

# SPECTRUM

## Smith-Magenis Syndrome Caregiver Study: Results and Recommendations

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### Part 1: Caregivers display significant symptoms of anxiety and depression

Stephanie Kozachek<sup>1</sup>, Rebecca H. Foster<sup>2</sup>, Surbhi Kanotra<sup>2</sup>, Marilyn Stern<sup>2,3</sup>, and Sarah H. Elsea<sup>1,3</sup> <sup>1</sup>Department of Human and Molecular Genetics, Virginia Commonwealth University, Richmond, VA. <sup>2</sup>Department of Psychology, Virginia Commonwealth University, Richmond, VA. <sup>3</sup>Department of Pediatrics, Virginia Commonwealth University, Richmond, VA.

Caring for an individual with Smith-Magenis syndrome (SMS) is challenging on a daily basis and is a role that the caretaker often assumes for a lifetime. Although a limited number of studies have shown that families of individuals with SMS experience increased amounts of stress in caring for their child, almost no research has systematically investigated what factors may promote healthy caregiver adaptation or serve as barriers to caregiver well-being (Fidler, Hodapp, & Dykens, 2000; Raina et al., 2005). Due to the lack of

information available to date, this study aimed to explore the many aspects of daily living faced by primary caregivers of those diagnosed with SMS in order to examine parental perspectives, behaviors, and current levels of functioning. This study included comprehensive data collection on a variety of factors including symptoms of depression and anxiety, caregiver well-being, sleep behaviors, caregiver satisfaction and self-efficacy, and career satisfaction.

*Continued on page 4...*

## Embracing the Inner Toddler in People with Smith-Magenis Syndrome

By Brenda Finucane, MS, CGC  
Executive Director, Genetic Services at Elwyn

In my role as a genetic counselor at Elwyn, a large nonprofit agency for people with developmental disabilities, I have worked with hundreds of children and adults who have a variety of genetic conditions, including many with Smith-Magenis syndrome (SMS). My colleague, behavior specialist Barbara Haas-Givler, and I have had the opportunity to meet dozens of PRISMS families in our travels as consultants for school districts and agencies throughout the country. Barbara and I consider it a privilege to be able to work so closely with people who have SMS. We find them interesting, fun, creative, loving, poignant, and always memorable. We gain new insights every day from their dedicated parents and teachers. Like all people with

developmental disabilities, those with SMS and their families face many common challenges related to their special educational, behavioral, and residential needs. Compared to people with other types of disabilities and syndromes, however, children and adults with SMS are among the most challenging we serve. We concur with the many parents who've commented that raising a child with SMS is a unique experience!

*Continued on page 14...*

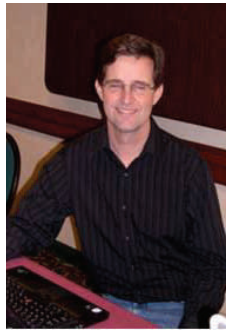


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# A Message from PRISMS President...

## Randy Beall

Parents know quite well the added stress that comes from having a child with SMS. This issue of SPECTRUM is all about *stress*.

In this issue, Sara Elsea reports on her research into the level of stress experienced by the primary caregiver of a person with SMS. She has documented what many of us have experienced, and while that may sound discouraging, it can be so helpful to recognize what is happening so that we can make plans to keep ourselves healthy.

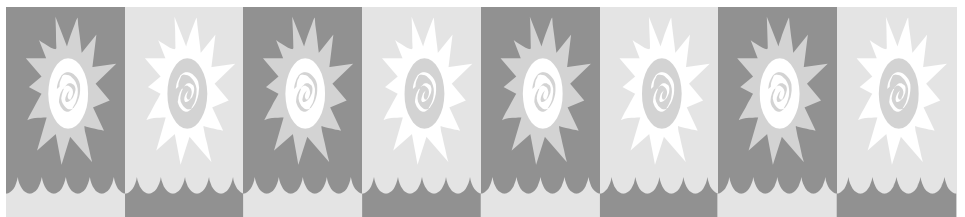
Brenda Finucane explains the reason for some of our children's most confounding behaviors in her excellent article about what she calls our children's "inner toddler". It can help us understand why it seems that our child continues to have so much trouble with emotional control...and understanding can give us more patience ...with our child, and with ourselves.

The PRISMS families are fortunate to have some very caring professionals who understand our experiences and are trying to help. Our Professional Advisory team is working on learning as much as they can about SMS, and publishing that information for us and for our communities. A Sleep Treatment Trial is beginning at the NIH, and you can read about that here also.

PRISMS families can help each other so much with problems and also with celebrating triumphs. Look inside this newsletter to find answers in "The Question Box", to read sweet stories about our darling children, and how you can get involved with the online SMS Discussion List to exchange information with other families.

I hope everyone is having a great summer.

Take care,  
*Randy*



# Meet the PRISMS Professional Advisory Board

## Dr. Andrea Gropman



Dr. Andrea Gropman has been a member of the PRISMS Professional Advisory Board since 2003 and an Associate Investigator on the SMS Research Protocol at the National Human Genome Research Institute, (NHGRI), in Bethesda, MD. Dr. Gropman has followed many of our families and individuals with SMS who have participated in the study, and has been a tremendous resource to PRISMS and its members.

