



Parents and Researchers
Interested in
Smith-Magenis Syndrome

Spectrum

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Smith-Magenis syndrome (SMS) is a chromosomal disorder characterized by a specific pattern of physical, behavioral and developmental features. It is caused by a missing piece of genetic material from chromosome 17, referred to as deletion 17p11.2. The first group of children with SMS was described in the 1980's by Ann CM Smith, MA, a genetic counselor, and Ellen Magenis, MD, a physician and cytogeneticist. Although the exact incidence is not known, it is estimated that SMS occurs in 1 out of 25,000 births. SMS is underdiagnosed, but as awareness of it increases, the number of people identified grows every year.

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Questions?

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Looking for the right Genes?

Dr. Sarah Elsea's laboratory at Michigan State University is focused on identifying and characterizing the genes that fall within the chromosome 17p11.2 region. Dr. Elsea and her colleagues are most interested in the genes that are deleted in all persons with SMS, so their research is focused on the "critical interval" of chromosome deletion. The critical interval is the smallest region of the chromosome that is deleted but still gives the most complete SMS phenotype.

Thus far, 27 genes have been shown to lie within the SMS critical interval. The genes that contribute to the specific features of the syndrome have not been identified. One gene of interest is called *SHMT1*. This gene is important for folate metabolism and may contribute to the higher incidence of minor neural tube defects and other midline defects (heart defects, cleft lip and/or palate and others) observed in those

with SMS. Studies are underway to determine the possible role of this gene in the development of these features.

Dr. Elsea's research on the SMS critical interval includes an interest in identifying those persons who have smaller or unusual deletions of 17p11.2. That is, persons who may not have a documented deletion of 17p11.2 but have features consistent with SMS or those with a documented deletion of 17p11.2 who have an unusual phenotype that doesn't include all or most of the typical features observed in SMS are of great interest to Dr. Elsea.

Research on the genes deleted from chromosome 17 will lead to a better understanding of specific features of Smith-Magenis syndrome, as well as a better understanding of human development and behavior that may be beneficial to others who have disorders with

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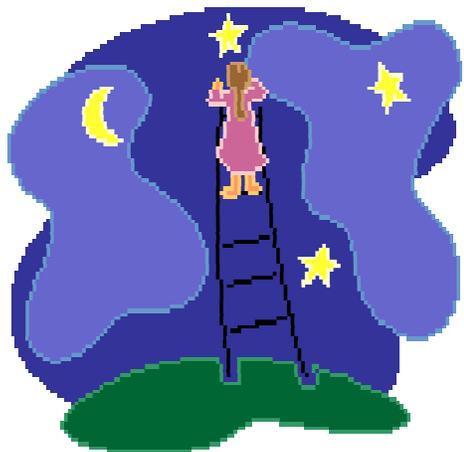
PRISMS BOARD MEMBERS NEEDED!!

In July, the two-year term for the present board will be coming to an end and there will be open positions. If you are interested in serving, please send a letter of interest, including any relevant skills or experience to:

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VISIONS, DREAMS AND WISHES

By Connie Bessette, PRISMS President



PRISMS board members gathered to set organizational long and short term goals and objectives. It began as a burdensome task, the kind that you may want to assign someone else; perhaps someone who owes you a favor.

We were surprised to find just hours into our meeting that this task was instead the antithesis of “burdensome.” The dialogue that unfolded was stimulating and exciting laden with passion, sensitivity and incredible wisdom. Guided by the PRISMS mission statement (below) the PRISMS WISH LIST was developed.

As you can see below we are sharing that list with you. Before you begin reading, here is some context for how we arrived at the 24 “wishes”: The full board of directors including Ann Smith was asked to dream, to think out loud about **what if**, in setting goals for PRISMS...you were not restricted by money or science, and what if...you could have anything you wanted for your SMS family and all other SMS families? And to take it one step further...what if you could change society or the professional community...or even your immediate or extended family, what would you wish for???

After hours and hours of discussion and deliberation we came to agreement on the following draft. Clearly some of the items below are more objective/task-like however, we did not want to restrict any creative thinking nor inhibit the process.

As you read through be ready to share your thoughts with us—more on this following the list.

WE WISH THAT:

1. Every person with SMS worldwide was diagnosed early. Ideally, before age 3.
2. The understanding and study of SMS would have application to society (i.e., education, medical mgt., behavior, etc.).
3. Society had an increased recognition and awareness about SMS.
4. PRISMS could influence law making (i.e. laws that support families with special needs children, tax relief, etc.).
5. SMS families had a better awareness of current PRISMS programs.
6. Educators understood SMS better and how do deal with our kids.
7. SMS families had easy access to information about SMS.
8. PRISMS would have a greater role in pragmatic support for families (i.e. respite services, behavior management, etc.).
9. PRISMS were working with a variety of other organizations (public and private) for research, support and funding.
10. Research could understand neurological pathways of medications in order to benefit maintenance.
11. We had a cure for SMS.
12. PRISMS had a portion of our budget set aside for research.
13. PRISMS had an endowment fund for research.

