



Traveling with SMS

By Pat Boschetto

Want to travel but are hesitant because of your SMS child/adult? Understandable. But you don't have to give up much needed vacations or miss out on a necessary trip. There is no assurance that you won't have issues, but there are a number of things you can do to minimize the chance of undesirable behavior. The key to a safe and comfortable trip is planning. Traveling with a "special" person is hard work, but if well-planned it can be a wonderful adventure for all.

Involve your SMS person in all aspects of planning the trip. Be enthusiastic when talking about your plans; this will encourage him to feel the excitement and enjoy the anticipation. Tell him when and where you are going and the mode of transportation, whether by plane, car, ship, train, bus. Prepare an itinerary for your convenience and give your child/adult a copy of his own. Use maps to show him the route of travel. Utilize a calendar to mark off the days until the date of departure; include the return date. Show him pictures of your target location, points of interest along the way, accommodations, planned activities and anything else that he will encounter. Family photos and the Internet are good sources to get pictures. Make a list of rules he is expected to follow; for the younger ones, pictures of rules are best. Discuss all of these things and let him know if he will have access to a swimming pool, youth activities, walks

along a beach, horseback riding, sightseeing -- anything.

Make reservations and special requests as soon as possible. On public transportation, ask for seating that will have the least impact on others in case of a behavioral problem. Instruct your SMS person on taking turns boarding and disembarking. Alert crew members of SMS special needs. Often, a crew member will have more success asking your traveler to buckle up, stow away luggage or other proper traveling requirements. Request a hotel room at the end of the hall to avoid disturbing other guests "when he gets loud." Discuss respect for other travelers. Don't hesitate to ask for assistance from staff.

If you're taking public transportation, allow plenty of time to arrive at the place of departure. Don't cut your time too short and create stress for everyone trying to make the plane, bus, etc. Encourage your SMS child/adult to pull his own suitcase; this gives him a sense of independence and keeps him focused on an important task. Take a small bag of essentials, i.e., medications, rewards for good behavior, while your luggage is out of reach for an extended time. Remember to reward good behavior more often than you do at home. Have your child keep a journal of his travels and include souvenirs to glue into his book.

For security purposes, attach your SMS child/adult's name, diagnosis and your cell number to his person and carry a recent picture of him in case he gets separated from you. If you will encounter security check-in, explain this process. Inform security person-

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nel of your special needs traveler in case you need their help getting through security.

It is wise to schedule an appointment with your doctor to ensure that your SMSer is healthy for traveling. For lengthy and out-of-country trips, ask your doctor to write a prescription with the GENERIC names of all medicines in the event you need to get more meds while away. If motion sickness is a potential problem, ask for a medication that will not conflict with other medications he is taking, and try it at home first for possible negative reactions. You may also want to discuss any sleep or bedwetting issues. Nobody wants to have to apologize for a wet bed at their hotel or at the home of a friend or relative. Check with your doctor before trying any new medications, including over-the-counter.

During the travel phase, have favorite items that your SMS person will be able to focus on to pass the time quietly. Suggestions are a



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A Message from PRISMS President...

Randy Beall

Greetings!

It's summertime and that means it's almost conference time!! Yahoo. This year's conference is planned for September 17-20 at the Reston Hyatt Hotel in Reston, VA. It's our major program and a significant financial commitment. We hope to see you there!

The planning for this event began several years ago and a handful of folks have worked for a long time to make this event a reality. Maggie Miller (conference chair), Alan Kleinfeld, Ann Smith, Julia Hetherington, Jeri Gawlowski and Mary Beall. Maggie and her team have worked countless hours to make this year's conference a success. You guys are the best!

As in the past, we hope to have our conference speakers provide their information for our website and future newsletters. In case you miss this year's conference, our next conference will be in Denver, CO in 2012. (...start saving now!)

