prisms
Parents and Researchers Interested in Smith-Magenis Syndrome

spectrum

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Dr. R. Ellen Magenis A Pioneer for SMS

R. (Ruth) Ellen Magenis, MD, FACMG1
Tribute by Ann C.M. Smith, MA, DSc (Hon), CGC
Senior Genetic Counselor, OCD/NHGRI/NIH

On February 4, 2014, we lost a true pioneer in the fields of clinical cytogenetics and medical genetics with the death of American College of Medical Genetics Founding Fellow R. (Ruth) Ellen Magenis, MD. Dr. Magenis, age 88, died at home with her family in Portland, after a long illness.

Born September 24, 1925 in Gary, IN, Ellen received her BA in Zoology from Indiana University in 1946 and earned her MD from Indiana University Medical School in 1952. She then dedicated time to raising seven children and completed her residency in Pediatrics in 1968 at the University of Oregon Medical School, now Oregon Health & Science University (OHSU). Working with Dr. Frederick Hecht at OHSU, Dr. Magenis completed a fellowship in Medical Genetics in 1971. In 1970, over two decades before the Human Genome Project, Ellen had mapped the haptoglobin gene to a heritable 16q fragile site using standard techniques of the day; this was only the second gene mapped to a specific autosome. Ellen next helped launch the OHSU Genetics Clinic, and was named Director of the Cytogenetics Laboratory in 1977. Beginning as a faculty Instructor at OHSU in 1965, she rose in rank to Professor in 1980, followed by Professor Emerita of Molecular and Medical Genetics and Pediatrics in 2008. She was board certified in Pediatrics (1971) and in Clinical Genetics and Clinical Cytogenetics (1982).

During a productive career spanning 40 years, Dr. Magenis was a dedicated clinician-scientist, whose tireless energy, clinical acumen, and passion for cytogenetics earned her worldwide respect. She served on many editorial boards and authored or co-authored almost 300 publications. A dedicated teacher and mentor, Dr. Magenis was known for her egalitarian management style; she treated techs and trainees as colleagues, encouraging all to stretch their abilities. Not afraid to swim against the current, she questioned, thrived on discourse, and embraced the future by taking advantage of new technological breakthroughs to remain on the cutting edge of cytogenetic research

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and clinical testing. Her research interests focused on gene mapping, numerical and structural aberrations of chromosomes—understanding their morphology, function and clinical correlates, roles in malignancy, relationships to sex determination—and the clinical delineation and molecular origins of microdeletion syndromes.

Early in my genetic counseling career I was fortunate to meet Ellen at the 1982 ASHG annual meeting; after she heard my presentation describing two children with interstitial deletion 17p11.2, Ellen excitedly felt she had a third case with a similar deletion. So began our long-term collaboration and a special friendship that spanned over three decades. Together in 1986 we described nine patients with a new microdeletion syndrome that now bears both our names, forever linking us with the legacy and story of Smith-Magenis Syndrome (SMS). Dr. Magenis's devotion as an advocate for patients and their families led to her active roles on parent advocacy boards for families impacted by Angelman, Prader-Willi, and Smith-Magenis syndromes. For Parents and Researchers Interested in SMS (PRISMS) and families of children with SMS, Ellen was a STAR—the kids knew her and she knew each of them and their families. Ellen's true essence is exemplified by advice she shared with her former graduate student, Susan Olson, PhD, FACMG, who now directs the OHSU Clinical Cytogenetics Laboratory, "Remember the patients for whom you are trying to make a difference. Value them, respect them, because they are part of you and your work." Dr. Magenis will be greatly missed by her family, friends, and colleagues in the genetics community, where her star will forever burn brightly.

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A Message from PRISMS President *Randy Beall*

As you may have heard, I'm stepping down as PRISMS president after 10 years of service. However, I will continue to serve on the board.

Laura, our SMS daughter, was 16 when I first joined the board. Sometimes it was hard to be her parent and also the PRISMS president. There were times when I was on the phone trying to advise a parent, while my own child was melting down in the next room. At conferences, I often couldn't speak into a microphone until she got to do that first. On those days I asked myself (and my wife, Mary, asked me): "What was I thinking taking this on?"

