downloads the critical rest/activity data to create an activity/rest profile as shown in Fig. 1.



RESOURCE MARKETPLACE

You Be the Judge

By Laurie Bellet

Have you ever noticed how often you are called upon to evaluate something? At every turn, there is a new product, a new program, a book, an article and, who knows what, that we must assess and decide upon. So, how do we go about this process? While nothing is fool proof, there are some simple guidelines you can use to help you make informed decisions.

Products such as toys, clothes and equipment are the easiest. Some may just be so inexpensive that you can buy it and try it. For the more costly or ongoing purchases, consider first the vendor. Is this a company that has been around for a while? If you ask around, are acquaintances at all familiar with the company or the product? If the product is available in stores, or will be demonstrated at disability fairs, find out where, and test-drive it. Consider whether you will need something that will last or something that will look great in the short run. My favorite clothes come from the Hanna Anderssen catalog. They are so durable you can pass them through generations. They are costly though and, likely not a good investment, if rips and stains are part of the wardrobe profile. If you are buying from a specialty catalog, take a look at the models. Are those able bodied folks in the pictures? I feel much more confident if the models clearly need what they are modeling. Check into the replacement warranty. Our optician once convinced me to buy an “unbreakable” pair of glasses because of the quality of the warranty. He was so smart! We went through 3 pairs of those glasses for the price of one!

When evaluating programs, look carefully at how the actual program suits your true needs. A program may be terrific in what it does but not a good fit for your situation. Because of funding and contractual limitations, programs cannot always do what we wish they could. Spend time in observation and be specific as to elements of appropriateness and elements that don't mesh. Look at the qualifications of the staff, ongoing training opportunities, and the rate of staff turnover. Look at the program consumers and the environment for expressions of happiness and evidence of cleanliness.

When evaluating professional papers, look to the source. Who has authored the paper and what institution backed the research? When assessing treatments or therapies, learn the parameters of the studies. Was the patient sampling large? Were double blind controls run? Can the results be replicated in another center? Does the author benefit directly from the results? The easiest thing to check is the date of publication. If you find reports of a “successful,” but apparently underutilized treatment, that was documented a decade ago, you would be wise to wonder why the treatment is not accepted practice. A new innovation may be promising, but not have a proven track record long term. Whenever possible, check up on subsequent papers. When once researching a much lauded surgical procedure, I found that the surgeon published a later paper saying he was no longer recommending his procedure, because it resulted in an unacceptable recurrence rate.

There are books on every subject and to document every opinion. First off, look to the publisher. When an author or the author's business (if not itself a publishing house) self-publishes the work, your red flag should go up. If the author is the only provider who seems to have this success or to know of a given treatment, proceed with caution. Do not consider the author's bio to be encyclopedic in its accuracy. Authors craft their own biographies for book jackets, press releases and the like. Hmm, now that I consider it, I think I will add the letters P-H-D after my name on my next book. I have decided they will stand for “Parenting of the Highest Degree”!!

Genetic Alliance Membership - Sign Up Today!

Genetic Alliance membership is free to everyone beginning January 1, 2003

The Genetic Alliance is moving from fee-based membership to free membership beginning January 1, 2003. In an effort to represent every individual and group concerned with genetics, we are opening up our membership. We invite you to register on-line at <http://www.geneticalliance.org/membership.html> Help us continue our work in education, outreach and advocacy, as well as initiate new activities that meet our common goal. Advocacy groups, medical advisory board and individual members are urged to register as Genetic Alliance members. If you know of groups or individuals who do not have access to the Internet, please have them telephone or write to us to register their interest. Our phone numbers are 1-800-336-GENE or 202-966-5557 or you may write to us at:

Genetic Alliance
4301 Connecticut Avenue, NW, Suite 404
Washington, D.C. 20008-2369

As a broad and inclusive coalition, we will work together to ensure that genetic research results in improved health care for all.

PRISMS, Inc., as an organization, is a member of the Genetic Alliance.

NOW AVAILABLE

Audiotapes from the 3rd International Smith-Magenis Conference Denver, Co - July, 2002

If you were unable to attend the 3rd International Smith-Magenis conference or attended and would like to be able to review information heard at the conference, you may want to order a set of our audiotapes. Our tapes can be purchased for \$4.00 each plus shipping and handling or as a (9) audiotape set for a total cost of \$43.00.