Dr. Gropman is a Pediatric Neurologist and Clinical Geneticist and is on staff at the Children's National Medical Center in Washington, DC. Dr. Gropman is also the Principal Investigator, Children's Research Institute Center for Neuroscience Research (CNR). Dr. Gropman completed her residency training at Johns Hopkins Hospital, and fellowships in neurology and child neurology at the Children's National medical Center and George Washington University. Dr.

Gropman has been involved in research since that time with a focus on neurological phenotypes in neurometabolic and genetic disorders. Dr. Gropman has co-authored several papers and publications about Smith-Magenis Syndrome, including:

**Gropman AL, Duncan WA, Smith ACM (2006).** Neurologic and developmental features of the Smith-Magenis syndrome (del 17p11.2). *Pediatric Neurology* (invited review). 34(5):337-350.

Ann CM Smith, Judith E Allanson, Sarah H Elsea, Brenda M Finucane, Barbara Haas-Givler, **Andrea Gropman**, Kyle P Johnson, James R Lupski, Ellen Magenis, Lorraine Potocki, Beth Solomon (August 2006) Smith Magenis syndrome Overview In: *GeneReviews: Genetic Disease*

Online Reviews at GeneTests-GeneClinics [database online]. Copyright, University of Washington, Seattle. Available at <http://www.geneclinics.org>.

Smith ACM and **Gropman A** (2005). "Smith-Magenis syndrome" In: Cassidy, S.B. and Allanson J (eds). *Management of Genetic Syndromes*, 2nd edition. New York: Wiley-Liss, Inc., pp. 507-526.

**Gropman AL, Elsea S, Duncan WC Jr, Smith AC (2007).** New developments in Smith-Magenis syndrome (del 17p11.2). *Curr Opin Neurol.* 2007 20(2):125-134.

Dr. Andrea Gropman has been a presenter at the PRISMS International conferences, (specifically neuroimaging strategies, and behav-

ior in infants through adolescence with SMS), and has also participated in the PRISMS Advisory Board Research Roundtable. Dr. Gropman is a frequent presenter at professional conferences and university classes on a variety of neurological subjects and has presented at Grand Rounds at several hospitals across the country.

PRISMS is honored to have Dr. Gropman as a member of the PAB, as she brings her countless expertise and experience to the "research table," while sharing her caring and kind heart with our families.

### 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 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.

Results will be presented in three installments beginning with an in depth examination of symptoms of depression and anxiety among primary caregivers, including how to identify symptoms of these disorders and how to seek help.

### Methods and Analyses

Caregivers were recruited between May 2007 and September 2007 via e-mails sent through the PRISMS (Parents and Researchers Interested in Smith-Magenis Syndrome) listserv and Yahoo SMS Listserv. Recruitment also took place at the PRISMS conference in May 2007 (Reston, VA). Those interested in participating completed an online survey, which included a variety of valid and highly reliable measures such as the Beck Anxiety Inventory (BAI; Beck, Steer, & Brown, 1993) and the Centers for Epidemiologic Studies Depression scale (CES-D; Radloff, 1977). For the first installment of results, primary analyses aimed to determine whether the caregiver demographic variables such as gender, age, level of education, num-

ber of children in family, and marital status were significantly related to self-reported symptoms of anxiety and depression. Analyses also explored whether factors such as sleep and counseling directly affected symptoms of depression and anxiety.

### Participants

Participants included 115 primary caregivers of individuals diagnosed with SMS. Among those who responded ( $M_{age} = 41.67$  years,  $SD_{age} = 9.64$  years), most were female (85.2%), white (94.8%), married (80.0%), had other children (85.7%), and had at least some college education (92.0%). Data shows that the caregivers are taking care of their health with the majority attending annual physical (67.5%) and dental exams (69.6%). Responses indicated that participants desired counseling after the diagnosis of their child with 58.3% attending counseling or receiving medical treatment for anxiety or depression following the SMS diagnosis. Most caregivers received social support from their spouse, other family members, friends, medical professional, and/or teachers/school systems. Even though the

respondents were primary caregivers, most were employed at least part-time (73.0%). Many (70.0%) of the caregivers rated raising a child with SMS as one of their most stressful experiences compared to other stressful experiences in their lives (score of 8 or higher on a scale of 1 to 10).

### Correlational Data

Correlations were examined to assess which demographic variables related to symptoms of



anxiety and depression. Endorsement of fewer depression symptoms (i.e., lower scores on the CES-D) was more likely among those who had received counseling both before and after beginning the caregiving role. Those reporting fewer anxiety symptoms (i.e., lower scores on the BAI) also had obtained higher levels of education, reported getting more sleep, had sought counseling after beginning their caregiving role, and had been to counseling within the past year.