We hope you know that we are trying to keep the costs of attending the conference as low as possible. In fact, the registration fee remains the same as the 2007 conference. The actual conference cost (which is primarily the hotel food costs), as well as the childcare cost, is much higher than the fees we charge. PRISMS is committed to providing a high quality, educational conference, presenting the latest research and most useful information available in an organized format. We really appreciate those of you who help us with grants and fundraisers. We couldn't hold this conference without your support. Thank you!!!

We still need corporate sponsors and folks to conduct local fundraisers to benefit PRISMS. In fact, we've enclosed a corporate sponsorship form with this newsletter. Our silent auction is another great way to get involved. Kellie Cooney (silentauction@prisms.org) is this year's Silent Auction Coordinator. If you can donate something, please contact Kellie. Please keep in mind that a dinner for 2 at your local eatery might be more valuable to you than the other folks at the conference. However, national chains work great. Also, many folks fly to the conference, so think about items that can be easily transported.

If you have a talent and would like to serve as a PRISMS volunteer, please contact Julia Hetherington, our Newsletter Editor and Volunteer Coordinator. Julia can be contacted via email at editor@prisms.org or phone 843-521-0156.

Finally, I'd like to welcome John Mayer, our newest board member. John is married to Debora Mayer and dad to Charley, age 24, with SMS. You may have read about "Light Bulb" Charley in our last newsletter. On behalf of the PRISMS board, welcome to the team, John!

The PRISMS board is here to serve you. If you have suggestions, concerns or comments, we would love to hear from you.

Warm regards,

Randy Beall
randy@prisms.org

Annual Report Enclosed

Meet the PRISMS Professional Advisory Board

Brenda M. Finucane, M.S., CGC

Many in the PRISMS family know our spotlighted Professional Advisory Board member, Brenda Finucane. One of the numerous things she does for SMS individuals is provide consultations and trainings about SMS to school districts and agencies across North America. Those families on the receiving end of these services know just how awesome she is!

Brenda is a genetic counselor by training and has a primary interest in learning and behavior in people with genetic conditions. She has a particular soft spot in her heart for all people with SMS and their families.

She has been employed by Elwyn as their Executive Director of Genetic Services since 1985. She is responsible for direction and provision of genetic diagnostic and counseling services at this large, nonprofit corporation serving adults and children with developmental disabilities. Prior to working at Elwyn, Brenda worked as a Peace Corps volunteer in an isolated equatorial village in Zaire, Africa.

At Elwyn, she is immersed in the special education and psychology fields. Her work allows her to combine her two passions: genetics and behavioral research. She is committed to increasing awareness about SMS and developing practical applications of research knowledge that families can use in their daily lives.

Working closely with children and adults with SMS has shown Brenda that their potential is often derailed by their challenging behaviors. For this reason, she feels it is essential that research focus on the underpinnings of their behavior and the development of practical interventions which can allow their true potential to shine through.

In 1993, having diagnosed several families with SMS in the Philadelphia area, she was instrumental in hosting the first regional SMS gathering. Soon after, her group joined forces with Ann Smith's families to form PRISMS. Brenda considers it a privilege to

have been involved with PRISMS from its inception.

In May of 1985 she received her M.S. in Human Genetics from Sarah Lawrence College located in Bronxville, New York. But this has not been her last academic endeavor! Brenda has published articles in numerous professional journals.

A few publications specific to Smith-Magenis Syndrome include:

Finucane B, Kurtz M, Scott CI (1992): New mental retardation syndrome with deafness, distinct facial appearance, and skeletal anomalies. [Am J Med Genet](#) 43:844-847.

Finucane B, Jaeger E, Kurtz M, Weinstein M, Scott CI (1993): Eye abnormalities in the Smith-Magenis contiguous gene deletion syndrome. [Am J Med Genet](#) 45:443-446.

Finucane B, Kurtz M, Babu V, Scott CI (1993): Mosaicism for deletion 17p11.2 in a boy with the Smith-Magenis syndrome. [Am J Med Genet](#) 45:447-449.

Finucane B, Konar D, Haas-Givler B, Kurtz M, Scott CI (1994): The spasmodic upper body squeeze: a characteristic behavior in Smith-Magenis Syndrome. [Dev Med Child Neur](#) 36:78-83.