Listed is a brief overview of the topic contained on each audiotape:

- Tapes 1 & 2 Welcome and Opening Introductions: Navigating the Maze of Professionals: Connecting the Docs
- Tape 3 Behavior Management in SMS: A handbook for Success.
- Tape 4 Newly Diagnosed Families: An Historical Overview.
- Tape 5 Riding Off Into the Sunset: Transitioning to Adulthood.
- Tape 6 Speech Language Development: A Road Map to Total Communication.
- Tape 7 Educational Issues in SMS. Part I: On the Road with SMS: Tales from the New Frontier
Part II: From Around the Campfire to Inside the Wagon: Education
- Tape 8 Research Updates: Part I: The Science of Sleep Part II: SMS Mice: What will they teach us?
- Tape 9 The Tools for Reaching the Peaks: Success Stories from the Home Front.

If you are interested in ordering any or all of these audiotapes, please contact: **PRISMS, INC.**
c/o Caroline Pope
411 Longfellow Drive
Williamstown, N.J. 08094-3205
Phone/fax: 856-875-7040
e-mail: CPopeRN@aol.com



Meet One of Our Kids...Christy McCauley

By Mary Kate McCauley

Christy was born April 6, 1977; platinum blonde hair and the most beautiful blue eyes, with perfectly etched brows and lashes. She was so still in utero that I worried all was not right. Some 16 years later we were to discover Christy had Smith-Magenis Syndrome.

She was such a puzzle. "When she was good she was very, very good.....but when she was bad she was horrid" She was bright, able to read almost anything, but unwilling to do what was asked of her.

The neurologist said, "Our goal must be to make her as socially acceptable as possible." Well now, wasn't that an interesting concept. That meant trying her on all and every medication out there.

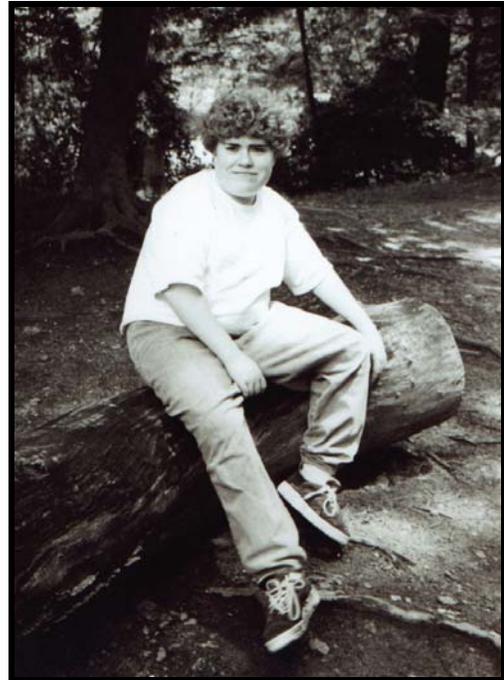
Friends, relatives and strangers were often shocked at her bluntness and lack of tact. The kindness of strangers was often rewarded with, "Did you forget your deodorant!" or "Did you brush your teeth with kitty litter!"

She did fairly well in a small private school for mostly autistic children till she was 13. After that we began to have increasing difficulty with school and transportation. The school district urged us to seek residential placement so Christy would have a future. This was a very difficult decision.

For two years we searched for a place our hearts could handle. Finally, we found Bancroft Neuro Health in New Jersey. We were able to get the school district and the state to fund an out of state placement. It was a difficult transition. Christy was almost 17 and we talked about her going to handicap college so she could learn a skill and get a job.

She presently is in a group home, living with three other young women. We speak on the phone every night or so and she comes home every other weekend.

For now it is working, not always smoothly but life is manageable. I still hope for the day that serenity reigns. But hey, Life is never dull.



Christy McCauley

GOODNIGHT, SLEEP TIGHT!?

By Rocehelle Wright

My 4-year-old son, Zachary, has SMS and is a fairly “normal” sleeper, without the use of any medications. Now that I have your attention I will add the disclaimer “to the best of our knowledge”. In July of 2002 we began using a Vail enclosed bed for Zachary. After a few nights to adjust to the new bed we have settled into a smooth bedtime routine. Each night Zachary climbs into the bed on his own, lies down to be covered up and says goodnight. Well, he actually says goodnight over and over while we zip him in the bed. We put him to bed at 8:15 PM and most nights we do not hear from him again until approximately 6:30 the next morning. On the few nights each month that we hear him during the night he has woken and turned on one of the toys that we keep in his bed or I have gone in to tuck him in and he is awake. We do not have to get up with him or take him out of his bed. If he is awake when I go in I just say goodnight, and go on to bed, or if I am in bed and hear him I can roll over and go back to sleep knowing he is secure in his bed.