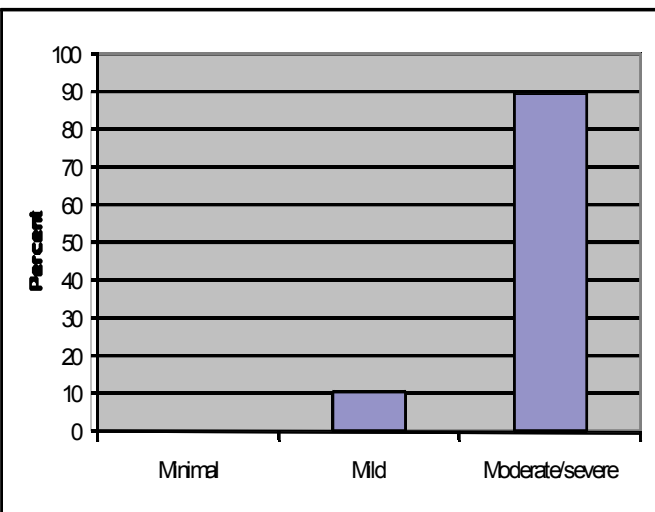
### Primary Results

#### Sleep

Caregivers reported that their sleep is interrupted 5.07 nights per week ( $SD = 2.07$ ) and that they received a total of 5.47 hours of sleep per night ( $SD = 2.54$ ). They reported needing 6.62 hours of sleep per night to function well ( $SD = 8.04$ ) and that ideally they would receive 7.05 hours of sleep per night ( $SD = 7.05$ ). In comparison, experts typically recommend that adults get 7 to 9 hours of uninterrupted sleep per night in order to function adaptively (National Sleep Foundation, 2008).

#### Depression Symptoms

The majority (89.6%) of caregivers endorsed moderate to severe symptoms of depression (see Figure 1). The remaining 10.4% expressed mild symptoms of depression. Caregivers who had engaged in counseling for depression or anxiety anytime after beginning their caregiving role re-



**Figure 1. Symptoms of Depression in SMS Caregivers. Results are based upon the Center for Epidemiologic Studies Depression scale.**

ported significantly fewer symptoms of depression than those who had not sought out counseling. Further analyses demonstrated that counseling after beginning the caregiver role and quality of sleep directly impacted self-reported symptoms of depression in such a way that those who had healthier sleep patterns and those who opted to engage in therapeutic counseling displayed better outcomes in terms of minimizing symptoms of depression.

### Anxiety Symptoms

Data show that 89.6% of the caregivers assessed displayed moderate to high levels of anxiety symptomatology (see Figure 2). Caregivers who had counseling anytime after beginning their caregiving role reported significantly fewer symptoms of anxiety. Respondents who held a bachelors degree or higher reported significantly fewer anxiety symptoms as compared to respondents with lower levels of education. Additional data analyses also showed that counseling after beginning the caregiver role and quality of sleep directly affected the intensity of anxiety symptomatology endorsed by participants in such a way that those with better sleep habits and those engaging in any type of therapeutic intervention were better equipped to minimize their anxiety levels.

### Additional thoughts on depression and anxiety from caregivers

As part of this study, caregivers were openly asked to provide additional thoughts related to their experiences caring for an individual diagnosed with SMS. Some commented directly on experiences related to depression, anxiety, and associated frustrations:

*“It is an emotional rollercoaster that I am only just coming to terms with through intensive counseling. My husband, 15-year-*

*old son, and I are all suffering from depression as a result of living with [my daughter] and her rages and self-injurious.”*

*“It is a lonely, isolating proposition because most people cannot grasp how extremely difficult and tiring raising an SMS child is.”*

*“It was incredibly challenging, especially [ages] 2 years to 11 years. I came very close to complete breakdown, physical, and emotional. If I had not had a lot of financial support, for lots of therapies and resources for [my child] that slowly improved her behavior, I would not have coped. I would have had a breakdown. Terror of what would happen to her in the future motivated me to continue even when it seemed hopeless...which was often.”*

Despite some of these challenges, numerous families additionally commented on benefits they had experienced and how their children continue to enrich their lives:

*“It is by far the hardest thing I do...all day, every day...it's life consuming and life draining. It's also rewarding. Every little accomplishment, from our SMS daughter but also our non-SMS daughter is treasured. We value the things that need to be valued.”*

*“[My daughter] has taught me how to appreciate life while also teaching me that I am a way stronger individual than what I ever thought was possible.”*

*“After having a pity party for myself the first few months after her diagnosis, I learned that me and my husband had strengths and patience that came from deep within.”*

Finally, some caregivers' comments seemed to directly reach out to offer advice and inspiration to other families

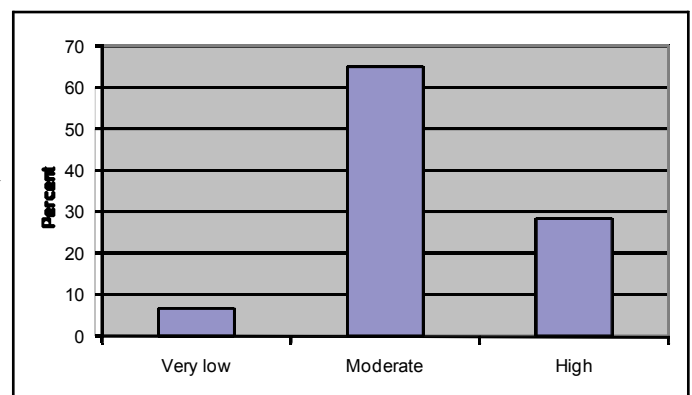
coping with the demands of raising a child with SMS:

*“Ask for help, let someone come in to your house so you can sleep and you don't have to go it alone.”*

*“This is the toughest job I can ever imagine. Sometimes I resent other 'typically developing families' with their insignificant challenges. However, I always remember that it could be worse and remind myself that GOD trusted me with this child. My motto is 'If you're handed it...you can handle it.'”*

