



Parents and Researchers
Interested in
Smith-Magenis Syndrome

Spectrum

Volume 6, Issue 2

Spectrum - The Newsletter of PRISMS

Spring • 2002

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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CONFERENCE COUNTDOWN



July 3rd - July 7th, 2002

The 3rd International Smith-Magenis Conference will be taking place in Westminster, Colorado July 3rd through the 7th, 2002 and we are getting very excited. Here is a hint of what's in store:

NAVIGATING THE MAZE OF PROFESSIONALS -

Dr. Judith Allanson will be our keynote speaker. She will discuss the multidisciplinary team approach needed for caring for persons with SMS. She will then moderate a panel presentation/discussion of: **CONNECT THE DOCS -**

Can Docs A and B talk to Doc C and therapist ONE and TWO? How do I, the SMS mom/dad explain a problem that most professionals do not understand? The panel will provide the ingredients necessary to make this dream a reality. You will learn to ask questions and outline concerns with words that will command attention—"those words the professionals understand and react to."

FATHERS ONLY! MOTHERS ONLY! SISTERS AND BROTHERS

ONLY! - This will be an opportunity to appreciate all the great things that you do in your family and get a few new tips. It will be a safe place to share struggles or just to listen; for sure a place to experience support.

TOOLS TO TREASURE – A panel of parents who really have "been there and done that" will share their favorite hints and secrets which may indeed have led to success!

FROM SLEEP TO MICE! - All the latest research in one place. Moderated by Ann Smith, Sleep experts will review the latest research and implication for behavior, mood changes, medications etc. Our SMS mice have experienced some struggles; however "Ann and Jim" have prevailed—we hear that they have been hard at work. We will get an update on that research also.

SPEECH LANGUAGE DEVELOPMENT: A ROAD MAP TO TOTAL

COMMUNICATION - It is important to understand normal speech and language acquisition to help children who are delayed in developing these skills. This interactive workshop will review speech, language development and various total communication approaches for fostering speech and language with the SMS child.

(Continued on Page 5)

Who's Who in



Officers

PRISMS President:

Connie Bessette
Francestown, NH

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Fairfield, CT

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Naperville, IL

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Houston, TX

Ellen Magenis, M.D.
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Portland, OR

Lorraine Potocki, M.D.
Baylor College of Medicine
Houston, TX

Beth Solomon, M.S.
NIH, W.G. Magnuson Clinical Ctr.
Bethesda, MD



Third Annual SMS Golf Tournament

August 2, 2002 - Francestown, NH

Just when you will start to recover from all the great things that went on at the 3rd International SMS Conference in Colorado, it will be time once again to get out those clubs and get swinging to raise money at our 3rd Annual SMS Golf Tournament to be held Friday, August 2, 2002 at the Troy Pines Resort in Francestown, NH.

Again, we are looking for not only sponsors and volunteers, but for our enthusiastic participants. If you would like to get involved in this wonderful and may I say most effective research and educational fundraising event, please drop us a line and let us know how you would like to help.

Additional information will be coming to you in the form of a brochure. Please continue to check out our website at <http://www.smithmagenis.org> for more information.

Should you wish to contact PRISMS sooner for more information, please feel free to do so, especially for hole sponsorships and donations. We would like to be made aware of any sponsorships or donations prior to June 15th, 2002.

IMPORTANT NOTICE

Investigators at Baylor College of Medicine would like to update their files!!!

If you have ever participated in the Smith-Magenis Syndrome Clinical Research Protocol at Texas Children's Hospital (TCH), Dr. Lorraine Potocki would like to be able to contact you to get an update on the health of your SMS child.

Please contact Dr. Potocki's office as soon as possible to provide her with your demographic information (address and phone number) so she may contact you to inquire on the current health of your SMS child. You may reach Dr. Potocki at lpotocki@bcm.tmc.edu or by fax at 832-825-4294 or via phone by calling 832-822-4292.

The past clinical data is being reviewed by Dr. Lupski and Dr. Potocki and other collaborators at TCH and the updated information that only you can provide is a vital part in the research of SMS.

Thank you for your continued participation in TCH's clinical research. Your help can make a difference!

Before the Conference, Before the Presentations, Before the Fellowship
Comes
The

2nd SMS Research Roundtable

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

With funding support from an NIH Bench-to-Bedside award, *PRISMS'* Professional Advisory Board (PAB) will again convene the 2nd *SMS Research Roundtable* on Wednesday, July 3rd preceding the parent educational conference.