Zachary Wright and his Vail bed

Now you may be thinking that I am exaggerating, or he cannot possibly have SMS, or saying just you wait, it will change. Although I certainly hope he continues to sleep well, I am convinced that if he were not in the Vail bed he would be up early and often now. When on vacation or away from home without the enclosed bed we have experienced first hand the frustration of having Zachary up early, between 1:00 and 3:00 AM, and often, we tended to give up on trying to get back to sleep around 5:00 AM. He always seems to be in a good mood,

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and full of energy in the middle of the night, and he refuses to stay in bed long enough to fall back to sleep.

Zachary can climb quickly out of a crib, pack and play or traditional bed with side rails. We have tried them all. In fact he is so disruptive to everyone's sleep when we are away from home that I devised an enclosed bed for travel. It consists of a twin bed air mattress, an air pump for quick inflation/deflation, and a twin bed tent. The tent is a child's racecar design play tent that fits on a twin bed. To keep Zachary safely inside I sewed the opening on one side of the tent closed and sewed in three

zippers on the other side, all of which close from the outside. The three zippers zip close to a central spot and can then be connected together with a twist tie to keep Zachary safely inside. Zachary has tried very hard to get the zippers open to date has not succeeded.

Now I will admit that we do not know if Zachary is actually asleep until 6:30 each morning. However, I can tell you that the rest of the family is asleep, and if Zachary is awake he is very quite because we do not hear him. It is a great benefit to know that Zachary is not up wandering, and he is not keeping the rest of the family awake.

I do not work for Vail Beds,

and I in no way make any guarantees about the product. I am simply sharing my families success story in hopes that it will help others struggling with their child's sleep problems.

If you are interested in finding out more about this bed you may contact the company on-line at www.vailbed.com or call 800-235-VAIL. They will work with you to obtain insurance or other agency funding for the enclosed bed as it is fairly expensive. This process took our family three months from the time the completed paper work was submitted to Vail to the time we received the bed. It was definitely worth the wait for our family.



Help PRISMS document the benefits of a VAIL bed

The article by Rochelle Wright discusses the potential benefits for use of a Vail bed. Several parents who have used the bed acknowledge that it may assist their child in sleeping better. Since we do not have any data to validate this observation, families considering purchase of a Vail bed can assist in collecting sleep data (using Actiwatch and the Monthly Sleep Diary Form) before getting the bed and then continue after their child is sleeping in the Vail bed. If collected, such data can be used to determine if the Vail bed does indeed make a change in sleep. Please notify PRISMS, Ann Smith or Wally Duncan if you have plans to begin use of a Vail bed. See contact information below:

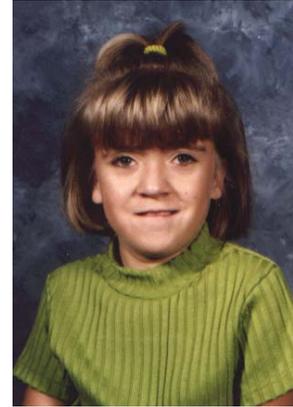
PRISMS, Inc. 76 South New Boston Road Franconia, NH 03043 Phone 603-547-8384 Email: info@smithmagenis.org	Ann CM Smith, MA D.Sc. (hon) SMS Research Unit, NHGR, NIH 10 Center Dr., Bldg. 10, Rm. 10C103 Bethesda, MD 20892 Phone: 301-435-5475	Ann CM Smith, MA D.Sc.(hon) Email: acmsmith@nhgri.nih.gov Wally Duncan, Ph.D. Email: wduncan@mail.nih.gov
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Miracles In Education

PRISMS has been pleased to have had an opportunity to publish a series of articles regarding an educational approach to teaching children diagnosed with Smith-Magenis Syndrome. In the last installment of our series, Christina Miller and Leanne VanDover provide us with their final thoughts and insights into the educational approach of Leanne's daughter Evin.



