

## SPECTRUM



## Dental and Craniofacial Findings in Smith-Magenis Syndrome

By Natalia Tomona, D.D.S.

*This article summarizes research conducted as part of the ongoing IRB-approved NIH protocol 01-HG-0109, Natural History of Clinical and Molecular Manifestations of Smith-Magenis Syndrome (SMS) by the NIH SMS Research Team led by Principal Investigator Ann C.M. Smith, MA, DSc(hon).*

This study was published in the American Journal of Medical Genetics Part A 140A:2556-2561 (2006). It is based on the comprehensive craniofacial and dental assessment from 15 SMS subjects with confirmed diagnosis of common 17p11.2 deletions at the National Institutes of Dental and Craniofacial Research at the National Institutes of Health (NIH protocol 01-HG-0109). Children ranged in age from 4 to 19 years. Extraoral and intraoral examination, 3D photographs (3dMD®), and radiographs were taken. Our findings are summarized below with recommendations for parents and clinicians caring for children with SMS.

A strikingly high incidence of developmental dental anomalies was present in our 15 cases. The presence of missing teeth (86.67%) greatly exceeds the 3.5%-8.0% prevalence rate in the general

population. The most frequently missing teeth were premolars, with the second lower premolars most commonly missing. Because these teeth are the last to develop they may have a greater predisposition to developmental alterations. Taurodontism (teeth with "bull-like" roots containing large, elongated pulp chambers) was also a common finding (86.57%) in both primary and permanent molars. Root dilacerations (an abnormal angulation or curve in the root or crown of a formed tooth) were present in 5 of 115 (33.3%) cases. The high prevalence of these dental anomalies may facilitate diagnosis of SMS, particularly in preschool children, when diagnosis of SMS can be problematic. Keeping regular dental visits is important, so your dentist can recognize any of these dental anomalies.

We found an increased number of caries, lesions, or restorations in our SMS cases. The number of teeth with current or past dental lesions in our 14 to 19 years old subjects was higher (range 4 to 9) than the values reported by the World Health Organization for the US population, that

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### 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.*



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## A Message from PRISMS President...

Randy Beall



I don't know about you, but I'm still on a high from our 5<sup>th</sup> International Conference held this past May in Reston, VA. We met many new friends and re-aquainted ourselves with families we had met at previous conferences. I want to personally thank all our donors! It takes a lot of money to put on these conferences and we depend on your generous support. Also, a big thanks to our speakers, conference committee and volunteers, professionals and attendees!

This year's Silent Auction raised almost \$8,800. Wow!! I want to extend a huge thanks to Barry Woodhouse for an awesome job with such a great event! Also, a big thanks to all of our silent auction donors! Our first conference silent auction was held in 2002 at our Denver conference and now it's a conference tradition – and a terrific fundraiser! Speaking of raising money, PRISMS board member Percy Huston spoke on Sunday about how fundraising allows us to have conferences and helps us keep costs low. Please consider helping us. Percy can be reached at

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

Birdies for Charity is a great way to get involved.

This year's John Deere Classic PGA event is July 9-15. Just ask friends and relatives to pledge a flat amount or an amount per birdie. For more information see the [birdies-forcharity.com](http://birdies-forcharity.com) website.

Also, Charlie and Tina McGrevy (and their son Garrett) are hosting a 5K Run and Family Carnival on August 25 in Springfield, OH to benefit PRISMS. They are collecting profiles of our kids so folks can better understand SMS. Please email [cmgrevy@yahoo.com](mailto:cmgrevy@yahoo.com) for more information.

This was my family's 4<sup>th</sup> conference and we've learned important lessons with each one. While some folks believe the conferences are primarily for newly diagnosed families, my family has always learned valuable information, met amazing people and felt re-charged after each conference.

Finally, please think about ways you can get involved and support each other – post conference. Our group is "all about" providing information and support. SMS is a journey and we so desperately need to support each other.

Have a great summer and try to enjoy your precious kids.\*

**Randy**

# Savor The Moments

By Mary Miller

As the grandmother of an SMS child, Deirdre, age 17 (daughter of my stepson Scott and his wife Maggie), I was excited about attending this year's conference. Several years have passed since I experienced the first two PRISMS conferences. And this time my mother (Deirdre's great grandmother) would be my roomie!!

What I remembered from the first conference, 10 years ago, was how moved I felt in the presence of so many SMS children. Up to the moment of check-in, Deirdre was the only child I knew with the characteristic features and husky voice of SMS. The opening moments of the 1997 conference made the strongest impression. Deirdre was not the only child who struggled!

Fast forward to 2007! I wondered if there would be a similar moment—a revelation, a reassurance—something that would speak to my heart as well as my head? Well, yes!! It was early Friday morning –breakfast (wasn't the food good?) and then a time of welcome. We all looked for a place to sit, introduced ourselves around the table and initiated new friendships. And there it was— "out of the blue" - a warm and defining moment coming from two sides!