This one-day session held exclusively for the exchange of scientific data and treatment approaches, brings together an invited group of basic science and clinical researchers to discuss their current research efforts and foster future collaborations.

A special morning *SMS Sleep Symposia* will concentrate on research to understand the chronic sleep disturbance in SMS in the context of current knowledge about the biology of sleep.

A summary of the exciting research findings will be presented on Saturday morning during the Research Update session.



EDITOR ERROR!

My apologies to Dr. Sarah Elsea for leaving out the contact information for her research project at the laboratory at Michigan State University.

Dr. Elsea's research 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.

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.



Bulletin Board

July 3rd - 7th, 2002
3rd International SMS Conference
Westminster, Colorado
For more information, please contact:
PRISMS at 603-547-8384
For room reservations at the Westin
Westminster, please call: 1-800-
WESTIN (1-800-937-8461)

May 25th, 2002
Heidelberg, Germany
1st Meeting of SMS Families
For more information, please call:
06221 395759 or 06221 759173

August 2nd, 2002
Third Annual SMS Golf Tournament
Francestown, New Hampshire
For more information, please contact:
PRISMS at 603-547-8384

August 3rd and 4th, 2002
SMS Family Gathering - York, PA
For more information call:
Sandra Tome at 717-757-6600



Meet One of Our Kids....Caroline Comford

♪♪Sweet CAROLINE♪♪

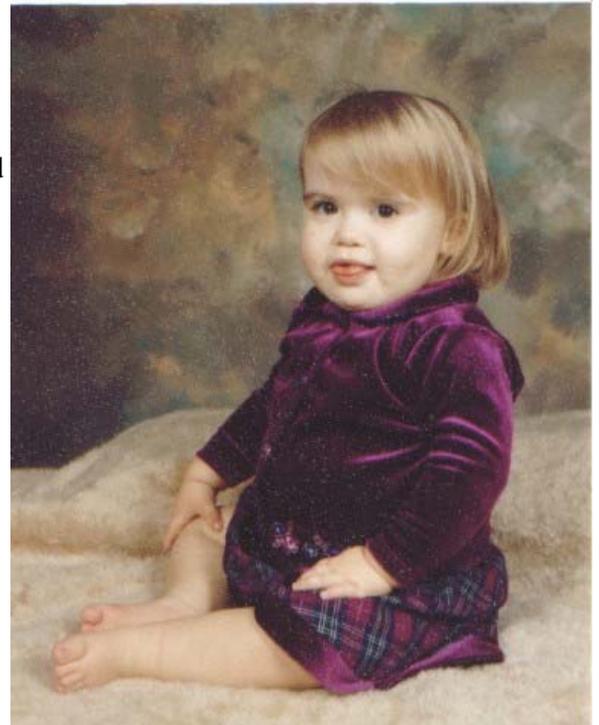
By Jennifer Comford jcomford@comcast.net

Hello, my name is Jennifer Comford and I want to share with you our family's story of our beautiful little girl, Caroline.

After what seemed to be a very uneventful pregnancy came a time of shock and dismay. Our beautiful little girl was thought to have Down syndrome. This prediction was made about two hours after her birth by a pediatrician sent over by the local children's hospital. He told us he suspected our baby might have Down syndrome and asked us for permission to start genetic testing. My husband and I agreed to the testing and there began our short journey. Ten days after her birth, Caroline was diagnosed with Smith-Magenis syndrome.

Those early days were awful times filled with uncertainty and fear, but also with overflowing amounts of love. We knew our task was not going to be easy but here it is a little over two years later. I cannot begin to describe the amount of joy Caroline has brought to our home. Things aren't always easy but we manage through. We feel blessed every single day that Caroline was diagnosed so early.

Caroline began speech therapy and occupational therapy at 2 months old and picked up physical therapy at about 6 months of age. Her progress has been tremendous and her support staff is absolutely wonderful. Caroline of course was and is still a late bloomer, but in my mind there is nothing she can't do until she proves it to me herself.



Joe and I want nothing more for Caroline than we want for Maddy, our seven-year old daughter. We want her to reach her fullest potential and whatever that turns out to be is fine, just as long as we know in our hearts we have afforded her every opportunity in life she deserves. What a truly spectacular feeling it is to see her accomplish things that she couldn't do just the week before.