And then I thought about the ones who came before me. Scott and Maggie Miller, Ann Smith, Brenda Finucane and Barbara Haas-Givler.They started with nothing except a knowledge that there were families who were suffering and a desire to help.They did the legal work, held conferences, researched and wrote articles, and started the newsletter and the website (remember dial-up internet connections?).They were on the phone with parents, doctors, and other researchers, sending them information by making multiple trips to the post office each week. They had children too, and yet they managed to do all of the work to start PRISMS. We all owe so much to our founders, who still are working hard for us more than 20 years later. So I knew I needed to keep working, too.

PRISMS has improved our lives in so many ways. Before we joined PRISMS, I remember wishing we had other people to talk to. PRISMS connected us. We now have PRISMS friends from all over the world, and some in Texas that we get to see quite often. I remember wishing there was something written that I could give to teachers. PRISMS had articles, and now we have a book, published by PRISMS. There was so much I needed to know about SMS sleep and behaviors. PRISMS sent us articles and invited us to conferences. And now, when I need information, a few clicks on the PRISMS website give it to me, and I can easily share it with others. I can connect with the PRISMS facebook page, and get tweets about the latest news. We've come a long way!

I want to thank all of you who have volunteered: PRISMS board members (you're the best!), conference planners, fundraisers,

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PRISMS Smith-Magenis

researchers, conference speakers, and those who have shared your experiences with other families. Keep going; it's worth it! Mary and I won't stop either, because we *believe* in the work of PRISMS. And besides, you don't say goodbye to family.

It may be hard for me to retire from the limelight. My friend, Rachel Hetherington, might stop calling me "The Big Cheese", and my friend, Deane O'Connor, might stop collecting my boarding passes. I will be back to just being "Laura's dad", which I suppose is OK with me, since that is how this whole thing started.

The future of PRISMS looks very promising and exciting! Our new PRISMS president, John Mayer, has a lot of energy and passion plus a wealth of non-profit experience. And, as you may have heard, PRISMS will be hiring a fulltime executive director in 2015 to help us provide even more education, research and awareness for our families. So here's to a bright future for PRISMS, all of you, and your children! Together we are making a difference!

Warm regards,
Randy Beall, PRISMS ex-president, Laura's dad

Transitioning to a Supportive Living Environment

By Kevin O'Connor

At some point in an SMS young adult's experience, it may be useful to explore alternative living arrangements where the SMS individual lives outside of the parents' home. Depending on the state, there are a number of group home options that are integrated into local communities that can provide the support, supervision, and structure that would facilitate such a transition.

Deane O'Connor, 23, is an SMS male currently living in his parents' home in Evanston, Il., a Chicago suburb. Deane is preparing to move to a supportive living environment—most probably a group home of four to six residents in a local community setting. This article details steps which can ease this process on Deane and his family, and suggests strategies to ensure a successful transition.

1. **When do you know it's time to start this transition?**
   a. When you realize that you really can't help your SMS son or daughter progress any more in their current environment.
   b. When your SMS young adult needs more/different structure and support.
   c. When your SMS young adult would benefit from a set of full-time role models, peers, and supports
   d. When it becomes time to accelerate independent life skills and social skills.
   e. When you have difficulty filling in your SMS young adult's day with meaningful activities.
   f. When your SMS young adult tells you (with words or actions) that he or she is ready for this new stage in life.

2. **Why go through the effort of transitioning?**
   a. Because you're ready to downsize to a smaller house and can no longer accommodate your SMS young adult on a full-time basis.
   b. Because your SMS young adult is bored with the current arrangement and needs stimulation and new relationships.
   c. Because your SMS young adult is "ready for the next step", much the same way a typical young adult is ready to leave for college or move to an apartment.
   d. Because the retirement plan you've envisioned includes your SMS young adult on a visiting basis, as opposed to a full-time responsibility.
   e. Because you realize that your SMS young adult deserves a fuller life experience with friends, peers, and housemates than you can offer in your home.
   f. Because you're just plain tired.