To my right sat the parents of a 13 year-old daughter who had waited 8 years for SMS to be correctly identified. During the years of searching for a diagnosis, one doctor had commented, "It doesn't matter. You know how she is." Sitting at the PRISMS conference, her mother adamantly disagreed! "But, it does matter! You get all this" - as she swept her arm around the room to include the parents, the doctors and researchers, the speakers, the information—and yes, the other SMS children.

To my left sat the father and grandmother of a 2 year-old girl, diagnosed at birth!! A doctor had noticed facial features and ordered a FISH test. I literally had goose bumps! The family knew at birth! Reflected in stories from these two families was the heart and mind of PRISMS.

## **Connection matters!!**

At that precise moment I wondered "Scott and Maggie, do you know how much you have helped families all over the globe?" I know it wasn't done alone, but the two of them were so central to the very beginnings of PRISMS. In 1990 when Deirdre was born and later diagnosed, there was no such organization. No source of instant

connection with other parents and medical professionals—no SPECTRUM newsletter. The year 1990 was the same year that Ann Smith moved from Colorado to Northern Virginia, Scott and Maggie's "neighborhood." I'm not sure whose kitchen table they sat around, but the support for parents, et al, everywhere began to take shape.

At the 2007 conference, SMS families told me how much they love Scott and Maggie. I know how hard they have worked these 17 years and how committed they are to keeping PRISMS strong. Patrick, Deirdre's 13 year-old brother, is following his parents lead as evidenced in his presentation at the Saturday morning Sibling Session. The extended Miller family loves and admires each of them so much! It appears that the SMS family feels the same way!! Their legacy of care, compassion and counsel is, indeed appreciated and treasured. I heard it myself in the stories of just two families. I am certain there are countless others. And that reality is truly something to savor!!\*



# Research updates from Dr. Elsea's group

## **Smile!**

**Maria Gyure, VCU genetic counseling graduate student**

We would like to thank all of those who participated in our study investigating the usefulness of considering a child's smile during a genetic diagnostic evaluation. We have collected many smiles from many children with a variety of genetic syndromes and were able to successfully design a survey for geneticists which is already starting to show a few interesting trends.

So far, we have found that although a smile might not be particularly helpful in diagnosing Smith-Magenis syndrome, other conditions such as Down syndrome and Williams syndrome may benefit.

Another trend we are seeing is that although it appears geneticists have a difficult time diagnosing specific conditions based on smile alone, they are able to correctly identify the smiles of children without a genetic condition. This suggests that perhaps smile is helpful in an evaluation to aid in determining if a condition may be present, but it must be used in combination with all other features of the child to correctly and accurately determine any condition that may be present.

We hope these studies will be useful in the characterization or new genetic syndromes as we seek to identify other genes that cause SMS-like disorders.

## **Caregiver Study**

**Stephanie Kozachek, VCU genetic counseling student**

We are interested in issues that face the primary caregivers of children with Smith-Magenis syndrome. We know that caring for a child with SMS can be stressful, so we would like to explore the issues parents and primary caregivers encounter regarding their personal lives. Our main focus is to learn about different aspects of caregiver health, including exercise, diet, and sleep, to annual physician and dentist visits. We are interested in your physical and mental health, and we hope to learn more about the causes of your stress and how you cope. Another focus area is how you feel about your quality of life. We would like to know more about your life goals and if they have been fulfilled. Other interests include the benefits you see from caring for a child with SMS and the amount of help you receive from family, friends, school systems, and respite services.

By learning more information about these topics, we hope to increase awareness in the medical community, the SMS community, and the general public. Your health is an important part of taking care of others. This awareness will hopefully lead to an increase in the assistance you may seek from schools, respite services, and friends/family.

This project is a collaborative effort with the VCU Dept. of Psychology. Please contact Dr. Elsea for more information: sel-sea@vcu.edu

## **The Role of *RAI1* in Smith-Magenis Syndrome**

**Santhosh Girirajan, VCU Human Genetics graduate student**

Our research focus is to understand the clinical and molecular role of the retinoic acid induced 1 (*RAI1*) gene in Smith-Magenis syndrome. We have been identifying 17p11.2 deletions that include the *RAI1* gene, as well as mutations in *RAI1* in cases that do not have a chromosome deletion, using various molecular genetic techniques. In the process, we have been able to correlate the SMS clinical features based on the deletion size or mutations in *RAI1*.

We have identified and evaluated 12 individuals with mutations in the *RAI1* gene and ~25 patients with different sized SMS deletions. Our analysis indicates that the vast majority of the clinical features seen in persons with SMS are primarily due to disruption of *RAI1* function. Our data also show that *RAI1* has a small role in features such as reduced muscular tone and speech and motor delay but we conclude that other genes in 17p11.2 contribute to the greater severity and variability of these features in those cases of

*Continued on page 5.....*

17p11.2 deletion. Interestingly, *RAI1* appears to have no contribution toward short stature, cardiac defects, and kidney and urinary tract abnormalities which are often seen in cases with 17p11.2 deletion, suggesting that other genes in 17p11.2 cause these abnormalities.