Finucane B (1997): Smith-Magenis Syndrome (letter). [Ophthalmology](#) 104:732.

Finucane B, Dirrgrl K, and Simon EW (2001): Characterization of self-injurious behaviors in children and adults with Smith-Magenis Syndrome. [Am J Ment Retard](#) 106:52-58.

Finucane B: Psychiatric diagnoses as they relate to Smith-Magenis Syndrome. *Spectrum* newsletter, Winter 2001, PRISMS.

Finucane B: The long road to diagnosis, by way of a needle in a haystack. *Spectrum* newsletter, Summer/Fall, 2003, Parents and Researchers Interested in Smith-Magenis

Continued.....



"I am humbled every day by the endurance, creativity, and humor of families meeting the challenge of SMS."

Brenda Finucane

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.

Syndrome.

Finucane B (2008): Embracing the inner toddler in people with Smith-Magenis Syndrome. *Spectrum*, Summer 2008, PRISMS.

Finucane, B. and Haas-Givler, B. Smith-Magenis Syndrome: Genetic basis and clinical implications. *Journal of Mental Health Research in Intellectual Disabilities* (in press).

Dykens E, Finucane B, Gayley C (1997): Brief report: Cognitive and behavioral profiles in persons with Smith-Magenis syndrome. *J Aut Dev Dis* 27:203-211.

Edelman EA, Girirajan S, Finucane B, Patel P, Lupski JR, Smith ACM, Elsea SH (2007): Gender, genotype, and phenotype differences in Smith-Magenis syndrome: A meta-analysis of 105 cases. *Clin Genet* 71:1-11.

Elsea SH, Juyal RC, Jiralerspong S, Finucane BM, Pandolfo M, Greenberg F, Baldini A, Stover P, and Patel PI (1995): Haploinsufficiency of cytosolic serine hydroxymethyltransferase in the Smith-Magenis syndrome. *Am J Hum Genet* 57:1342-1350.

Juyal RC, Finucane B, Shaffer LG, Lupski JR, Greenberg F, Scott CI (1995): Apparent mosaicism for del(17)(p11.2) ruled out by FISH in a Smith-Magenis syndrome patient. *Am J Med Genet* 59:406-407.

Smith A, Allanson J, Elsea S, Finucane B, Haas-Givler B, Gropman A, Johnson K, Lupski J, Magenis E, Potocki L, Solomon B (updated August 2006) Smith-Magenis Syndrome in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. Copyright, University of Washington, Seattle. 1997-2006. Available at <http://www.genetests.org>.

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It's **silent auction** time again! This is a major fundraiser at our conference and we need everyone's help! Please consider making a donation. Items which can be transported easily are especially appreciated by those traveling to the conference. But remember, great items WILL find a way home! Do you have a craft or a business, or know someone who could donate something special?? Even if you are unable to attend, this is a great way to support PRISMS. Please contact Kelly at silentauction@prisms.org if you have any questions or anything to donate!!!



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 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.

portable CD or DVD player, laptop, MP3 player, Ipod, books on tape (be sure to have earphones to avoid bothering others). Consider packing a surprise bag of new [toys] that you can pick from about every hour or whenever he becomes restless. These can be small, inexpensive items that if lost, no worries! On an airplane, bus or train, get up and stretch, and take a short walk when possible. Keep hydrated to avoid cramping. When traveling by car, plan frequent stops to get out and run off some steam. Be sure to wear comfortable, loose-fitting clothing and shoes. Bring along healthy snacks and try to avoid fast food restaurants. Maintaining a diet similar to daily life at home will help alleviate possible behavior and stomach distresses.

Remember to schedule plenty of down time for everyone to relax and regroup. Overstimulation is the enemy, as you know. Plan well to avoid mishaps, and be flexible -- something will always come up to change your plans. Keep as much normalcy as possible -- eating and sleeping times, etc. If staying with friends or relatives advise them in advance what to expect with your SMS child/adult and their needs so that they can be prepared. If you can, travel off-season to avoid the crowds -- and the peak season pricing! Go and explore the world. It's fun out there!