14. PRISMS would have a strong working relationship with the Professional Advisory Board.
15. We understood the cyclical changes of SMS kids better and how we could better manage them. Are their cycles lunar, seasonal, etc.?
16. There was a SMS Action Team that could be dispatched for crisis situations. This team would provide consultation.
17. PRISMS would be able to undertake every worthwhile project without resource restrictions.
18. We had an SMS camp for kids.
19. We had recreational and social opportunities for SMS parents as a group.
20. We had a SMS medical advisory and consultation team available for home professionals. Ideally, regionally based.
21. PRISMS had an annual operating budget of \$250,000 with an eventual budget of \$1,000,000.
22. PRISMS could provide a resource book for doctors and teachers (information re. SMS, management, covering birth to adults, family issues, support and encouragement. This book would be a practical handbook.
23. PRISMS had an 800 number.
24. All SMS Families had home access to a computer and the Internet.
25. We had more information about dealing with older kids (e.g., housing, jobs, etc.)

NOW FOR YOUR HOMEWORK:

I am asking that everyone who reads this gets involved.

Please take some time to think about the “wishes” on the list and feel free to add a wish that we have not thought of. Is there one way that you can help propel one wish, one piece of the puzzle into reality? Be as creative as you like or as simple as you like. Keep in mind, we will not be asking you to carry it out. It will become a working document of the PRISMS membership. We hope to post your responses—without your name or identifying information on our website and perhaps in a future newsletter. Your volunteering this information means nothing more or less than that. We simply want your ideas and thoughts.

Here are some ideas that board members shared: For # 3: distributing brochures (already achieved in part—through PRISMS membership drive). For # 6: develop a booklet/video on education strategies for SMS students.

While some wishes will not come to fruition for years it is reasonable to expect that many more will be realized in much less time.

PRISMS Mission Statement

PRISMS is dedicated to providing information and support to families of persons with Smith-Magenis Syndrome (SMS) and fostering partnerships with professionals to increase awareness and understanding of SMS. PRISMS is also committed to the support and promotion of research into the cases, optimal management and treatment of SMS.

As I leave you with this assignment, I am reminded of the words of Helen Keller: “Alone we can do so little. Together we can do so much.”



Miracles In Education

By Leanne VanDover and Christina Miller

PRISMS is pleased to have an opportunity to publish an article regarding an educational approach to teaching children diagnosed with Smith-Magenis Syndrome. In a continuing series of articles, Christina Miller and Leanne VanDover will provide us with insights and applications into the educational approach of Leanne's daughter Evin. In this issue of Spectrum we would like to introduce you to our authors and a very wonderful young lady named Evin VanDover. Please see our next issue of Spectrum for part 3 in this series.

Leanne VanDover, Mother to Evin

In the second part of this series I would like to share with you from my experiences of being Evin's mom as well as how my teaching career helped me to make a positive difference in Evin's education process.

In our last article you heard from my daughter's teacher, Christina Miller, about Evin developing independence at school as a result of the successful program that was implemented. The surprising thing for me was that as Evin began to gain independence so did I. As Evin improved I was able to stop worrying and relax. School is the only place that I can send Evin and not have some type of anxiety. This also came about as a result of Mrs. Miller's and the staff's wonderful attitude.

Prior to this program I was always being told about Evin's weaknesses. Mrs. Miller focuses on Evin's strengths. Every time I have a meeting at school, I leave feeling like I am a better Mom than before I came in, and that Evin is a great kid who contributes positively to others. Through this positive attitude Evin flourished and so did I. It also makes it so much easier when you have to address the weaknesses. I feel that this positive attitude is one of the key factors to the success of this program.

As I began experiencing the positive affects going on at school I began sending notes to say thanks. During the three years I taught special education I only heard from parents when they were angry or unhappy. No one ever said thanks for all your hard work, or I appreciate what you do for my child.

I don't think that most people realize what hard work it is to be a teacher, and a special ed. teacher works harder than most. So I began sending notes and thanking the teachers in person. I also tell the principal, director of special education, other parents, etc. when appropriate what a wonderful job the teachers and aides are doing. This may sound like a lot of work, but it is not. When in conversation with people I make it a habit to bring up the positive. I have found that this positive cycle builds and brings more positives about.

I have also found that when there are problems that need to be addressed no one takes offense, we work together for the good and positive because this is the established pattern. I also learned from my experience on the teacher's side of the desk that when I do become upset about something at school, that 90 to 95% of the time I do not have the whole story because I was not there. Many times when parents call they make the teacher feel attacked. I always try to talk to the teacher without my negative feelings showing through, because I usually find out some piece of information that changes my feelings. By doing this the teacher is not put on the defensive and the problem will be resolved much more quickly.