This project would not have been possible without the help and encouragement of SMS patients and families, collaborators in the US, Europe, and Australia and referring physicians and geneticists.

**Identification of New Genes for SMS-like Disorders**

*Stephen Williams, VCU Human Genetics graduate student*

SMS shares many of the physical and developmental characteristics of other genetic disorders ranging from Down syndrome to Prader-Willi syndrome and beyond. Even within SMS itself one can see a broad range of variability. However, we have identified a large group of children who share many SMS characteristics but do not have a 17p11.2 deletion or a mutation in *RAI1*. Thus, we think other genes are involved. So we are looking for new genes that might play role in SMS-like disorders.

In collaboration with Eli Hatchwell from the State University of New York, we are using an exciting new technique called "array Comparative Genomic Hybridization" (aCGH) to evaluate these cases. aCGH is

a method by which we are able to scan the entire genome of an individual to search for copy number variations (i.e. duplications and deletions). We are now screening DNAs from cases that were referred to our lab for *RAI1* evaluation but in which no abnormality could be detected. Thus far we have evaluated ~20 cases for genomic variation using aCGH and will be following up these studies with another 30 patients. Hopefully, these results will give us insight into new genes that play a role in the development of persons with SMS or an SMS-like disorder.

**Developing Molecular-based Diagnostic Tools for SMS**

**Diagnosis**

*Lily Truong, Charles Sturt University (Australia) Biomedical Sciences graduate student*

The incidence of SMS is estimated at 1 in 25,000 births, which is believed to be an underestimation, in part due to the reliability of currently available diagnostic methods. Classically, SMS has been diagnosed by either cytogenetic analysis or fluorescent *in situ* hybridization (FISH). In most cases, these diagnostic methods are adequate to give a definitive SMS diagnosis. How-

ever, there are instances where deletions can be missed based upon the limiting factors of these tests.

In order to ensure the earliest possible diagnosis and hence allow for early intervention of the clinical manifestations of SMS, we have set out to develop a highly efficient, rapid and reliable molecular-based diagnostic tool that can be used to identify individuals with SMS. We have been successful in adapting a technique known as real-time PCR to measure relative gene copy numbers. The SMS real-time PCR assay specifically amplifies a region within the *RAI1* gene and thus, can be used to determine the 17p11.2 deletion or duplication status of an individual.

We will continue to examine the viability of real-time PCR for routine SMS diagnosis, as well as any other SMS related platforms for which this technique may be adapted. This project has been an ongoing collaborative effort between the labs of Chris Blanchard, Ph.D. (CSU), Sarah H. Elsea, Ph.D. (VCU), and Ann Smith, M.A., D.Sc.(Hon) (NHGRI/NIH).

✱

**Have you updated your address book?**

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Figure: Presence of dental plaque and gingival inflammation around teeth as a consequence of inadequate oral hygiene habits.

is, 1.7 for ages 5-17 and 6.6 for ages 18 years and older [WHO Oral Health Country/Area Profile Programme]. Therefore there is a need for constant oral health care and maintenance with help of parents as the child with SMS gets older. Use of fluoride toothpastes and regular dental check-ups at least twice a year are recommended. A balanced diet, low in sugar and rich in fruits and vegetables is also important to preserve the oral health.

The poor oral hygiene associated with gingival inflammation observed in the older individuals with SMS suggest a need for more dental care in the adolescent years. These findings are consistent with the inverse relationship reported by Martin in 2002 between the child's age and performance of daily living skills. Consider getting an electrical toothbrush for your kids and monitor their oral hygiene habits not only during childhood but also in the adolescent and adult years.\*

#### Acknowledgments:

We thank all the children with SMS and their families for their cooperation with this study and the SMS interdisciplinary research team at NIH, established by a Clinical Center Bench to Bedside award. The work was supported by the intramural programs of NHGRI and NIDCR/NIH, Department of Health and Human Services.



## Tips for Parents

It is very important to maintain the oral health in your children. Teeth play a role in speaking, eating, and appearance. As we found in our study, individuals with SMS seem to have more difficulty, as they get older to maintain a good oral hygiene causing an increased presence of dental caries and gingivitis in comparison to the general population. These are a few dental recommendations that can be helpful for you as SMS parent or caregiver:

Supervise how your children brush their teeth even in their adolescent or adult years. If they have trouble getting all the food in their teeth after brushing or you notice that the gum is red and swollen, you may consider getting an “electrical” toothbrush for them, this will make tooth brushing easier for them.

Make sure your children brush their teeth everyday after each meal or at least twice a day.

Use of dental floss is also important because food between the teeth cannot be removed completely by the toothbrush and this may cause the appearance of caries or gingival inflammation between teeth. This requires fine motor skills so you will probably need to help them with the dental floss at least once a week. There are different products in the market; we recommended those ones that have a plastic holder so it will make your task easier.

Diet is another important factor in dental caries. Avoid sodas, juices with high sugar, snacks such as chocolates, candies, etc. We recommend drinking water, and eating fruits, vegetables and nuts as snacks. Since it is difficult to eliminate carbohydrates completely in our diet, you can eat them with meals but avoid them between meals.