### What do these results mean?

The results of this study clearly suggest that caregivers of individuals diagnosed with SMS are at an increased risk of experiencing symptoms of depression and anxiety. However, when considering the meaning of these results, please keep in mind that the measures used for this study are screening tools. Although a large number of caregivers reported heightened levels of depression and anxiety symptoms, self-reported symptoms do not necessarily translate into a diagnosis of an anxiety or depressive disorder. Additionally, while it is likely that the symptoms reported by participants may be worsened by the poor sleep quality experienced by many SMS



**Figure 2. Symptoms of anxiety in SMS caregivers. Results presented are based upon data from the Beck Anxiety Inventory.**

## How can I learn more about anxiety and depressive symptoms?

Further online resources related to anxiety and depression are available online at:

<http://www.webmd.com/depression/default.htm>

<http://www.webmd.com/anxiety-panic/default.htm>

<http://www.mayoclinic.com/health/depression/DS00175>

<http://www.mayoclinic.com/health/generalizedanxietydisorder/DS00502>

caregivers, seeking counseling may be especially helpful in not only alleviating these symptoms but improving quality of life among caregivers and families.

## What should I watch for in terms of symptoms of anxiety and depression?

Identifying and differentiating between symptoms of anxiety and depression and the typical ups and downs of the caregiving role can be extremely difficult. Although only a trained health-care provider can diagnose anxiety and depressive disorders, family members are often quite good at recognizing behavioral changes in themselves and their loved ones that may indicate a problem above and beyond everyday manageable stressors. Below are two checklists that can be used to begin to identify whether you or your loved one *may* be at risk and whether you may want to consider seeking additional help.

### *Symptoms of Depression*

Have you or someone you know experienced five or more of the following symptoms during the same two week period?

- Depressed mood most of the day nearly everyday (e.g., feeling sad or empty, tearfulness)
- Loss of interest or pleasure in activities, things, or people you used to enjoy
- Significant weight loss or weight gain; significant increase or decrease in appetite nearly every day
- Too much sleep or too little sleep nearly every day
- Motor agitation or retardation (e.g., physically sluggish movements, slow speech)
- Fatigue or loss of energy nearly every day
- Feelings of worthlessness or excessive guilt nearly every day
- Inability to concentrate or make decisions nearly every day
- Recurrent thoughts of death or suicide

If the answer is *yes*, you or your loved one *may* be experiencing a major depressive episode. If you checked the last box concerning suicidal ideation, seek help immediately (see next section).

### *Symptoms of Anxiety*

Have you or someone you know experienced both of the following?

- Excessive anxiety and worry occurring nearly every day for at least six months
- Difficulty controlling the worry

In addition to the above, have you experienced at least three of the following within the last six months, and have these symptoms impaired your

ability to complete your daily activities?

- Restlessness, feeling on edge
- Easily fatigued
- Difficulty concentrating, mind going blank
- Feeling irritable
- Tense muscles
- Disruptions in sleep (trouble falling asleep or staying sleep)

If you answered *yes* to both question, you or your loved one *may* be at risk for having an anxiety disorder.

## What should I do if I (or someone I know) am displaying symptoms of depression or anxiety?

Although it can be difficult to seek help for symptoms of anxiety or depression, there are a number of options available that can help alleviate your symptoms in a short amount of time. Consider sharing your feelings and thoughts with those you trust such as a close friend or family member. Having the support of loved ones is crucial in working through your symptoms and lessening stigmas. Consult with your primary care physician, your child's pediatrician, or your child's SMS specialist. Primary care/family doctors and pediatricians are trained to do initial screenings for depressive and anxiety disorders and make referrals



for counseling services (both individual and family services) and/or psychotropic medications. Consulting with an SMS specialist can be beneficial as well. Such a specialist may be able to make a referral to a therapist that is specifically trained to work with families coping with chronic illnesses.

If you or someone you know is feeling suicidal, you need to act immediately. Do not leave the person alone. If you feel the person is in imminent danger, call 911 or take the person to the nearest emergency room. Additional help and advice are available 24 hours per day at 1-800-SUICIDE or 1-800-273-TALK.

### Summary

Over 50% of our respondents had attended counseling or received medical treatment for anxiety or depression since beginning their caretaking role. Data analyses showed that those that have attended counseling and those that get more quality sleep have significantly fewer symptoms of depression and anxiety. Although the demands of raising a family may make it difficult to seek help in dealing with symptoms of anxiety and depression, seeking out this type of help may greatly improve individual coping abilities and ultimately improve the entire family's long term quality of life.

### In future issues of Spectrum . . .

Part II: SMS Caregiver Wellbeing

Part III: SMS Caregiver effects on Education and Career

### Acknowledgements

We would like to thank all of the caregivers who took the time to complete our survey. Your time and efforts have provided a wealth of

information that will allow us to inform other caregivers and healthcare providers of the unique needs of families coping with SMS and develop innovated approaches and interventions aimed at serving SMS families in the future. This study was supported, in part, by a generous funding from an anonymous donor who saw the difficulties faced by a friend whose child has SMS.

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## Save the Date

**PRISMS 6<sup>th</sup>  
International  
Conference**

**"BUILDING  
BRIDGES OF  
HOPE"**

**Sept. 17-20, 2009  
Reston Hyatt Hotel  
Reston, VA**

\*This is the same site as the 2007 conference. Our families loved the location and were able to easily enjoy the shops and restaurants in immediate walking distance. We hope to see you all return AND we welcome new families and faces to the 2009 conference. Start planning and saving now! Be a part of the celebration!!