Caroline has been walking independently for six months now and in fact is getting so strong that they reduced her physical therapy to one time per week. Speech of course is our biggest delay, but we are slowly progressing. Caroline is currently trying to say about three words, but you need a Caroline-to-English dictionary to understand them. Of course, Mom has already come equipped with that because I am now able to translate for her. Funny how moms just know.

Our journey with Caroline has really just begun. Sometimes I read what some of the other parents are going through and just weep and other times I'm almost jumping for joy at some of the other children's accomplishments. Please allow me to thank you all for sharing your knowledge, frustrations, strengths and accomplishments. You are leading the way for these uncertain times ahead and I have quickly learned that those of you who have already been there are the greatest source of information and support.

We have only been at this for two short years and we have a long road ahead, but even still I could write chapters on life with Caroline. I will spare all the details because I know you all "get it".

Thank you so much for allowing me to share a little piece of our life with Caroline.

RESOURCE MARKETPLACE – Have you been in the mood for a good catalog lately? This will be your one stop shop for those special and oh so elusive products.

CURBSIDE CONSULTS - Store up your questions and bring them directly to the doctors, scientists, and specialists: one-on-one time will be provided.

BARBECUE – A 4th of July tradition. Let PRISMS be your neighborhood and someone else will do the cooking!

MAKE AND TAKE ART ACTIVITIES – Designed just for the artist with SMS. Clean and creative! Parents and children will have an opportunity to have fun together.

EDUCATION

ON THE ROAD WITH SMS: TALES FROM THE NEW FRONTIER - Part one will be an overview of the most common educational difficulties encountered among students with SMS. It will offer practical strategies for working with school districts to create an optimal learning environment for students at all grade levels. **FROM AROUND THE CAMPFIRE TO INSIDE THE WAGON** - Part two will focus on classroom strategies and behavior modification techniques which have been successful for a variety of students with disabilities including SMS. It will offer practical strategies for positive home/school connections.

RIDING OFF INTO THE SUNSET/TRANSITIONING TO ADULTHOOD - Once a person with SMS reaches adolescence, families may face unfamiliar financial, vocational, and residential issues, as well as new challenges surrounding sexuality, choice, and independence and not least of all, the emotions that accompany such change. While there are no easy answers, and policies may vary from state to state, this workshop will explore these important issues while encouraging open dialogue among the participants.

MANAGING BEHAVIOR IN SMS: A HANDBOOK FOR SUCCESS! - Explore methods that actually work...and those that definitely don't...with an experienced special educator who is also Laura's Mom (SMS, 17 years old, and the inspiration for learning everything possible about behavior). We'll talk about "Why do they act like that?", "What can I do about it?". Ideas to be presented were gathered from many professionals and (best of all!) from successful experiences of parents of children with SMS. You bring your best methods too!

ACTIVITIES - Here are just a few of the activities we have planned both for families and children: Face painting, music, sing-a-long and dancing, puppet shows, bingo and other games. Also, we will have a visit from a loveable clown.

PRESENTERS, FACILITATORS, MODERATORS - This is a partial list of confirmed speakers: Ann C.M. Smith, *M.A.,D.Sc. (Hon.)*, Judith Allanson, *M.D.*, Sarah Elsea, *Ph.D.*, Brenda Finucane, *M.S.*, James Lupski, *M.D., Ph.D.*, Ellen Magenis, *M.D.*, Lorraine Potocki, *M.D.*, Beth Solomon, *M.S.*, Barbara Haas-Givler, *M.S.*, Connie Bessette, *MSW*, Randy Beall, Laurie Bellet, Margaret Miller, Ellen Elijah, *M.D.*, Mary Beall, *M.ED.*, Helene De Leersnyder, *M.D.*, Beth Kurtz, Caroline Pope, Staci Martin, *Ph.D.*

NOTE: At press time, the reduced rate of \$89.00 per room, per night at the Westin Westminster 1-800-WESTIN (1-800-937-8461) was still available. Also, please consult our website and or contact PRISMS should you have any questions regarding registration deadlines.



YOU'RE INVITED TO SERVE AND GUIDE PRISMS...