Your children should visit their dentist twice a year for regular maintenance and ask your doctor for preventive treatments such as fluoride gel application.



# The 1st Scandinavian Smith-Magenis Get-Together Has Been Held!

My daughter was diagnosed with SMS in February 2004. My husband and I searched the net and asked the doctors, to find out everything we could about the syndrome. Like many others we didn't find much, and what we found in Danish was almost nothing.

The PRISMS website was our source of knowledge, and we soon found that we knew more about the syndrome than the doctors! But we wanted to know more, and to get in touch with other families with SMS. So I participated in the 2005 PRISMS conference, I was overwhelmed and learned so much. I felt that the rest of the Scandinavian SMS-population (especially those who don't understand English that well) should have access to

more information and I wanted to find the families "out there."

After the 2005 conference I made a web-site about Rebecca and SMS in Danish ([www.smithmagenis.dk](http://www.smithmagenis.dk)). I have constantly been updating the homepage, and last spring I received the 1<sup>st</sup> call from the mother of a 17 year old boy. She had never met anyone else with the syndrome, and she didn't even know how long she could expect her son to live. She was shivering and over excited to finally talk to another SMS mother, and she asked so many questions of which I had answers to most.

Before that, we had contact

with another boy through our doctor, and a girl in Norway through PRISMS. Since last spring I have had 3 parents contact me with newly diagnosed children, and one who suspected SMS. It's such a pleasure for me to be able to help and support new SMS families. My husband and I decided to invite everyone to a get-together.

So that was the 9<sup>th</sup> of June, and we managed to gather 5 families out of the 9 families we know of in Denmark and Norway. I did a speech about the 2007 conference so everybody who wasn't at the conference got an idea of what it was all about.

It was lovely to see all the kids and families, of which 2 had never met another SMS individual before, and I think everyone enjoyed themselves and learned something too. We're definitely going to repeat the event, and I'm definitely coming to the next conference, hopefully with a small handful of Scandinavian families.

Thank you PRISMS for being such a huge support and help to all us SMS families out here!!!

Pernille Fox, Denmark





**Dave Thomen, magician**

The exuberance was notable – not just in the Smith-Magenis children, but also in the 300 attendees at the 5<sup>th</sup> SMS conference.

Thursday evening, all were welcomed with red carpet treatment. The food was excellent and the entertainment inspirational. The Rhythms of Hope Dancers graced the stage with celebration and joy. The company is composed of persons with disabilities and allowed all to join them on stage for wonderful musical expression. This was followed with music by Alex Smith. Alex is an accomplished musician who wrote the lyrics and music to “Building Bridges of Hope”. Several of our SMS youth were gladly welcomed on stage by Alex, who showed remarkable kindness and patience as they “directed” his act and “shared” his microphone, all the while providing musical entertainment to the audience. We are sure his mother, Ann Smith, was extremely proud of him.

Dave Thomen could have been mistaken for the Pied Piper as he made his way through the room performing his magic show. “Mr. Dave”, father of Sarah, brought laughter to all those around him. What a great way to kick off an awesome conference!

A very full Friday began with the presentation of “Rocksoup” by Sally and David Sloop from North Carolina. They shared their journey with us regarding their autistic son, Peter. Encouragement was given to all to use your parental wisdom as well as the professional’s wisdom as you travel the often rocky, twisting roads of parenting the child with special needs. Sally spoke of turning your sorrows and rage into a productive driving force. David was humble as he shared how his telling of *his* story led him to a pathway for healing. Peter, through video presentation, shared the very wise words “Never give up on hope, have faith in yourself and your child.”

Full attention was given to Dr. Ann Smith and Dr. Ellen Magenis as they took us from Identification

to Understanding with an overview of the last 25 years of Smith-Magenis discovery. Dr. Magenis also provided us with a glimpse into the life of the eldest person living with Smith-Magenis Syndrome.

Sleep updates were given by Wally Duncan, PhD, and Helene DeLeersnyder, MD. If you are not aware of the melatonin/betablocker information available, you may want to go to the PRISMS website to review this.

Tim Sweeney, LCSW, presented on SMS and the Stress on Marriage and Family. Tips on parenting special siblings include giving them quality on-on-one time, encouraging honest discussion of feelings, and providing them with a positive role model on how to handle

challenges. He also encouraged us to nurture our marriages in order to provide a healthy environment for our families.

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**“Never give up on hope,  
have faith in yourself and  
your child.”**

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Photographer and founder of Positive Exposure, Rick Guidotti, provided such an open, heart-warming presentation which included many wonderful photos





**Dr. Ann Smith and Jacqueline**

of our SMS children. If we could just tap into his energy and store it up for use after one of our “hard day’s nights” we would be blessed!!

The serious subject of estate planning and special needs trusts was addressed with humor and authority by attorney Robert Bullock. He provided a roadmap for navigating the maze of planning needs. This dove-tailed wonderfully into a later session on special needs finances. We were all reminded of the importance of having a Durable Power of Attorney, an Advanced Medical Directive, and a Will. These are just a start in our planning needs as parents of SMS children and it is invaluable to have the data presented in an understandable manner.