PRISMS 6th INTERNATIONAL CONFERENCE  
Reston 2009

building bridges  
of hope





# Church and the SMS Child

## *A Parent's Story by Ken Bolt*

We have attended the same church now for over 20 years. If you want an example of love and tolerance, you'd have to come to our little church. Until recently, we had a separate contemporary Praise and Worship service. Since my oldest son, Daniel, plays keyboard in the praise band, we opted to attend that service. Jeremy, our SMS child, is ten years old and we also have a beautiful daughter, Sarah, who is 14. Many of you know Jeremy as the red-headed "celebrity" on the SMS bookmark.

Occasionally, I would be asked to collect the offering. Well, the first time I got up to pass the plate, Jeremy jumped up and "helped" me. He grabbed the plate and flew down the aisle. I had to get a hold of his collar to keep him slowed down. Of course, some folks didn't have their checks quite ready. On one occasion, Jeremy blurted up "Hurry UP!" Since everyone knows Jeremy, they just laughed. On another occasion, someone put a check in the plate and Jeremy grabbed it, staring at it. It was like he was going to say, "is that ALL?" Thankfully, he put it back after I told him to.

After we finished, we'd deposit the plates in the last pew and return to our seats. Jeremy, of course, ran the length of the aisle full-tilt. No matter how many times we'd tell him to slow down, I think he had it in his mind to set the world speed record for offering collections!

Now, we have combined our services into a "One Voice" service. So, we have a lot more folks and many of them are older. Jeremy has joined the Children's choir. In their first per-

formance, he got part way through the first song, before something set him off and he ran down the aisle crying. I, of course, had to follow him out to try and calm him down. We ended up, appropriately enough, in the "Crying Room". I took his robe off and told him that his choral career was over. He instantly stopped crying and said "I be good". So, I told him that he could go back out. We put the robe back on and he ran, flat out, down the main isle to the front of the church during the second song.

And, do you know what happened...the whole congregation gave him a standing ovation!

When the diagnosis came in, some 8 plus years ago, we asked the question "Why?" like so many other parents of special needs children. I think I know the answer now. Jeremy brings smiles to people who otherwise might not smile. We go to the local nursing home a couple of times per month and the ladies just love him. He doesn't really notice a lot of the disabilities of those elderly folks, he just loves them as they are.

Isn't that what we're ALL supposed to be about? We can learn from people like Jeremy.

Most every Sunday, at the close of the service, people are invited to come up to the altar to pray. Jeremy is usually the first one to arrive and he always asks me to go with him.

And, every Sunday as I kneel there, I thank God for the gift of Jeremy. He is truly a blessing to me and to others.✱



# Do You Have an SMS Question?

Randy Beall ✧ PRISMS, President ✧ SMS Discussion Moderator  
[prismsbb@prisms.org](mailto:prismsbb@prisms.org)

## The PRISMS SMS Discussion List on our website has now been improved!

We are now offering an email digest option to make it easier to participate on this forum. Digests are email summaries of messages posted on the board. Digests can be sent daily or weekly and at an hour of the day you select.

You specify those particular sub forums (such as daily chat, managing behavior, adults with SMS, etc) for which you want message summaries, or you can easily elect to receive all messages for all forums for which you are allowed access. (Since this board is just getting started, you might want to select all at this point.)

Consistent with our privacy policy, digests contain no "spam", nor is your email address used in any way connected to an advertisement. You can, of course, cancel your digest subscription at any time. Most users find digests to be very useful. We encourage you to give it a try!

Excerpts from posts are presented with a convenient link back to the original message if you want to read more.

This discussion board is only as good as the posts that we receive. Please share your SMS experiences. How's it going? What issues are you facing? What's working? Maybe consider answering someone else's post. We need to support each other and share our mutual experiences.

It's easy to get to the PRISMS SMS Discussion List. Simply go to the main <http://www.prisms.org> website and click on the icon on the lower right hand side for

SMS Discussion.

Once you've registered and logged in, there are 2 links at the top of the SMS Discussion home page. One says "profile" (where you can change the basic registration settings) and the other one says "digests" (where you can change your email digest settings).

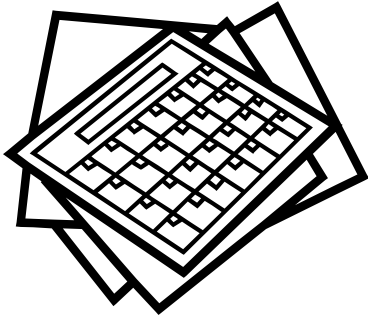
Click on the link that says digests and you can see your settings for email digests. There's also a link below.

<http://prisms.org/phpBB2/digests.php>

Please join the discussion and post something today! ✧



# It's time to CELEBRATE and EDUCATE!!



We have dedicated the month of September as **SMS Awareness Month**. **Celebrate** all the wonderful attributes and talents of our SMS'ers while **educating** our community, school, churches, medical facilities, families, friends and strangers! Let's take our mission "to the streets," and teach the world about SMS. Spread the joy that is SMS and create a community of greater understanding, acceptance, and support. Let's all contribute to the building blocks of our "Bridges of Hope," and help design a future that readily understands Smith-Magenis Syndrome and regards our children as precious and important!