Helpful websites:

<http://specialchildren.about.com/od/travelwithspecialneeds>

www.tsa.gov/travelers/airtravel/specialneeds

www.iser.com/resources/traveling-specialneeds.html

Preparing Emotionally for the Annual PRISMS Conference

By Mary Beall

If this is your first PRISMS conference, or if you are newly diagnosed, here are some words of wisdom from one who has been there. Actually, even if this is not your first conference, it is good information to think about before and as you go through the next few days.

1. Don't take every word as gospel. You may hear "You should do this..." "Make sure you..." "Do everything you can to..." In reality, what you are hearing are good ideas, and while the information is based on research and practice, you need to find what works for you, your child, and your family. You cannot do everything, nor should you be expected to—make sure you always take time to be a mom or dad and just enjoy your children.

2. Spend time with some of the other families. Try to find a family who lives near you, another family who has a child your child's age, and a family with an older child. You may find that you feel an instant bond with them. They will understand your experiences and will enjoy your stories, both good and bad. You will probably find yourself laughing a lot as we delight in the antics of our precious kids. (Look for "Parent Connect" opportunities in the conference schedule).

4. Don't feel like you have to go to every session to qualify as a good parent. The reality is that you cannot go to every session because there are

so many of them, and they will not all pertain to you anyway. Pick a few topics that you want to learn more about and focus on those.

...You need to find what works for you, your child, and your family.

5. Use the conference to take the time to go for a walk, have a cup of coffee, exercise, or get some much needed extra sleep. Give yourself the time to be introspective; look at all you have done, and while there is more to do, there always will be. Pat yourself on the back and then set a couple of goals for yourself.

6. You will have a chance to hear from the principal researchers and specialists who study Smith-Magenis Syndrome. They will provide us with the most up-to-date information available, and will inform us about their research projects. Come prepared to take notes. You may even have time during these sessions to ask general questions that apply to all of the participants.

7. Take advantage of the Curbside Consults session. This is your chance to speak to the specialists about your individual concerns. Be prepared for this unique opportunity by bringing specific questions they can answer quickly. (As you can imagine, there will probably be a long

line of other parents waiting for their chance).

8. Come early enough to attend the opening party, and be sure to stay for the last day's sessions where you will get to be a part of PRISMS plans and will hear a parent panel talking about the everyday experience of raising a child with SMS.

9. Participate in the Silent Auction benefitting PRISMS, so that we can continue to support parents and families. Bring an item to sell, and bid, bid, bid!

If you are able to come, you will find it extremely worthwhile. Have a great time!



SMS Medical Management Guidelines

These guidelines were developed and approved by PRISMS Professional Advisory Board. They are published as part of the in-depth review of SMS that appears in GeneReviews. A link to GeneReviews is on the PRISMS website.

Management involves evaluation for manifestations of SMS and treatment to mitigate symptoms.

Evaluations.

At the time of diagnosis of SMS, a series of baseline evaluations are recommended to guide medical management, including the following:

- Complete review of systems at the time of diagnosis
- Physical and neurological examination
- Renal ultrasound examination to evaluate for possible renal/urologic anomalies (~20% of individuals with SMS)
- Echocardiogram to evaluate for possible cardiac anomalies (<45% of individuals with SMS); follow-up depending upon the severity of any cardiac anomaly identified
- Spine radiographs to evaluate for possible vertebral anomalies and scoliosis (~60%)
- Routine blood chemistries, qualitative immunoglobulins, fasting lipid profile, and thyroid function studies
- Ophthalmologic evaluation with careful attention to evidence of strabismus, microcornea, iris anomalies and refractive error. Treatment with corrective lenses as indicated.
- Comprehensive speech/language pathology evaluation, with special emphasis during early childhood
- Assessment of caloric intake, signs and symptoms of gastroesophageal reflux (GER), swallowing abilities and oral motor skills with referral as warranted for full diagnostic evaluation
- Otolaryngologic evaluation to assess ear, nose, and throat problems, with specific attention to ear physiology and palatal abnormalities (clefting; velopharyngeal insufficiency). Recurrent otitis media may require treatment with tympanostomy tubes.
- Audiologic evaluation at regular intervals to monitor for conductive and/or sensorineural hearing loss. Amplification may be required.
- Multidisciplinary developmental evaluation, including assessment of motor, speech, language, personal-social, general cognitive, and vocational skills
- Early evaluation by physical and/or occupational therapists and implementation of services as needed
- Sleep history with particular attention to sleep/wake schedules and respiratory function. Sleep diaries may prove helpful in documenting sleep/wake schedules. Evidence of sleep-disordered breathing warrants polysomnogram and overnight sleep study to evaluate for obstructive sleep apnea.
- Assessment of family support and psychosocial and emotional needs to assist in designing family interventions
- Parental chromosome analysis to permit accurate recurrence risks and provision of genetic counseling

Recommended Annually.

- Multidisciplinary team evaluation is optimal, including physical, occupational and speech therapy evaluations, and pediatric assessment to assist in development of Individualized Educational Program (IEP). Periodic neurodevelopmental assessments and/or developmental/behavioral pediatric consultation can be an important adjunct to the team evaluation.
- Thyroid function
- Fasting lipid profile
- Routine urinalysis
- Monitoring for scoliosis
- Ophthalmologic evaluation
- Otolaryngologic follow-up for assessment and management of otitis media and other sinus abnormalities
- Audiologic evaluation at regular intervals to monitor for conductive or sensorineural hearing loss

Recommended as Clinically Indicated.

- EEG in individuals who have clinical seizures to guide the choice for antiepileptic agent. For those without overt seizures, EEG may be helpful to evaluate for possible sub-clinical events in which treatment may improve attention and/or behavior. A change in behavior or attention warrants re-evaluation.
- Urologic workup if history of frequent urinary tract infections
- Neuroimaging (MRI or CT scan) in accordance with findings, such as seizures, and/or motor asymmetry
- Individuals with SMS documented to have larger deletions:
 - Specific screening for adrenal function; and

Detailed assessment and attention to peripheral neurologic function in individuals with SMS with large deletions involving the PMP22 gene, which is associated with hereditary neuropathy with liability to pressure palsy (HNPP)
Monitoring for hypercholesterolemia and medical treatment if indicated

Treatment Recommendations Include the Following:

Ongoing pediatric care with regular immunizations

From early infancy, referrals for early childhood intervention programs, followed by ongoing special education programs and vocational training in later years

Therapies including speech/language, physical and occupational, and especially sensory integration

During early childhood, speech/language pathology services should initially focus on identifying and treating swallowing and feeding problems as well as optimizing oral sensory motor development.

Therapeutic goals of increasing sensory input, fostering movement of the articulators, increasing oral motor endurance and decreasing hypersensitivity are needed to develop skills related to swallowing and speech production.

The use of sign language and total communication programs as adjuncts to traditional speech/language therapy is felt to improve communication skills and also have a positive impact on behavior. The ability to develop expressive language appears dependent upon the early use of sign language and intervention by speech/language pathologists.

Published data about the optimal intervention and behavioral strategies in SMS are limited to anecdotal and experiential findings.

Use of psychotropic medication may increase attention and/or decrease hyperactivity. No single regimen shows consistent efficacy.

Behavioral therapies are integral in behavioral management. Special education techniques that emphasize individualized instruction, structure, and routine can help minimize behavioral outbursts in the school setting.