Both Friday and Saturday were filled with many options during concurrent sessions. Professionals and parents presented on SMS “tactics.” There is no way to review them all here. The handouts will be available on a CD later this summer. Look for future information in the upcoming infomail. Please note that handouts cannot convey the ins and outs of a lecture. Plan now to attend the

next conference!

The value of the presentation by Barbara Hass-Givler, MEd, BCBA, and Mary Beall, MEd, parent, cannot be understated. They gave practical advice on behavior management in the school and at home, being refreshingly specific to students with SMS. Talk about teamwork, Mary had a coach (SMS daughter Laura) from the floor, and her husband Randy pushing the laptop buttons (or was that Laura pushing her buttons!). Mary showed true parental multitasking as she presented great information in a wonderful way. Barbara was empathetic and knowledgeable as she gave an understandable breakdown of a complicated process. She made it real, while being a true professional who “gets it.” She has promised to write an article for a future issue of Spectrum regarding this lecture.

A research update was provided by Ann Smith, MA, DSc (Hon), and Sarah Elsea, PhD. There have been advancements in the previous 2 years with improved diagnostics and more in depth clinical studies. Future studies are planned for determining the function of the RAI1 gene in the cell and its role in development and behavior, improving diagnosis with better testing and whole genome studies, and also to evaluate the role of other genes in this deletion. Studies you can easily participate in include a SMS Caregiver Study found at <http://survey.vcu.edu/surveys/TYJBX5> and the upcoming prospective web-based SMS medication database that is accessible through the

PRISMS website. Dr. Smith is also leading a treatment study that will study the effects of bright light therapy versus use of delayed time release melatonin tablet (being made by NIH) on sleep in SMS. The Silent Auction ended the day’s activities on Saturday. It was a great success, and there was an item for every price point and every interest. Serious bidding took place, and some of our attendees have mastered the art of Silent Auction bidding! Their efforts paid off for PRISMS, with a grand total of \$8,763 raised for PRISMS. Thank you to all who organized, donated and participated in the Silent Auction.

On Sunday, we celebrated Mother’s Day, and all the women at our conference were given Mother’s Day gifts, (SMS purse charms). We wanted to thank all these wonderful women, who have made such an imprint on our children’s lives and have been there along the way to help us raise, teach and love these children. We also had a professional harpist come and play during our Mother’s day breakfast, and the children were fascinated and sooo quiet during the harp music. This was a soothing backdrop to what had been a very busy few days.



**Alex and Julia**

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## Conference Review

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The conference concluded with parent presentations from four families who presented on their own individual SMS journeys: Cheryl & Ed Huber, parents of David, age 14 ; Tina and Mark Thomen, parents of Sarah, age 15; Heather and Dan Wilde, parents of Madison, age 10; and Patricia Pearson parent of Deieah, age 14, whose twin brother, Demetrius joined the parent panel. Insights, funny stories, achievements and “battles” won were shared --- a bit like “Chicken Soup” for the SMS Soul. Our stellar group of parents ended the conference on an encouraging note, and challenged us all to strive onward with humor and grace, and celebrate our children.

We cannot end without sharing a true “SMS moment.” There was a young man Deane, SMS, who was the “official” greeter. Early Friday morning he greeted a nicely-dressed gentleman at our breakfast buffet. The questions were fast and furious....”What is your name? Where are you from? Who is your child? Where is your nametag?” He was relentless! The gentleman answered his questions...even admitting that he wasn’t actually with our group (busted!!!). A parent kindly said “Guess you are getting an education on Smith-Magenis Syndrome” and walked off leaving him in the very capable hands of Deane. Priceless!

Thanks go to the PRISMS board and conference committee, for all their hard work in setting up this very successful conference. Start planning now to attend the next conference which will be announced soon.✱



Deane and Randy

## GROWTH PATTERNS in SMS



NIH is working to develop syndrome specific growth curves for SMS. Data gathered to date on 68 children with SMS show the following growth patterns:

- Most infants with SMS are born at term and have normal birth parameters for length, weight and head circumference.
- During the first year of life, infants with SMS show a decline in weight and height from normal birth range to less than or equal to 5 %tile.
- By age 8-9 years, heights are within the normal range (approx 25%tile) and weights are higher for boys than girls.
- Weight gain to levels suggesting obesity (>95%tile) is frequently noted after age 9 years with onset of puberty.

Additional growth data is needed to complete the project, especially for children from age 4 years into their teens and final adult height. By checking with your child’s pediatrician who keeps a record of height/weight for each annual checkup or reviewing your own records, this data can be pulled together. By year’s end, we hope to have sufficient data to develop growth curves from birth to age 3 years and 3 years to adulthood . Parents willing to provide growth measurements on their child with SMS (heights, weights and if available head circumference) are encouraged to download the SMS Growth Curve form from the PRISMS website ([www.prisms.org](http://www.prisms.org)) under the RESEARCH section.