3. **What is an appropriate fit (what's important to Deane and his parents)?**
   a. A room of his own.
   b. Housemates that he likes and can get along with.

c. Not too large of a group (seven individuals max, three or four ideally).
d. Geographically close to parents.
e. Access to public transportation.
f. Adequate and appropriate support for social, life skills, and work/enrichment.
g. Healthy and aligned attitudes and views about consensual sexual activities.

4. **Other factors important to a successful transition.**
   a. Get into the state system earlier than later—not so much for the financial support, but more for the visibility.
   b. Select a good life skills coach. A solid, reliable LSC can have a tremendously positive effect.
   c. Get comfortable with your SMS young adult having more independence (this can be hard).
   d. Help to migrate toward more independent activities and life skills with cooking, laundry, cleaning, shopping, eating out, money management, and public transportation.
   e. Strive to "fill up the day" with meaningful activities for enrichment.
   f. Understand and accept that you'll always be involved in your SMS adult's life. That does not end when they move out.

Volunteer Opportunities

Journal assistant editor sought! PRISMS is seeking a volunteer with organizational skills, computer skills, and an eye for design. We are specifically seeking an individual with experience in publishing software. We need someone to get the data from the Editor of Spectrum and place it into the right format for electronic distribution. This position does require computer experience and good communication skills. There is a well-developed newsletter committee to support the editor and assistant editor. For more information on how you can help PRISMS please contact editor@prisms.org.

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

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

Caregiving Responsibilities/Sibling Interview Study

Current and future caregiving responsibilities among adolescent and young adult siblings of individuals with Smith-Magenis Syndrome: A report from the Sibling Interview Study

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As the PRISMS community is well-aware, Smith-Magenis syndrome (SMS) is a rare chromosomal disorder characterized by craniofacial anomalies, intellectual disability, global developmental delays, chronic sleep disturbances, neuropathies, and extreme behavioral/emotional dysregulation. Given the numerous SMS complexities and the fact that challenges exist throughout the lifespan, siblings of individuals with SMS may worry about current and future sibling caregiving responsibilities. To better explore perceptions of these responsibilities, 20 adolescent and young adult siblings (average age = 18.0 years, range = 13-25 years, 60% male, 40% older than the sibling with SMS) participated in the Sibling Interview Study at the recent PRISMS International Conference that was held in St. Louis from July 31st – August 3rd, 2014. Siblings participated in an audiotaped interview and completed a questionnaire on sibling responsibilities.

Results showed that although most siblings denied worrying about *future* financial concerns and sleep disruptions, the majority reported concerns that *future* sibling responsibilities will:

- Be physically demanding, inconvenient, overwhelming, and confining,
- Require changes in family dynamics, personal plans, and work schedules, and
- Include managing difficult sibling behaviors.

Female siblings reported somewhat greater worries about *future* caregiving responsibilities than males. There were no differences in *current* or *future* sibling responsibility concerns based on whether or not the participant was older or younger than their sibling with SMS.

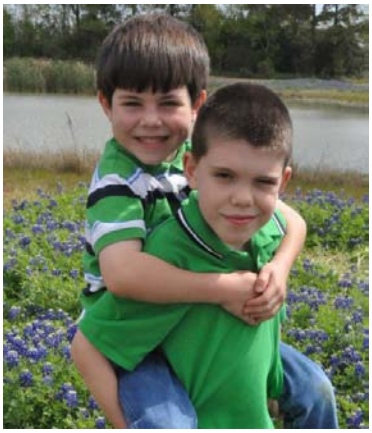
When asked to discuss *current* responsibilities, most discussed babysitting and/or helping care for the sibling with SMS at home.

- Examples:
  - "I'm the designated babysitter."
  - "[I] make sure she's fed, in bed by a certain time, and basically just that she's happy."

Several siblings discussed the need for constant supervision, as explained in the following statement: "I hang out with [my brother] at home when my parents are working because someone needs to be home with him *at all times*." It was not uncommon for siblings to indicate that these are normative activities that they would likely be doing regardless of whether or not they had a sibling with SMS.

- Examples:
  - "[It's] just like normal helping."
  - "It's just like having a younger brother."