I have been able to build such a wonderful relationship with Evin's teachers that I now trust that they will always have Evin's best interest at heart.

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Christina Miller, Evin's Teacher

I began working with Evin when she was in second grade. At that time Evin was included in the general education classroom for opening, calendar, lunch, recess, art, music, P.E., science, and social studies. She had special education support for math, reading, and written language.

Each time Evin transitioned between classes she seemed confused and unsure of what was expected of her. Due to the open communication between home and school, I was able to develop a program that enabled Evin to better understand the routine and become more independent.

My special education classroom was structured using strategies from the TEACCH program developed in North Carolina. The most important strategy was the use of posted schedules for each student. Each student in the classroom had a schedule made up from photographs or digital photos, *Mayer-Johnson Board Maker* icons, and/or words. These schedules were posted inside the door of the classroom. When the students entered the classroom they removed the top "ticket" from their schedule, went to the area indicated, and placed their "ticket" on the Velcro in that area. When it was time to transition, the students were each given an index card with his/her name on it and were asked to "check their schedule". The students then went to the main schedule board, placed their index card in a library pocket above their schedule, and removed the next ticket.

Independent seat work was set up in a similar fashion. Each student's desk had a "work job" schedule on it. The students took the top "ticket" from their desk and matched it to a bin located on shelves across the room. Each bin had one "work job" with a task/activity that reinforced concepts already taught in a one-on-one or small group setting. The students took the bin to their desk, completed it, placed it back into the bin, and then into a larger "finished basket" located to the right of their desk. The "work jobs" were made to increase fine motor skills, alphabet and number recognition, writing skills, reading vocabulary, and math concepts. Seat work time was implemented to increase independence and time-on-task. This also allowed me time to pull each student individually for reading instruction.

A traveling schedule was eventually implemented to increase independence outside of the structured resource classroom. The traveling schedule was made of strips of poster board or one half of a manila folder. The activity/event icons were lined up on the schedule from top to bottom. Once the activity/event was completed, the icon was moved to the finished side of the strip or folder.

Within a very short period of time I saw an increase in independence and self-confidence in Evin. She understood what was expected of her and began to improve in all areas, both academically and behaviorally. The structure and consistency implemented in my classroom brought out the strengths in all of my students. Evin quickly began to thrive on the success that now comes more easily to her.

(Continued from page 1) *Looking for the Right Genes?*

features that are similar to or overlap those seen in SMS.

These studies will not lead to a "cure" for SMS but will open new avenues for treatment and other therapies that may make living with SMS much easier. If we know the genes that are involved, then we can understand how these work and how their absence leads

to the specific features of SMS--then perhaps we can design pharmacological therapies to circumvent or augment the function of a particular gene product.

Dr. Elsea's studies are supported by NIH (NICHD) and a research grant from the Michigan State University Foundation. Families who desire more information or who are interested in participating in research may contact Dr. Elsea directly at elsea@msu.edu or by phone at (517) 353-5597.



THIRD SMS INTERNATIONAL CONFERENCE

~ Conquering the Peaks and Valleys Together ~

We will gather at the Westin hotel and as we look out at the Rocky Mountain Range we will discuss the big issues—the ones about sleep and behavior. There will be many opportunities to experience the unusual—understanding and acceptance! Those who will gather for this conference know about impossible situations, and SMS problems that defy the experts (parents and professionals). You may find yourself apologizing, or embarrassed only to realize that no one has even noticed your child staring or touching or hugging or falling to the floor. You will be reminded over and over that you are not alone! And, everyone will have full bragging rights; our children will be the cutest, the smartest, and the funniest.

RESERVE YOUR HOTEL ROOM—REGISTER FOR THE CONFERENCE

You may register for the conference at the PRISMS website or through the conference brochure that has been mailed.

Dates:

Conference Registration:	
Wednesday, July 3rd	6:00 - 8:00 pm
Thursday, July 4th	7:00 - 8:30 am
Conference Begins:	
Thursday, July 4th	8:30 am
Conference Ends:	
Sunday, July 7th	11:30 am

Please Note

Registration is available online at our website:

<http://www.smithmagenis.org/conf2002.html>

Place:

The conference hotel is the beautiful Westin Westminster, 10600 Westminster Blvd., Westminster, Colorado. The hotel is conveniently located between downtown Denver and Boulder (approx. 15 minutes from both). There are also several campgrounds within a 15-35 mile radius of the Westin.