Evin VanDover

Parent Perspective

By Leanne VanDover

In this closing article I would like to touch on how Evin has been used to bring about miracles in my life. The following is my story as a child with special needs that has been turned around and used for good to help Evin and others like myself.

As a child I was diagnosed with a learning disability, however I was told that I was mentally retarded and worthless by teachers, students and family members. It has been a long journey to overcome the effects that this brought about in my life, but this journey has prepared me to be Evin's mom.

Because of my experience as a child I realized how important it was to instill in Evin her great value and worth. We began by giving her the name Evin which means gracious gift from God. If I did nothing else in the first five years of her life I instilled in her that she was loved and how very important she was.

Evin has such a positive effect on people. I have had numerous people come to me and tell me how Evin made them feel so loved. Others tell me they have gotten involved helping people with disabilities due to the impact Evin has had on their lives. I have even had women who do not have children tell me that being around Evin and experiencing her loving nature made them want to have children.

We live in a world that showcases beauty, intelligence and talent, and our kids must struggle to survive in that world. But when I look at how Evin affects people I believe that she is the one with the above qualities. Evin changes the world's definition of what beauty is.

I don't want to mislead you and make you think that we don't have our struggles, we certainly do. It is not easy battling with all the things pertaining to SMS. But it is the above attitude that makes the battles easier and worthwhile.

I write all this to say please find the good in your situation. Know that you and your child

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have worth. Know that there is a purpose for you, even when it doesn't feel that way. We as parents need to instill in our kids their great value, but we must first believe it ourselves.

Now what does all this have to do with education? I believe everything! Our kids need to believe in themselves and know that they have purpose. When they do they will have greater success in school.

Teacher Perspective

By Christina Miller

Evin walked into my elementary L.I.F.E. skills classroom as a second grade student. It was quickly evident that she had a strong foundation of love and support from home. She always had a smile and a hug for anyone who needed one. She was ready to learn, although at times not always willing.

The classroom was structured to ensure success in learning and develop independence. Icon and written schedules were posted for each child in the classroom. Independent work station tasks, which included basic and fine motor skill activities, were taught to each student in a one-on-one situation until mastery. Once tasks were mastered, they were moved to student desks where they were done independently according to a posted work schedule at the desk.

While the students worked independently, I pulled them one at a time for reading instruction. Writing instruction was done as a small group with the help of paraprofessionals and the district's Occupational Therapist. Math instruction was also presented in a small group setting. Work was modified as needed for each child. Evin seemed to benefit most from color coding strategies.

Evin used a weekly behavior chart which targeted two behavioral goals. This chart was sent home daily and was graphed at every grading period. This helped pin point specific trends in behavior so adjustments in instruction or routine could be made as necessary. Evin displayed occasional behaviors such as work refusal and off-task behavior. Her behaviors at school were never significant. When behaviors would appear over a period of time, Leanne and I would conference in person or over the phone to come up with a strategy. Changes were made and behavior continued to improve.

Over time Evin's confidence in her academic ability blossomed. Her performance in all academic areas increased, as she became more independent and confident.

I believe that Evin's foundation from home prepared her to overcome the challenges faced by children with SMS. She was ready to learn and loves school to this day. She has seen and felt her success at school. She is a fighter with a winning spirit, which will carry her far into her future.

PRISMS Board Visits NIH

By Margaret Miller

On Tuesday, Nov. 12, 2002, the newly inducted board members got a chance to visit the Smith-Magenis protocol and its staff at the National Institutes of Health, in Bethesda, MD. The PRISMS Board had just spent two very intense days brainstorming ideas and engrossing themselves into PRISMS business. A field trip was just what we needed! Ann Smith was kind enough to organize and present this meeting, and even prepared a wonderful lunch for all to enjoy!

The purpose of the meeting was to indoctrinate the board to all of the facets of the protocol and to introduce the board to the “key players” involved in the SMS protocol. We were also given a step by step tour of the different clinical departments that are part of the protocol and presentations from the staff on what takes place during the child’s stay at NIH. The information we gathered that day will help PRISMS to more accurately answer parent questions regarding the protocol and also substantiates the partnership between “parents and researchers.” We now also have names to faces and an easier sense of correspondence with the staff at NIH.