Siblings expressed appreciation to parents as well. As one sibling stated, “Besides babysitting and the occasional running errands and taking her places when my parents are busy, I’ve never really had a lot of responsibility, and I’ve always appreciated that.” While most siblings discussed practical sibling responsibilities (e.g., babysitting, preparing meals, providing rides), one sibling expressed that his primary role is protective in nature; “I feel like I have to protect her because some people don’t understand how she is. They may see her acting the way she is, [and] they may [not understand]. So I help her [and] protect her.”



Siblings were also asked to discuss what *future* responsibilities they believe they will have. Most siblings reported a belief that they will play a primary caregiving role. One participant reported, “I’m going to have to take care of her whether she’s in a group home and I live five minutes away, or we live together.” Most siblings indicated that although they are accepting of this role, they did not know how they will prepare themselves for becoming primary caregivers.

- Examples:
    - “I’m going to have to learn all that stuff my parents do; I’m going to have to do that at some point, so I have to prepare myself for it.”
    - “I do believe that eventually I will be taking care of my sister after my parents are gone…But I don’t know how well I will take to it. I’ve never had the responsibility of fully taking care of her. So I don’t know what medications she has. I don’t know who all of her doctors are. I don’t know a lot of things.”

A few siblings suggested that caring for their sibling with SMS may be a burden; “You watch out for your blood because family’s family…it’ll be a very frustrating, guilt-ridden thing, and ultimately why I’ll have to take care of my brother…I’ll be my brother’s keeper. And, that’s how it feels really.”

Some siblings reported that although they will not be living with their sibling with SMS in the future, they plan to be involved and helpful in their lives in a number of ways.

- Examples:
    - “If they [my parents] want to go do something, I’ll come over and help watch [my sister with SMS].”
    - “I’m going to get [my brother with SMS] and come to my place for birthdays and take him out for special days. I also want to take care of his money.”

A few siblings reported that they have discussed future caregiving needs in detail with their families. One sibling reported, “My parents have gone out of their way to make it as easy for me as possible and never make me feel like ‘Hey, you’re going to have to do this when you get older. This is coming.’ Financially my parents have been working with people to save money and invest money that will go to taking care of her so I don’t have the financial burden, which is amazing.” Another reported feeling included in the decision-making process; “*We* were thinking assisted living. So I would go visit him once a week, or how often I go just depends [on] what happens.”

While some siblings reported having discussed future caregiving plans with their parents, others stated that they were unaware of future plans for their sibling with SMS.

- Examples:
    - “I don’t really know what my parents are doing when she gets older, but I don’t think I’ll have any big responsibilities.”
    - “I have no idea [what my responsibilities will be]. It just depends on what happens with him.”

Other siblings may be underestimating the complexities involved in caring for someone with SMS. As one sibling stated, “I think once my parents are gone I might have to take care of him, but I don’t think it’s going to be too hard.”

Many of the siblings in our study discussed a desire to talk to their parents in greater detail about future plans for their sibling with SMS. One sibling reported, “I’ve always asked my

parents, 'What are you going to do when she can't go to school anymore?' [And they've said], 'You don't need to worry about that.' And then they'll always tell me, 'It's not your responsibility. Don't worry about it. Be a teenager. Live your life. We'll worry about it. It's our job. She's our kid.' But I still worry about it. I mean, I don't want my mom to have to take care of her [for] her entire life...So they've been looking into group homes and stuff like that but knowing [my sister], I don't know if she could handle it because she's like hip to hip with my mom all the time. She always has to be with her. But, they always tell me not to worry about it, so I try not to, but I still do because I care about my family."

Finally, siblings were asked about any specific *caregiving role concerns* they may have and what *resources* they may like to have available in supporting and navigating these roles. Siblings discussed how important it is to them to have their sibling with SMS continue to be a part of their lives.

- Examples:
  - "I love her and I want her to be in my life and be involved in my kids life, if I have any, and I'd like her to be there."
  - "I worry about how [my brother] will *feel* when he is in a residential home."

Siblings frequently discussed concerns about how future significant others will play a role as well. One sibling stated, "So he'll [my husband] have to be accepting of [my brother] as well. And that might be a little tricky because if you don't grow up with someone [with SMS] or knowing someone with special needs. It can be a little awkward, so he would have to get used to [my brother], which could be hard. Yeah, so [my brother] living with us could be a little tricky."