# Speech Therapy Ideas for Parents and Therapists

By Christine Brennan, M.A., CCC-SLP/L

For the past three years, I have been working with a young man who has SMS. Over time I have learned a lot about the speech and communication strengths and difficulties that can occur with this syndrome.

In May, I attended the PRISMS 5<sup>th</sup> International Conference. It was fabulous getting to meet so many individuals with SMS and to connect with the attending professionals, researchers, parents, and families. I got the idea to contribute to the newsletter after sharing ideas about speech and language therapy with some of the parents I met. In this article, I share three ideas that promote language development in children with SMS. I am also including some speech-lingo to help parents when talking with the SLPs who work with their children. So good luck; I hope this information is helpful!



## **THERAPY IDEA 1**

**AGE/LEVEL:** Younger child, communicating with 1 or 2 words at a time.

**LINGO: MLU or Mean Length of Utterance** refers to the average number of words used in a single utterance or spoken sentence. Children without a language delay typically have a:

MLU of 1 at 12 months of age

MLU of 2 at 24 month of age

MLU of 3.5 at 3 years of age

**GOAL:** Increase average number of words used expressively (state modality: i.e. sign, verbal, AAC, PECs) to an MLU equivalent of 3-4 using visual cues as needed. Recommended criteria: determining MLU given a sample of 30-50 expressive “sentences.”

## **SUGGESTION/ACTIVITY:**

Supplement expressive language tasks with the use of picture or photograph cues for each word in a targeted sentence. Unlike speech or sign, pictures are more permanent and can better illustrate sentence building in a way that spoken words or sign can not. Use of pictures can be used to address sentence and question formulation, and the length of utterances produced can be increased little by little. Pictures should be recognizable and should include the printed word even if the child is not yet reading. Providing verbal models in combination with pictures can result in improved pre-reading skills (exposure to left to right progression and text) as well as opportunities to work on sentence building across various levels of language.

**TWO WORD LEVEL:**

noun + verb (dog run)

adjective + noun (pink pig)

verb + adverb (run fast)

**THREE WORD LEVEL:**

noun + verb + adverb

(bunny jumps high)

noun + verb + noun

(cat drinks water)

adjective + noun + verb

(little boy sleeps)

An additional component to increase MLU is to build up a child’s vocabulary of verbs, adjectives, and adverbs, as well as nouns. In English, early vocabulary tends to emphasize nouns. When children have language delays and are using only one word at a time, they tend to use nouns. They may need some extra emphasis on action words as well as descriptor vocabulary so that they have the necessary building blocks for lengthier sentences. Picture cues can also be used to build questions in the same way they are used to cue sentence building.



## **THERAPY IDEA 2**

**AGE/LEVEL:** School age child/adult, communicating at sentence level.

**LINGO: Visualization** refers to the ability to imagine things not present. Visualization combined with strong language allows us to

Continued from page 11....

communicate ideas with others about abstract or removed things in the past, in the future, or in our thoughts.

**GOAL:** Identify features/ characteristics and describe objects/items/events not present using visual cues as needed. Recommended criteria: Independently generate and identify 3-4 features and/or characteristics of a given object/item/event with 85% accuracy.

**SUGGESTION/ACTIVITY:** Twenty questions / Hedbanz™/ Cranium Conga® /Playing games like Twenty Questions which use visual cues for categorization and features improves visualization of items not present while also addressing synthesis of information to draw logical conclusions. Games like Hedbanz™ and Cranium Conga® can be used in therapy sessions or to play with at home as a fun way to work on these skills. It may be necessary to teach a child what possible questions are and what questions are helpful. These games are also helpful in giving the child opportunities to consider and answer yes/



Deane wearing card from Hedbanz™ (a game similar to “twenty questions”).

no questions about an item someone else is guessing about. The ability to synthesize information, generate descriptions and talk about items not present is critical to our ability to successfully communicate with others. Usually when talking with others people reference things not present. Lack of visualization, impaired expressive language, and poor synthesis of information will all result in deficiencies in social discourse.



### **THERAPY IDEA 3**

**AGE/LEVEL:** School age child/adult, communicating at sentence level.

**LINGO: Social-pragmatic language skills** refer to the ability to interpret and use language in social contexts. It includes using sentences, questions, and stories to engage in conversations and play with others. In addition to articulation and language formulation difficulties, children and adults with SMS often have difficulty with social-pragmatic language skills.

**GOAL:** Improve social conversation skills by generating on-topic comments and questions for a given topic with visual cues as needed. Recommended criteria: 4-6 comments/questions per topic.

**SUGGESTION/ACTIVITY:** Practice making basic comments including:

- I like \_\_\_\_\_.
- I don't like \_\_\_\_\_.
- I have \_\_\_\_\_.
- I don't have \_\_\_\_\_.

I've done that.

I've never \_\_\_\_\_.

I've been \_\_\_\_\_.

I've never been \_\_\_\_\_.

I would like to \_\_\_\_\_.