Ann Smith introduced the staff members that were able to attend this meeting. (Not all of the staff of the protocol were able to be present.) Those in attendance were:

Ann C.M. Smith, M.A., CGC
Principal Investigator, NHGRI/NIH

Carmen Brewer, Ph.D.
Audiology, NIDCD

Donna Kraneswich, M.D., Ph.D.
Medical Advisory Investigator

Beth Solomon, M.S., SLP
Speech/Language Pathology, CC

Sue Parada, RN, CC - Nursing
Study Coordinator, Research Nurse

William Gahl, M.D., Clinical Director
National Human Genome Research Institute

Hanna Hildebrand, OTRL, CC
Rehab Med Occupational Therapist

Michaele R. Smith, B.S., PT, CC
Rehab Med. Physical Therapy

Wally Duncan, Ph.D.
Psychologist/ Sleep Researcher, NIH

Ann Smith presented the complete title of this project. It is, “The Natural History Study of the Clinical and Molecular Manifestations of Smith-Magenis Syndrome.” What this means is that this project will study the physical, cognitive, developmental and neuro-behavioral phenotype in SMS through a comprehensive and longitudinal clinical analysis from early infancy to adulthood. Basically, the project seeks to study the characteristics of SMS over a person’s lifespan. This information can only further define a diagnostic criteria and enrich the variety of interventions necessary for improving the quality of life for persons with SMS.

Ann also specified the other goals of the project, such as establishing a cooperative and

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all-inclusive SMS Research & Core Tissue Bank. Also, further studies of sleep disturbance, maladaptive behaviors, speech/language delays and the psychological impact of SMS on the individual and the family will be performed.

Wally Duncan our resident “sleep expert,” gave a fascinating presentation of the tools now being used to research and document the sleep patterns of persons with SMS. The Actigraphy wrist watch is now being used, if possible, on in-patients and also for out-patient use. The Actigraphy watch will document activity/rest patterns for several weeks, and hopefully months, if tolerated. Parents are also asked to keep sleep logs and the Home Assessment of Mood and Behavior, (HAMB). As sleep-deprived families, we are all very anxious to see what results Dr. Duncan will collect and what interventions may come about from this research. Dr. Duncan, Ann Smith, and some of the staff have been wearing the actigraphy watch to provide “normal” controls for the trials.

After the formal presentations, we were taken on a tour of the different clinical departments that participate in the protocol. Michael Smith, PT, gave us a demonstration of the computer-based stride analysis. I was a somewhat willing subject, and we had much fun stepping in the “shoes” of our kids! Beth Solomon, SLP, gave us a presentation of the oral/sensory motor/swallowing evaluation. She demonstrated what “black spaghetti” is and the process of capturing your child’s voice on audio and video. We were able to see laryngeal function up close, very close, and how it ties into proper speech function. This was fascinating! Dr. Carmen Brewer gave us a tour of the audiology department and the sound-proof rooms used to give exams. This had to be the last stop on our tour due to time constraints and board members needing to catch airplane flights. Too bad for us, as we could have asked questions of the staff for days to come!

The most impressive thing about the protocol was not the fact that it takes place at NIH, not the fact that they have the most advanced equipment and research tools, not the fact that you have access to top medical consultants and research. Though all of these facets are awesome, the most impressive features of this protocol are the dedication, sensitivity and compassion of the doctors, scientists, nurses, therapists, technicians and support staff. These are folks with very impressive credentials. They are authors of well-respected papers and medical journals. These are professionals who could probably work at higher paying institutions and in an area with less traffic! But they chose to work at NIH, on the Smith-Magenis protocol, and with our children. Trust me, we are very lucky to have them on our team. I can only believe it is the example of Ann Smith who motivates and attracts these professionals to want to work with our kids. She has recruited the best because she only gives her best.

It was, to say the least, a great day for the PRISMS board and a wonderful way to start off the new term! For any of you families traveling to NIH this year, you are in good hands. Safe travels!

\$\$\$ RAISING FUNDS, RAISING AWARENESS \$\$\$

PRISMS is at the point of evolving to the next level to serve our growing SMS population. PRISMS will be creating an informational video, medical pamphlet and resource guide for both professionals, educators, doctors, donors, and the general public. We are also seeking funding so we can be more proactive in research and in developing behavioral interventions.