Several siblings reported that they plan to rely on family to help assist them in caring for their sibling with SMS. For example, one sibling reported, "I'll try to get my parents to help and maybe my younger sister. She can help too." Another reported, "Family will definitely be a big support; talking to my siblings about it." Others acknowledged PRISMS, sibling groups, and conferences in supporting their families needs (e.g., "PRISMS will be helpful for me.") As one participant stated, "I definitely need to stay in the loop on SMS and trends and what I can do for [my sister] so something like this conference is important."

Overall, results suggested that siblings want to participate in their SMS siblings' lives as they age. However, siblings have concerns about the challenges that may exist in acting in both primary and secondary caregiver roles. It appears that siblings desire to be a part of the discussions that are occurring about their siblings' future care needs, even when these conversations may include sensitive information that can be difficult to discuss. Families may find it helpful to initiate conversations with their adolescent and young adult typically developing children. It is recommended that parents/current primary caregivers work with siblings to put together a binder or portfolio that contains significant information about the sibling with SMS (i.e., educational, medical, social, behavioral needs and supports) and provides space to document plans to support future care needs. The following topics/questions may be useful to consider when facilitating ongoing discussions with typically developing siblings:

- What are the options for where my sibling with SMS will live as an adult? Which of the potential options would be the best fit for our family?
- Will my sibling with SMS have a guardian, power of attorney, representative payee, health care proxy, or other supports? What do these terms mean? Who will those people be? Will my sibling with SMS have advanced health care directives in place?
- How will finances be handled?
- Who will be my sibling's adult health care providers and therapists? What health care needs and medications do we need to discuss now? What medical challenges may we need to think about in the future?
- What supports are available for helping my sibling with SMS have a job or volunteer?
- What supports are available for supporting siblings in caregiving positions? What sort of respite care or adult day services are available where we live?

Molecular Function of RAI1

By Liqun Luo
Ph.D., Stanford University

The genetic causes of Smith-Magenis Syndrome are now well understood. Most individuals with SMS are missing a segment on one copy of chromosome 17 that includes many genes. Individuals with mutations in a single gene in that region, called Retinoic Acid Induced 1 (RAI1), present with symptoms similar to those of individuals with the chromosome 17 deletions, suggesting that loss of function of one copy of this single gene is responsible for most of the symptoms of SMS. However, the mechanisms through which this genetic abnormality leads to the diverse features of SMS are poorly understood.

The precise function of the protein encoded by the RAI1gene is not known, but lab work by such researchers as Jim Lupski, Sarah Elsea, Katherina Walz and their colleagues suggests that it functions as a regulator of the expression of other genes. As a result, RAI1 could be involved in regulating a variety of different cellular functions.

The goal of our work is to determine the molecular function of RAI1, how its loss affects cellular development or function, how the resulting cellular abnormalities influence the development and function of circuits of neurons in the brain, and how this neural circuit dysfunction leads to the symptoms of SMS.

Understanding this pathogenic mechanism at multiple levels—molecular, cellular, neural circuit, and behavioral—is important for the rational design of therapeutics. If we can understand how loss of RAI1 leads to SMS, it may be possible to design interventions that could alleviate some symptoms of SMS and improve the quality of life of SMS patients.

To study the function of RAI1, we use mice as a model system. The biological processes that occur in humans and that may be disrupted in SMS are likely also to occur in mice—and mice have their own version of RAI1 that is similar to human RAI1. Nearly a decade ago, Jim Lupski's lab generated mice that were missing one copy of RAI1 and found that these mice had many characteristics that resembled SMS in humans: for example, they became overweight, and had distinct craniofacial features.

To expand on this work, we produced genetically engineered mice that allowed us to inactivate RAI1 in restricted populations of cells in the brain. By enabling us to identify where in the brain RAI1 is important for normal behavior, these studies provide a starting point for identifying the mechanism through which loss of RAI1 produces SMS-like features in mice.