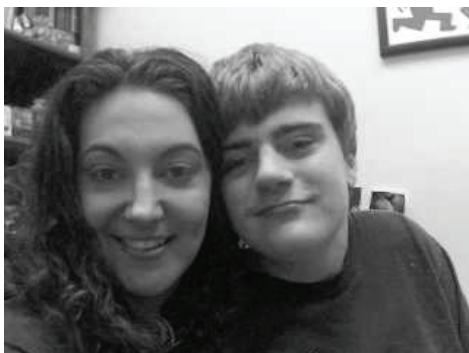
I wouldn't like to \_\_\_\_\_.

It is important to work on just one or two comments at a time. Start the activity by sharing information about an activity or preference that would be used to initiate a conversational exchange. For example, “I went out to lunch today.” Individuals with SMS like to ask questions and they like to get answers to their questions. They may want to ask where you went or who you went with. Remember to work on questions that are on topic. Topic selection can be alternated via turn taking. Possible topics can be made visually available using pictures and/or text. When working on making comments, it is helpful to establish a routine that includes a required comment on the topic when initiated and before a question can be asked and answered. For example, sign “C” near the face as a visual cue to remind an individual to make a comment. When an individual improves at making comments, start increasing the requirement to 2, 3 and even 4 comments per conversational topic. Always remember to work on topic maintenance when generating questions as well. Practice with multiple people including familiar adults and peers.

### **A Final Note**

The key to improving any individual's language skills includes: (1) building from established

strengths, (2) providing the tools needed to make gains, and (3) making activities motivating and fun. No one communicates about things they are uninterested in and no one learns when they are not motivated. Be creative and modify suggested activities to fit your child's needs and interests. Do not stop when your child masters the targeted goal; there is always a next step for everyone.\*



*Christine and Deane*

Christine Brennan, M.A. CCC-SLP/L is a private speech-language pathologist in Skokie, IL. Christine has over 17 years experience working with children and adults with developmental disabilities. She frequently lectures at Northern Illinois University as well as at national and state speech pathology conventions. Questions for Christine can be sent to [christine@brennanandburns.com](mailto:christine@brennanandburns.com).

## PRISMS SMS Discussion Forum



The new PRISMS SMS Discussion Forum or Bulletin Board is online! This great new resource is designed to build a "knowledge base" for SMS - created by parents, professionals and people affected with SMS - to help and support each other. Because of the structure, good posts with valuable information should be easy to find and won't get automatically archived because of age. It's easy to search the various forums - either by category and forum and topic, or just by using the search function to find entries.

Other features:

- \* Information is well-organized to help newly diagnosed parents and persons find what they need
  - \* It's easy to begin new discussion topics
  - \* It's easy to post daily updates
  - \* Registered users can easily access all posts which have been made since their last visit
  - \* You can edit your own postings - if you make a typo or need to explain something
  - \* If you post a question, you can click "watch this topic" and you get email notification when someone responds to you
  - \* Private messaging (PM) allows you to contact others privately
  - \* Anyone can view most areas of the forums; to post you must register
  - \* Personal information (real name, location, personal website) may be made public or kept private - it's up to you
- We hope you will go to the PRISMS website – [www.prisms.org](http://www.prisms.org) – and check out the link to this important new resource. If you have questions, please email [prismsbb@prisms.org](mailto:prismsbb@prisms.org)

### Monthly Emails



We have been sending out monthly informational emails to keep everyone up to date on fundraising, reminders, conference information, etc....

Please add [prismsnews@prisms.org](mailto:prismsnews@prisms.org) to your address book and spam exception list to insure you keep receiving your infomails.

If you have not received these emails and would like to, send your name and email address to [info@prisms.org](mailto:info@prisms.org) or [prismsnews@prisms.org](mailto:prismsnews@prisms.org)

# "Connected

Our son Garrett was diagnosed with SMS at the age of 18 months after failing to reach several milestones. At the time, my husband, Charlie and I could not imagine anything worse than that devastating diagnosis. Last summer, we learned that was not true. Ironically, **the only thing worse than knowing your son has SMS is not knowing your son has SMS.**

Garrett brought home a paper from school that said the county school board was holding a meeting for parents and families affected by autism. I went and met several parents who were dealing with the same issues: explosive behavior, obsession/compulsion and sensory issues. Robyn Foster was there along with her parents, but we only spoke in passing. That first meeting led into a parent support group that meets once a month to vent our frustrations with insurance companies, school boards and overall ignorance. When a parent suggested making a video of what actually goes on in our homes, Charlie brought the PRISMS "Smith-Magenis Syndrome Journey" DVD we picked up at the Cincinnati conference.

Although Robyn was not at that particular meeting, her mother was and she started to ask about Garrett. We went through the list: we have not slept in seven years, Garrett squeezes his hands or hugs himself when excited, and he flaps his arms and slaps his head when upset. She said that Garrett's picture looked a lot like her twelve year old grandson, Zan. We gave her the video and a PRISMS brochure to take to

Robyn and her husband, Harold.