PRISMS has a special room rate of \$89 plus tax per night. You are responsible for making your own hotel reservations. Please reserve your room early by calling 1-800-WESTIN-1 (1800-937-8461). Tell them you are with PRISM or the Smith-Magenis group. The quantity of rooms is limited and the deadline for reservations and the reduced rate is June 1st. The earlier you call the hotel, the better.

**Conference Costs:
Registration and Meals (US Dollars)**

	Regular (Postmarked After May 11th)	Early Bird (Postmarked on or before May 11th)
-Each Adult	\$115	\$80
-Each Child	\$35	\$25
-Each SMS	\$35	\$25
Individual		
-Professional	\$135	\$100
-Professional	\$115	\$80
In-training		

Important Note: Please add \$30/family and \$45/professional if you are not a PRISMS 2001/2002 member.

Six meals will be provided: breakfast each day (4), lunch on Thursday and dinner on Friday. The Westin has 1 restaurant and 9 additional restaurants are in the Westminster Promenade, adjacent to the hotel.

Although the schedule has not been finalized, the following are some of the topic- workshops and facilitated discussions that will be offered:

- Research/Medication Updates
- Behavior Management
- Mothers' Workshop
- Siblings' Workshop
- Curbside Consults with professionals
- Overview of SMS for newly diagnosed families
- Mothers' Tea
- Sleep Issues
- Residential Options (for children and adults)
- Fathers' Workshop
- Parents' Panel
- Total Communication
- Family Networking (look to the experienced for ideas!)
- Fathers' Coffee

At the Westminster Promenade adjacent to the hotel you will find:

- Subway Sandwich Shop
- Dave and Busters (with arcade!!)
- Jackson's Hole All Am. Sports Grill
- Rock Bottom Brewery
- Rubio's Baha Grill
- The Sun Microsystems Ice Centre
- Hibachi Japanese Steakhouse
- Marble Slab Creamery
- Johnny Carino's Italian Eatery
- Tuk Tuk Thai Bistro
- AMC 24 Theaters
- Skateboard Rink

Special activities are being planned:

(Everything proposed is tentative and may involve extra cost.)

- Butterfly Pavilion outing (adjacent to hotel)
- Balloon Artist
- Trip to the Zoo
- Westminster City Park, with 45 miles of jogging and bicycle trails.
- Face Painting and Clown
- Story Teller/Puppet Show

Transportation options to and from airport/hotel:

Denver International Airport (DIA) is approx. 40 minutes from the hotel. Many restaurants are within walking distance from the hotel so a rental car may not be necessary. Airport ground transportation options include a rental car, taxi or Super Shuttle. In case you drive a care, hotel parking is complementary. Our Super Shuttle group rate is \$30 dollars round trip or \$18 one way for individuals age 9 and older. Super Shuttle can be reach at (303) 370-1300.

*******FINANCIAL ALTERNATIVES*******

Are you on a tight budget and thinking "how can I afford this"? Or, perhaps you are thinking I cannot afford any of this...so there is no way I can attend? PLEASE take a minute to consider the following:

In the past, families have received partial and full assistance from community resources such as Social Services, State Developmental Disability Councils (<http://www.acf.dhhs.gov/programs/add/state.htm>), Local Churches, Synagogues, Special Education Services, Knights of Columbus (<http://www.kofc.org>), The Shiners (<http://www.shriners.com>), Lions Club (<http://www.lionsclubs.org>) Salvation Army (<http://www.salvationarmyusa.org>), Angel Flight—serving the western US/Canada — (<http://www.angelflight.org>) or 1-888-4-AN-ANGEL, etc.

In addition, a limited amount of financial assistance is available directly from PRISMS. Please make sure that you complete and return the financial assistance application form by May 1st.

Childcare

An Activity Center will be available to children age 4 and older. This service is only available during workshop sessions. Franklin Kyle (Yes it is the same Franklin from the last 2 conferences!) will be in charge of the center and coordinating activities with volunteer staff. If you plan to utilize the activity center, please fill-in the appropriate conference registration form section. Space is limited and available on a first come, first serve basis. The deadline for reservation is June 1st. There will be a \$30 per child or SMS adult fee (\$10 per day) for the Activity Center. Parents will be required to complete emergency consent forms for each child participating in the Activity Center. These forms will be available at check-in.

International Members: Please note that all fees listed here are in US Dollars. Please go to <http://www.xe.com>

THANKS!

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Parent-to-Parent Program

PRISMS receives calls from parents of children newly diagnosed with SMS. Often they ask to talk to parents with SMS children of similar ages. As a link between parents, PRISMS has a Parent-to-Parent Program.

If you are interested in serving or participating in this program, we require your permission to release your name, address, and telephone number as appropriate requests are received.

I am interested in being a resource/contact parent for PRISMS and give my permission to PRISMS to release my name, address and phone number as a contact parent.

Name (print): _____

Telephone: _____

Address: _____

Email Address: _____

Signature _____



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