As part of the SMS Awareness Month, we are having another photo contest! We would like you to send in photos, (digital is best), of all your family, friends, neighbors, etc., dressed in their grandest PRISMS wear. Any photo that shows your family or community spreading awareness of SMS is eligible for the contest. Here are some other ideas:

- ✓ Pass out PRISMS brochures in your church or school, or place of work.
- ✓ Present a lecture or power point presentation about SMS at your child's school.
- ✓ Hold a coffee for neighbors to educate them about SMS and your child.
- ✓ Get the siblings involved! Perhaps the sports team or school clubs they participate in would love to show their PRISMS style.
- ✓ Plan a get together with other PRISMS families in your area and celebrate your children.
- ✓ Wear your best PRISMS wear, have a party and rejoice in

your SMS child! Let them know how truly amazing they are!

Send us your photos either digitally, or mail the photos to the PRISMS address. Photos will be accepted until December 1<sup>st</sup>, 2008. **The winner of the best SMS awareness photo will receive a free night's stay at the Reston Hyatt Hotel during the September 2009 PRISMS Conference in Reston, VA.** (The McGrevy's of OH were our winners last time).

So, take it to the streets, "say cheese," and spread the message of SMS! Send photos to:

[info@prisms.org](mailto:info@prisms.org)

Or  
**PRISMS**

21800 Town Center Plaza  
 Suite #266A-633  
 Sterling, VA 20164



# Farewell and Thank You!

Many of our families have come to know Rebecca Morse over the past six years, and we sadly say good-bye to Rebecca as she begins her PhD study in Applied Developmental Psychology next fall.

Rebecca has worked with the SMS Research Team at the National Human Genome Research Institute, (NHGRI), as the SMS Study Coordinator. Rebecca also worked on analyzing hundreds of hours of data from the actigraphy watches, as part of the Home Assessment of Sleep and contributed to several abstracts on the research of Smith-Magenis Syndrome, including hyperacusis, sleep patterns, and family function-

ing and coping skills. She came to know each family that participated in the research study and was a welcoming face to our families and children.

Rebecca was also a part-time “babysitter” for the children who came to NIH, and needed a break to go to the playroom! Rebecca was a calm and helpful voice on the phone when families called with questions and concerns, and made them feel at ease.

Rebecca came to three of our conferences and pitched in and presented wherever we needed her. We are grateful for her kind dedication to our families and she will be

missed. But, we wish Rebecca the very best and look forward to what new insights and research she may be able to share with PRISMS, whilst she is involved in her new studies. Well done Rebecca and thank you for all of your dedicated work!



*Rebecca Morse and Ben Popper,  
(PRISMS Conference, May 2007)*

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## SMS Sleep Treatment Trial Begins at NIH

In April, 2008, researchers at the National Institutes of Health (NIH) began active enrollment for the first controlled treatment trial to determine if bright light alone, or in combination with a melatonin tablet preparation, is effective in treating the sleep disturbance in children with Smith-Magenis syndrome.

The melatonin tablet was developed by the Pharmacy Development Services at NIH and was approved last fall by the FDA (IND 77,802) for use on this study. Sponsored by the National Human Genome Research Institute (NHGRI) at NIH, the treatment study is conducted by the SMS interdisciplinary research team led by co-principal investigators Ann C.M. Smith, MA, D.Sc.(Hon) NHGRI/NIH and Wallace Duncan, PhD, NIMH/NIH and Wendy Introne, MD, Medical Responsible physician, NHGRI/NIH.



Children with a confirmed SMS diagnosis (del 17p11.2) who are between 5-16 years of age, have not reached puberty, and are seizure free may be eligible to participate in this study.

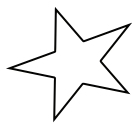
Since certain medications may exclude individuals from participation, a telephone screen to review your child’s medical history and all current medications (both prescribed and over-the-counter) will be conducted as part of the eligibility review. Partial funding is available to offset travel costs to/from NIH (based on standard government rates).

Parents interested in learning more about this treatment study should contact:

Ann C.M. Smith, MA, D.Sc.(hon),  
Principal Investigator  
Office of Clinical Director, National  
Human Genome Research Institute,  
NIH  
Bldg 10, Room 10C103, 10 Center  
Drive, MSC 1851  
Bethesda, MD 20892-1851  
Tel 301-435-5475  
Email: [acmsmith@mail.nih.gov](mailto:acmsmith@mail.nih.gov)



# SMS SuperKid!



## Connie Luo



Connie is 14 years old and is attending a private school serving children with special needs. Her school, the Achievekids, teaches youth with complex emotional and developmental disabilities the skills and knowledge necessary to achieve greater independence. Expert staff, supported by the community, provide programs for the students and families that are positive, individualized, and multi-disciplinary.



**Connie showing off her Super Kid T-shirt (actually she calls herself Super Girl whenever she wears this shirt).**

Overall the school has about 80 students from the San Francisco Bay Area. We are very lucky that the school is in our immediate neighborhood (right across the street from our house!). Connie is the only girl in her class of 8 students. Her class has 6 staff members. Because of this, Connie is really spoiled by her teacher and aids. The staff found out a great way to discourage Connie from pulling out her finger nails and toe nails, which is to paint her nails with nail polish if she keeps them intact!

Of course they change the nail polish colors almost every day to make it exciting.