The following week, we went with Garrett to the Dream Soccer Tournament at our local park. I heard Zan as soon as we stepped out of our van. I looked over at Charlie and he said "Don't scare them off." It was hard to contain my excitement when I finally saw Zan on the field and I knew without a doubt he had SMS. I did not want to seem pushy and full of unsolicited medical advice to someone I barely knew. Luckily though, I felt an instant connection to Robyn. She had already gone to the PRISMS website and she knew as much about SMS as we did. So much for my great advice!

After the Fosters received Zan's diagnosis, people started telling Charlie and I what a "help" we could be to their family. The truth is, Robyn and Harold are six years farther down the SMS Road and they have been an invaluable help to us! They know all the tricks from eating in a restaurant to potty training and they even go on vacation. Robyn and Harold have accomplished so much without even knowing the source of their son's disability; so we know we can believe them when they say we will survive these years, too.\*

With Wonderful Intuition-  
**Tina McGrevy**

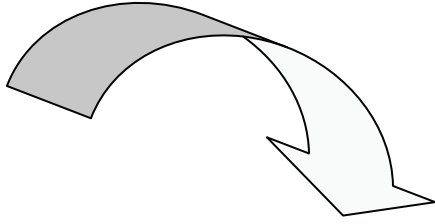


Zan and Garrett

With Awesome Relief-  
**Robyn Foster**

Tina has a wonderful way of saying "**the only thing worse than knowing your son has SMS is not knowing your son has SMS**". Those words speak of years of pain and anguish for our family. We noticed early on all of the typical traits of SMS babies, with my son's lack of sleep, feeding problems, choking issues, not reaching milestones, hypotonia, all of those awful things

# Stories"



that make you feel like an inadequate mother...."Maybe I'm just lazy and I am the one who sleeps too much."

Feeding him was so terrifying to me. He never ate more than an ounce at a time. I could raise wild and abandoned baby animals but this little human was not going to make it by my hands. I voiced all my concerns and fears to 2 pediatricians who never gave him a second look. I was told I was an "overprotective new mom."

I knew when I took him to the Health Department for his immunizations, someone finally saw something. I was told to go home and wait for him to gain some weight because he was too skinny to get an injection in his little legs.

I switched pediatricians, only to find the same mundane attitude of "Oh, he will catch up." I was getting a little more of my self esteem as a mother whittled away each time I saw him go no further in milestones.

We decided to enroll him in speech at around 18 months old and that was just the beginning of therapy. Speech, PT, OT, psychologists, neuro-psychologists, and geneticists. Yes, that's right, even a geneticist who didn't pick

up on SMS. We had accepted a diagnosis of Autism Spectrum Disorder from our 3rd pediatrician, who took an interest in my son. He found him very interesting because he did not have the classic autistic traits. We were always given undivided attention in the office but again, even this most caring and wonderful doctor did not know SMS.

This physician's office held an autism support group as Tina had mentioned. We did see each other once but never spoke. The night my mother spoke to Tina, she called me and was very anxious for me to speak with her. I agreed with my Mom that this sounded very promising. I knew my Mom would not create false hopes, so I was excited as well. My parents have been through this for years as my brother has many disabilities and presents as an undiagnosed high functioning autistic individual.

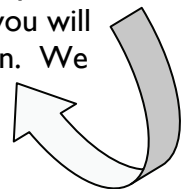
I found as much information on SMS as I could. What really made my hair stand on end was their facial characteristics and the sleep pattern explanation. I KNEW I had an SMS child. Tina referred us to yet another geneticist in Columbus, Ohio. We again go to the genetics lab and were told that they also thought we were right.

Within a week we got our call--- Positively an SMS child!!! I cried "Thank you, thank you, so much!!!" The lady on the phone said "I guess I'm stunned that you're happy. I don't get many of these types of responses." I told her that it was closure for me and our struggle to be heard was

over. I know my son has something REAL, Smith-Magenis Syndrome. It is real. I'm not a bad educator to my child. I never caused this by inhaling fumes, it's not Thimerisol. I did everything I should have!!!! My son has SMS. I said it over and over.

Needless to say I was on the phone first to Mom and Dad, then to my husband, and then to Tina. My mind sharpened as I marveled at who would hear "I told you so!." The list began with the elementary school psychologist, then to the principal and so on!!! I was back on top of my game. "Take me on now people" is all I could think of!! I am now the expert and I always was. Thirteen years of fighting, done.

I would never have guessed that nine months later I would join other families, just like us, in Washington DC. I apologize to anyone who thought I was staring or taking too many pictures of your wonderful children but I was absolutely blown away. It was very surreal for my husband and I to see so many mannerisms and behaviors and voices just like my son Zan. I was so at peace with all of you. You have all lived my life and were still laughing, like we do. I consider you our extended family and believe that the conference was one of the best things that ever happened in our life. I can truly say that I would do anything for any of you if it were in my power, especially to you-Tina and Charlie McGrevy- you will never be alone again. We love you. \*





Parents and Researchers  
Interested in  
**Smith-Magenis Syndrome**

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Sterling, VA 20164

## 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



**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.*

## THANKS!

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