Although this work is ongoing, we have made some interesting observations:

- We found that RAI1 was present in all, or nearly all, brain cells—including neurons, the cells that perform most information processing, and astrocytes, cells that support and regulate diverse aspects of neuronal function. However, we observed particularly high levels of RAI1 in some brain regions; these include the striatum, a structure import for movement, motivated behavior, and action selection, and the hippocampus, a structure important for learning.

- Although humans with SMS are missing only one of two copies of RAI1, to understand RAI1's function in the brain, we've also examined mice that are missing two copies of RAI1. Most mice that are missing both copies of RAI1 throughout the entire body die before they are born. We found that when both copies of RAI1 are removed selectively in the brain, mice are born alive but grow more slowly in early development, eventually become obese, have motor function deficits, have impaired learning and memory, and die in early- to mid-adulthood.

- Mice that were missing one copy of RAI1 throughout the brain became obese and had motor function deficits.

- The learning and memory dysfunction that we observed when both copies of RAI1 were removed throughout the brain were largely recapitulated when we removed both copies of RAI1 only from neurons whose function is to inhibit the activity of other neurons. This result suggests that RAI1 function in these inhibitory neurons is required for normal cognitive function.

- Removing RAI1 from excitatory neurons in the forebrain or from astrocytes did not produce any behavioral phenotypes that could be detected by our assays.

- When we deleted both copies of RAI1 from a sparse population of cells throughout the brain while simultaneously labeling these cells to examine their morphologies, we did not observe any obvious gross abnormalities. However, we found that when RAI1 is lost from a special type of astrocyte called Bergmann glia, those cells are present at lower numbers. This result suggests that RAI1 must be important for the development or survival of this cell type.

Considerable additional effort is necessary to follow up on these initial results. Future work will be aimed at identifying more narrowly defined populations of cells in which RAI1 is important for behavior, further characterizing the nature of cellular dysfunction resulting from loss of RAI1, and identifying genes that are regulated by RAI1 and that may account for some of the defects caused by loss of RAI1.

As neuroscientists, we are interested primarily in RAI1's role in the brain, but a similar approach could also be used to understand RAI1's function in other organ systems. We will make all our reagents, including genetically engineered mouse lines, available to the broader research community to facilitate research on diverse aspects of SMS.

A major challenge in the future will be translating the findings emerging from basic research programs such as ours into clinically beneficial therapeutics. Although such work is challenging, and the benefits likely won't be realized for decades, we believe that support of basic research will eventually translate into therapies that will improve the quality of life of individuals with SMS.

This work was supported by the Simons Foundation, the Howard Hughes Medical Institute, and the National Institutes of Health, and was conducted mainly by a talented Ph.D. student, Casey Guenthner. The behavioral analyses were performed as collaboration between our lab and the Stanford Behavioral and Functional Neuroscience Laboratory led by Mehrdad Shamloo.

PRISMS SuperKid

Austin is a super kid! My husband, Jeremy, and I could give you a million reasons why, but one reason is her uncanny ability to get what she wants despite obstacles (or her parents' reservations at times)!

We are huge Spurs fans — Austin especially loves Tim Duncan — and recently Jeremy took Austin to a Spurs game. At dinner before the game, Austin told us she was going to get Tim Duncan's autograph. Of course, we told her how that was not a possibility. Tim would be playing a game and unable to give her an autograph. Despite our rational explanation, she kept insisting. We eventually dropped the matter because no amount of sensible reasoning was adjusting her view.

Midway through the first quarter of the game, the Spurs' mascot, Coyote, was doing his antics a couple of rows below where we were sitting — 16 rows up at the corner near the Spurs bench. Austin began screaming to get the Coyote's attention. After several attempts and with help from a couple other fans, she caught the Coyote's eye. As he climbed over a couple of rows of seats, she came down the steps to meet him and eagerly gave the furry critter a typical SMS hug.

Austin pointed to Tim Duncan on the bench and told the Coyote she wanted to get Tim's attention so she could wave at him. Austin began to call Tim's name, but he did not turn around. The Coyote, with his excellent miming ability to communicate, somehow let all the fans in that end of the AT&T Center know to shout "Tim Duncan" on the count of three. They shouted, but Tim still did not turn around.