Connie loves colors. She works on art projects at school, together with her “buddy” Tulin, the school’s Development Director. According to Tulin, these art projects are some of her favorite times with the students. As they get creative, the students get talkative, even chatty. Connie would talk about colors, ask about others’ favorite colors and share her favorite of that day. For this piece of work, they drew the star together. Connie cut it out and then Tulin helped her hold it down while Connie painted the rainbow. She really spent a lot of time choosing colors to make sure that there was enough all the way around the star. This piece of art work was selected to be on the cover of the Annual Report of the Achievekids. It was also the front page of the official invitation letter for the school’s annual fundraising luncheon with the theme of “True Colors” on March 5, 2008. ✨



**Connie’s art**



*(A beautiful color version of this art can be viewed once this newsletter is available on-line).*



Do you have an SMS SuperKid? We would love to publish a picture and a story about your child. As we all know, even small accomplishments are encouraging for our SMS families. Please consider sharing your good moments with us. If you have questions or need help with the story, please call the PRISMS editor, Julia Hetherington, at 843-521-0156, or contact through email at [editor@prisms.org](mailto:editor@prisms.org)

# The Question Box

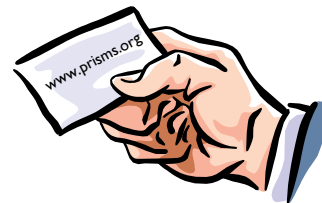
**My child has had frequent melt-down's in public, resulting in people standing around staring and sometimes making rude comments. How do you handle this?**



*“I recommend carrying the PRISMS bookmark or brochure. They can be kept in your purse or car. PRISMS can provide bookmarks, and the brochure can be downloaded from the website. I would get some for everyone who transports the SMS child.”* Bridget

*“Create a business card with simple information about SMS and suggestions, and possibly the PRISMS website listed on the back. Once people understand they tend to be less judgmental and often time even a bit helpful.”*

Stacey, mom to Brett, 18 years old

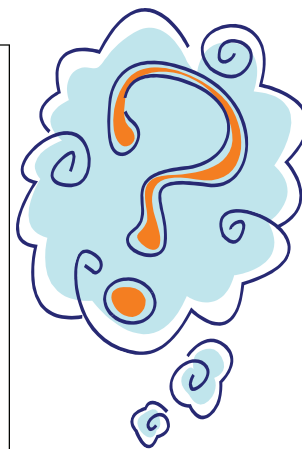


The Question Box is a new feature of Spectrum. Please email your questions/suggestions/answers to [editor@prisms.org](mailto:editor@prisms.org)

Fax: 843-986-9369

**Question:**

**“What are effective strategies for interrupting obsessive-compulsive behaviors, without getting into a battle or raising the anxiety?”**



What is it about SMS that makes this condition so challenging? One obvious answer is that SMS is associated with self-injurious behaviors, such as head banging and picking at fingernails. While these behaviors are distressing, parents rarely report self-injury as the most difficult aspect of raising a child with SMS. Significant self-injury doesn't usually take place on a daily basis, and these behaviors tend to occur in cycles with varying severity. Another possibility is that having a child with intellectual impairment can be a source of stress and sadness for parents. This is true, but no more so in SMS than for parents of other children with special needs. In fact, there is a wide range of functioning among children with SMS, with many having only mild intellectual disabilities and good communication skills.

**.....attention-seeking behavior by itself is not usually as problematic as the reaction**

Another potential way in which some people with SMS may pose extraordinary challenges lies in their insatiable need for individual attention. Attention-seeking behavior can be exhausting for caregivers. People with SMS are often in competition with siblings or peers for attention, and they tend to monopolize a parent's or teacher's time. Still, attention-seeking behavior by itself is not usually as problematic as the *reaction* of the person with SMS when attention is withdrawn and/or when things don't go exactly as expected. This heightened reactivity to situations and environments is almost universal among people with SMS, in our experience. The behavioral reactions of people with SMS often seem like those of a very young child, regardless of the age or intellectual level of the person. Parents often report that they feel like they are "walking on eggshells", sensing that their child is smoldering under the surface and ready to erupt into a major outburst at the least provocation. By necessity, parents often become the best experts on managing their children's behaviors because they've learned to adapt their own be-

haviors and reactions to avoid potential outbursts. They become hyper-vigilant to potential triggers and develop an innate sense of how to phrase a request. They often know just how hard to push and when to diffuse a situation with humor or distraction. Unfortunately, such nuances are very hard to teach or even explain to people unfamiliar with SMS.

### Developmental asynchrony

In the special education world, the term "developmental asynchrony" is occasionally used to describe unevenness in the intellectual and socio-emotional development of gifted children. It has been observed that some children with very high IQs can at the same time be socially and emotionally immature compared to their same-aged peers. Although this observation has not previously been described in SMS, we have observed a similar phenomenon in these children and adults. In SMS, intellectual development is generally slower than that of typical children, but emotional development is even more delayed.

With age, a gap develops between the intellectual attainment and emotional development of people with SMS, and this disparity poses significant behavioral and programmatic challenges. In our experience, this discrepancy is at the heart of what makes SMS so challenging.