Here are a few of our fundraising activities and planned events:

Holiday SMiles Letters - These letters were mailed out to our many SMS families to allow them to solicit donations from friends and family during the holiday season. As of the date of Spectrum's publication, \$9,050.00 has been raised by these letters alone. Great work everyone! Because of the success of this program, we will, of course, continue to accept contributions until we again send letters during this year's holiday season.

Entertainment Books - A near effortless way for PRISMS to raise funds, these books can be purchased online at <http://www.entertainment.com>. Remember to use our PRISMS account number (742332) when ordering in order for PRISMS to receive any profit. If you live in a metropolitan area, these books are a must have!

Paladini Sauces - Achille Paladini of San Mateo, CA has changed the labeling on his Paladini seafood sauces to include information about SMS. In addition to the SMS information, Mr. Paladini is donating a portion of the purchase price of these sauces to PRISMS. Please ask your local grocer for these sauces, they're fantastic. If the sauce wasn't enough, in April, Mr. Paladini will host three days of tours at their nationally renowned Japanese Garden. This has been a successful fundraiser in the past for other charitable causes and it is a huge opportunity for PRISMS to be this year's recipient of the tour funds. Mr. Paladini is a grandfather to one of our SMS children, Samantha Brown. A big "thank you" to the Paladinis for their contribution!

Golf Tournaments - Two golf tournaments are scheduled for this year. One in the Ohio area and the Fourth Annual SMS tournament in New Hampshire. Please see more information on page 15.

eScrip - This is PRISMS newest and perhaps easiest fundraiser yet. Here's how it works: Each time you use your credit card or grocery card with a participating eScrip merchant, a percentage of your purchase, whether at the grocery store, at the mall or on the Internet, will be contributed to our group. To get started participating in this near effortless fundraiser, you must first register your cards. You can do this by going online at www.eScrip.com. In order for PRISMS to get the donations, you will need to enter our group identification number. PRISMS number is 150239751. If you do not have access to the Internet, please contact Sue at the PRISMS office (603) 547-8384 and she will help get you registered. Please encourage all your friends and family to participate!

These fundraisers were all the result of hard work on the part of several individuals, including the Paladini's, Michele Zdanowski, Marvin Armstrong, and many, many others. We, as an organization, will continue to solicit corporate contributions from Pfizer and others, but it is individuals like you who drive PRISMS and provide the energy and resources to keep us going. If you need more information about these events or if you have an idea or project and need help, please contact us here at PRISMS. Please keep up the good work and support these projects if you are able.

Golf Outings



*Great Lakes Region
Smith-Magenis Syndrome
Golf Tournament*

June 21, 2003 (Saturday)
ValleyWood Golf Club
13501 Airport Hwy.
Swanton, (Near Toledo) Ohio

For more information email: GreatLakesSMSGolf@Comcast.net

Or Phone: John & Kellie Cooney at: 330-948-8357

Great Lakes Region SMS Golf Outing Coordinators (and SMS Parents):

• Joe and Michele Zdanowski • Lin & Julie VanNest • John & Kellie Cooney

We are currently accepting any donations for the Live and Silent Auctions, i.e. gift certificates, baskets, art, sports memorabilia, etc.

If you are not a golfer, just come for the live and silent auctions and dinner later in the evening.



4th Annual SMS Golf Tournament

Francestown, New Hampshire

August 2003

(Date to be announced at a later time)

For more information, contact Marvin Armstrong at 603-547-3132 or email: marmstrong@golfarchitect.com



If anyone is interested in organizing a golf tournament in their area to raise funds to benefit SMS, please contact PRISMS for a helpful guide.

MAKE THE CONNECTION

PRISMS is looking for computers! If you have an old computer still in working condition but just gathering dust somewhere in your house or perhaps work for a corporation or organization who may be upgrading their computers and need somewhere to recycle their old ones, let them know about us.



PRISMS would like to take these computers and offer them to our many SMS families who may need one and just can't find a way at this time to fit one in their budget. PRISMS unfortunately will not be able to provide technical support for setting them up in your home, but no doubt you have a relative who can probably do this for you.

If you would like to be placed on a waiting list to receive one of these computers, please contact PRISMS. Computers will be given on a first come, first serve basis.

THANKS!

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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 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 that you want. Here's how, contact:

Mary Beall
414 Birch Lane
Richardson, Texas
Phone: 972-690-1016
mary.beall@attbi.com



Important note:

PRISMS parent to parent program will only give out information on parents who have agreed to be contacted.



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