The Coyote went into action. Seeing that the first quarter was about to end, he swooped Austin up in his arms and carried her down to the floor. When the horn sounded to end play and the Spurs players stood to huddle up during the timeout, he approached Tim as he stood in front of the bench. Duncan, grinning from ear to ear, reached out and accepted a hug from our little girl. Austin got more than an autograph and Tim got something pretty special too — an SMS hug!

Submitted by Sylvia and Jeremy Farber

EASY SUPPORT FOR PRISMS

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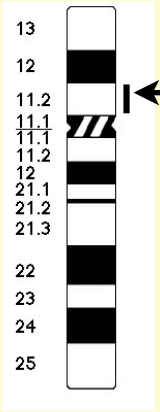
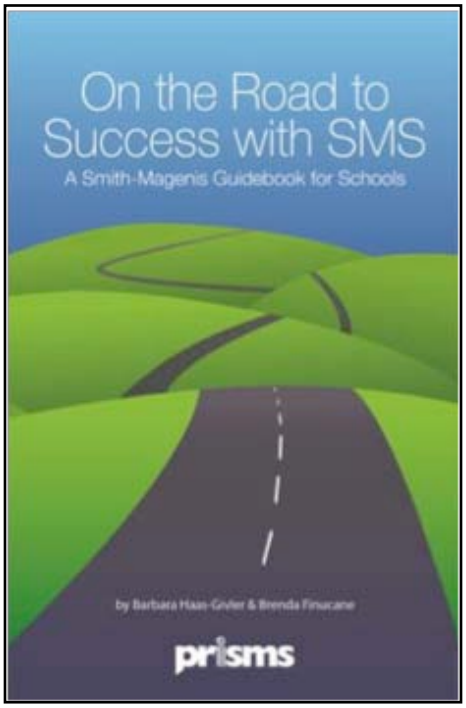
What is Smith-Magenis Syndrome?



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 1980s by Ann C.M. Smith, M.A., a genetic counselor, and Ellen Magenis, M.D., a physician and cytogeneticist. Although the exact incidence is not known, it is estimated that SMS occurs in 1 out of 15,000 births. SMS is under-diagnosed, but as awareness of it increases, the number of people identified grows every year.



On the Road to Success with SMS - A Smith-Magenis Guidebook for Schools

In this book, Barbara Haas-Givler and Brenda Finucane share practical tips, strategies, and accumulated wisdom from their many years “on the road with SMS.” The educational roadmap for students with Smith-Magenis Syndrome (SMS) is full of twists, turns, and detours, with occasional back-tracking as well as long stretches of wide open highway. The journey is not an easy one – these students can pose intense behavioral challenges that are not seen in most other children with special developmental needs. Along the way, both parents and teachers also learn to appreciate the uniquely positive aspects of raising and educating a child with SMS.

Available at:

http://www.prisms.org/us/sms-awareness-tools/sms-book-order-form

Study of Dental Pulp in SMS Individuals

Dr. Lawrence T. Reiter at the University of Tennessee Health Science Center in Memphis, TN is conducting a research study to determine if neurons can be grown from the dental pulp of individuals with various neurogenetic syndromes including Smith-Magenis Syndrome. For more information on how to participate, please contact Dr. Reiter directly by e-mail: lreiter@uthsc.edu.

New Clinical Study by Vanda Pharmaceuticals

A new clinical study is investigating circadian rhythms and sleep disturbances in people with Smith-Magenis Syndrome to guide the development of a possible treatment.

For more information contact SMStrials@vandapharma.com or call toll-free: 844-366-2424.

SMS Immunologic Diseases Survey

Smith-Magenis Syndrome patients experience a significant number of typical and atypical infections. Dr. Neil Romberg, Assistant Professor of Pediatrics at Yale University, is studying the immune system of SMS patients to determine the biologic explanation for this phenomenon.

If you were unable to complete one of Dr. Romberg's surveys at the recent PRISMS conference in St. Louis Dr. Romberg hopes that you will consider doing so now. The survey can be downloaded by families as a PDF and should only take a few minutes to be completed by hand. Completed surveys can be returned to Dr. Romberg by email, fax or post. All survey responses will be kept confidential. Go to www.prisms.org to download the PDF.

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