While there is a wide range of functioning among people with SMS, most have mild to moderate intellectual disabilities and can master many skills at the 6 to 8 year old level, if not higher. By contrast, the emotional development of individuals with SMS is usually much younger, with many reacting like toddlers in the way they process information and react to their environment. Many aspects of the SMS behavioral profile could equally describe the development of typical 2 and 3 year olds, including: a low tolerance for frustration; negativity; mood swings; a need to do things for themselves; short attention span; tantrums; anxiety about separation from loved ones; resistance to changes in routine; and repeated (unrelenting!) questioning. Typically-developing tod-

dlers often become emotionally attached and possessive of caregivers, need frequent reassurance about upcoming events, and have a poorly developed sense of time, all common SMS behaviors. Behavioral outbursts in adults with SMS can look very much like the "temper tantrums" seen in toddlers, with a person throwing herself to the floor, kicking and crying. Some of the more unusual behaviors observed in SMS may also have their roots in normal early childhood development. For example, many typical toddlers go through a period where they insert objects into their noses or ears. This behavior is seen in about a quarter of people with SMS, usually starting in late childhood and sometimes persisting through adulthood. Another very common behavior in children and adults with SMS is the "spasmodic upper body squeeze", a tic-like gesture that often takes the form of self-hugging or hand-squeezing. In people with SMS, this endearing behavior usually occurs in response to happiness and positive excitement. We consider this a positive aspect of the excitable SMS personality, and although it may seem unusual in an older child or an adult, a similar behavior can be observed among typical young toddlers when they are excited (figure 1).



Figure 1. This photo of a presumably typical child engaged in a full blown SMS self-hug is featured on a Hallmark greeting card

## Implications for Intervention

In our experience, developmental asynchrony seems to be more common among people with SMS than it is in most people with developmental disabilities, including those with other genetic conditions. A person with Down syndrome, for example, who functions intellectually at a 6 year old level also tends to react emotionally like a 6 year old. Unfortunately for people with SMS, regardless of intellectual level, their emotional development is typically at the 1 to 3 year old level which tends to be a difficult emotional age. (There's a reason why they call this period the "terrible twos"!)

The good news is that knowing about developmental asynchrony in SMS can provide us with important insights into intervention.

It is no coincidence that parents of children with SMS frequently look back on the early school years as a "golden age" of development and learning. While parents of preschoolers are adjusting emotionally to the reality of the SMS diagnosis, their children are learning new skills and developing at a slow but steady rate. Most children with SMS seem to be in sync with the preschool and early elementary environment. The school day is filled with hands-on, high interest activities of relatively short duration, and the classroom is filled with bright visual cues, schedules, and reminders. Teachers working in such settings have backgrounds in early childhood education and are experts in the use of distraction and redirection to keep toddlers engaged and happy. They avoid known behavioral triggers and emphasize unconditional positive regard. As children with SMS age through later elementary, junior high, and beyond, the world is not quite so adapted to the very young side of their emotional style. Class periods lengthen, colorful visual organizers, charts, and schedules all but vanish, and there is an increased emphasis on verbal instruction. Few staff in the upper grades and adult programs have a background in early childhood approaches. Behavior plans generally involve consequence-based strategies which may work

well for other students but frequently backfire in SMS.

As a child advances in academic areas, it often creates an expectation that the child's overall development should match up. A 13 year old who reads well at a 4th grade level may be expected to sit through a 50 minute Language Arts class with an emphasis on auditory learning and writing. Unfortunately, if the student has SMS, her "inner toddler" is unlikely to tolerate such a format. She might do better attending half the class and working on a related, hands-on activity in another setting for the remainder of the period. Likewise, even though many adults with SMS are quite capable of performing work tasks, they rarely adapt well to a typical 40 hour work week. Unlike many other adults with developmental disabilities who thrive in such settings, they often have difficulty staying on task without constant one-to-one support and attention. Emotionally and developmentally, adults with SMS often do best with a more eclectic work schedule, one with a consistent routine of different high interest activities in a variety of settings. Variety is the spice of life for many people with SMS! For example, one successful young man's schedule includes three afternoons a week at a traditional work setting, two mornings a week volunteering at the local animal shelter, time spent on chores and errands, and daily workout sessions at the local gym. His schedule is structured (i.e. certain tasks consistently occur at the same time in his weekly routine), but his activities are varied and motivating.

Acknowledging the young emotional level of people with SMS in no way implies that one should continue to treat them like toddlers as they age. Behavior plans that ignore a person's "inner toddler" cannot succeed, but strategies that neglect the person's chronological age and academic abilities do not promote growth and learning. A combined approach that incorporates fundamental practices of early childhood education, "age-progressed" to match the interests and skills of the older child or adult with SMS, is often the key to success in

working with this population. \*

*Brenda Finucane is the Executive Director of Genetic Services at Elwyn, a large nonprofit corporation in Media, Pennsylvania that provides day, residential, and consultative services for children and adults with developmental disabilities. For more information about Elwyn's services for people with SMS and other conditions, visit [www.elwyngenetics.org](http://www.elwyngenetics.org). Brenda is a co-founder of PRISMS and a member of our Professional Advisory Board.*



Brenda Finucane and Barbara Haas-Givler, consultants to the stars!

## 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 that you want. Contact:

Mary Beall

Phone: 972-231-0035

[mary.beall@tx.rr.com](mailto: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.*



Parents and Researchers  
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**Editor in Chief:**

Julia Hetherington  
editor@prisms.org

**Editors:**

Randy Beall  
Ann C.M.Smith, M.A., D.Sc (hon)

**Newsletter Committee Members:**

Margaret Miller

**SMS SuperKid!**