...the organization that supports extraordinary children, parents and caregivers; that serves as an international clearinghouse of information about the medical and educational needs of persons with Smith-Magenis syndrome to families, doctors, scientists, teachers, counselors and institutions and participates in research that is dedicated to improving the lives of our children.

Board of Director Positions to open in July 2002:

Secretary

At-Large Board Member - Public Relations

At-Large Board Member - Fundraising

At-Large Board Member - Family Support

The membership of PRISMS has doubled in the past three years! There are now 450 individuals, diagnosed with SMS, known to PRISMS!

PRISMS primary mission of providing support and information is more vital than ever. We need individuals with time and energy who can embrace the challenge ahead and lead.

All board positions are for a 2 year term. All board members have full voting rights and are expected to participate in the decision making process of the organization. Board members are expected to attend board meetings which are held in different locations approximately twice per year. Expenses for board meetings are paid by PRISMS.

The following positions will have basic requirements; however, each person's unique qualities and strengths will be integral in final definition of the positions.

The secretary will be responsible for maintaining the membership database, general record keeping of board meeting minutes and any telecommunications that involve changes/decisions in PRISMS administration.

The primary focus of the At-Large board positions will be raising awareness, fundraising and public relations. SMS continues to be vastly under diagnosed. And, although professionals now are better educated about SMS, many doctors, educators and service providers do not have the information they need to provide the best possible care.

We will be meeting these goals through a combined effort. Fundraising campaigns will be designed to educate and inform the general public about SMS. Improved public relations will bring awareness about SMS and the need for a strong support network. Our continued commitment to participate and support research will be strengthened through these efforts.

New board members will be selected by the present Board of Directors and representative from the Professional Advisory Board. Selection will be based on the individuals available time, relevant experience or skills/expertise, specific interests and goals as a member of the board. We are hoping to have a combination of SMS parents and others who may be professionals, relatives of an SMS person or someone who is simply interested in serving.

Since the birth of PRISMS the management of the organization has been handled by volunteers. While much of the work has been done by board members there have been others who have volunteered their time and services. The dedication of these volunteers has been defined by their compassion for all who are involved with SMS and the major contribution they have made of their precious resource—TIME.

Through the growth in our numbers and perhaps overzealous ambitions we have realized that fatigue and stress can sneak up on you; even when you are doing something that you love. Rather than lie down and take an extended nap we have made a few changes.

We are increasing our Board from a six member to an eight member board of directors. We will continue to seek and encourage others to come forward to volunteer for specific tasks. And, our greatest immediate change is the hiring of a part-time (ten hours per week) administrative assistant. This person will work during business hours at the home office in Frankestown, NH. (currently interviewing)

Please consider the difference you will make and the personal benefits to be gained by getting actively involved. Together we will make it better for our struggling families and dedicated professionals!

Application deadline is June 15th. Complete the enclosed form and mail it to PRISMS.

OUR SINCERE THANKS AND APPRECIATION TO MARY SCULLY, WHO HAS SERVED AS VICE-PRESIDENT AND LAURIE BELLET WHO HAS SERVED AS AT-LARGE MEMBER.

Continuing for a second term: Connie Bessette-president, Marsha Bach-vice president (previously serving as secretary), Randy Beall-treasurer and Margaret Miller-past president and consultant to the board. Ann Smith will continue as Chair of the Professional Advisory Board.

Application for PRISMS Board of Directors

Please complete and return by June 15, 2002 to: PRISMS 76 South New Boston Road – Francestown, NH 03043
*For additional information, you may contact PRISMS President, Connie Bessette, at cbessette@monad.net
Or PRISMS at info@smithmagenis.org*

Name:

Address: (Street, City, State & Zip)

Home Phone (with area code):

Work Phone (with area code):

Position Applying for:

- PRISMS Secretary**
- At-Large Member – Public Relations**
- AT-Large Member – Fundraising**
- At-Large Member – Family Support**

Please explain why you would like to be selected for this position:

What qualities and unique strengths can you bring to this position:

What are some of the goals you have for PRISMS if selected for this position:

Do you own a computer:

No

Yes

Email Address:

THANKS!

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Editors

Tracie L. Belcher

Beverly Frey

Brenda Finucane, M.S.

Beth Kurtz

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

IMPORTANT NOTICE

This will be the last hard copy mailed out regarding the conference.

Please see the website for any updates (sorry for the inconvenience to those without computers-hopefully your local library or a friend can help).



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Francestown, NH 03043**