Therapeutic management of the sleep disorder in SMS remains a challenge for physicians and parents. There are no published well-controlled treatment trials. Early anecdotal reports of therapeutic benefit from melatonin remain encouraging. Dosages of 2.5 mg to 5.0 mg (6 mg maximum) taken at bedtime have been tried, providing general improvement of sleep without reports of major adverse reactions. However, melatonin dispensed over-the-counter is not regulated by the FDA; thus, dosages may not be exact. No formal melatonin treatment trials have been conducted. A monitored trial of four to six weeks on low-dose (1-3 mg) melatonin may be worth considering in affected individuals with major sleep disturbance. A single uncontrolled study of nine patients with SMS treated with oral α -1-adrenergic antagonists (Acebutolol 10 mg/kg) reported suppression of daytime melatonin peaks and subjectively improved behavior [DeLeersnyder et al 2001]. This treatment, however, did not restore nocturnal plasma concentration of melatonin. A second uncontrolled trial by the same group [DeLeersnyder et al 2003] combined the daytime dose of Acebutolol with an evening oral dose of melatonin (6 mg at 8PM) and found that nocturnal plasma concentration of melatonin was restored and nighttime sleep improved with disappearance of nocturnal awakenings. Parents also reported subjective improvements in daytime behaviors with increased concentration. The contraindications to using α -1-adrenergic antagonists must also be considered, including asthma, pulmonary problems, cardiovascular disease, and diabetes mellitus. Prior to beginning any trial, the child's medical status must be considered. It is also beneficial to have an understanding of the child's baseline sleep pattern.

Enclosed bed system (Vail bed)

Respite care and family psychosocial support help assure the optimal environment for the affected individual

Developed and adopted by PRISMS Professional Advisory Board and revised 15 March, 2004. Smith-Magenis Syndrome. In: GeneReviews at GeneTests: Medical Genetics Information Resource (database online). Available at <http://www.genetests.org>

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Announcing the SibKids and SibNet Listservs

The Sibling Support Project of the Arc of the United States is pleased to announce SibNet and SibKids. SibNet and SibKids are the Internet's only listservs for and about brothers and sisters of people with special health, developmental, and emotional needs. Both SibKids (for younger brothers and sisters) and SibNet (for older siblings) allow brothers and sisters an opportunity to connect with their peers from around the world. Both listservs have members from the US, Canada, Australia, England, Japan and elsewhere. SibNet (started in 1996) and SibKids (started in 1997) are remarkably warm, thoughtful, and informative communities where young and adult brothers and sisters share information and discuss issues of common interest. Anyone who has email can subscribe to SibKids and SibNet. For a no-cost subscription and to learn more about SibKids and SibNet, please visit the Sibling Support Project's Web Page (see address below). Finally, if you have further questions about SibKids, SibNet, our Sibshops, or the work of the Sibling Support Project, please contact:

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Research and SMS

One of the very unique things about PRISMS is the fact that it is truly parents and researchers working together! The following is a listing of what is currently going on. More info can be found by going to PRISMS website and clicking on the research links.

GROWTH PATTERNS in SMS/NIH

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 & 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. 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.

Virginia Commonwealth University

Projects in Dr. Sarah Elsea's lab are focused on the molecular analysis of chromosome 17p11.2 deletions and the identification and analysis of the genes involved in SMS. The goals of the various studies are to identify and characterize the genes most critical to the syndrome. For this reason, Dr. Elsea is interested in the evaluation of small and/or unusual deletions involving chromosome 17p11.2.

Recent studies in Dr. Elsea's lab led to the identification of the RAI1 gene as the primary culprit in SMS. Mutation or deletion of this gene can lead to Smith-Magenis syndrome. The precise function of the RAI1 protein is not yet clear. Studies in the lab are also focused on determining the normal function of this protein in the cell and in development and behavior.

While Dr. Elsea is interested in all 17p11.2 deletions, she is particularly interested in individuals who have clinical features of SMS but who do not have a documented chromosome 17 deletion.

Evaluation of the RAI1 gene in persons fitting these criteria can be performed on a research basis. For more information, call Dr. Elsea at (804) 628-0987 or send an email to selsea@vcu.edu

Research Update! Doctors Sarah Elsea and Santhosh Girirajan of Virginia Commonwealth University have published a research paper on SMS with the European Journal of Human Genetics. The paper is available through the link on the PRISMS website.

National Human Genome Research Institute/ National Institutes of Health

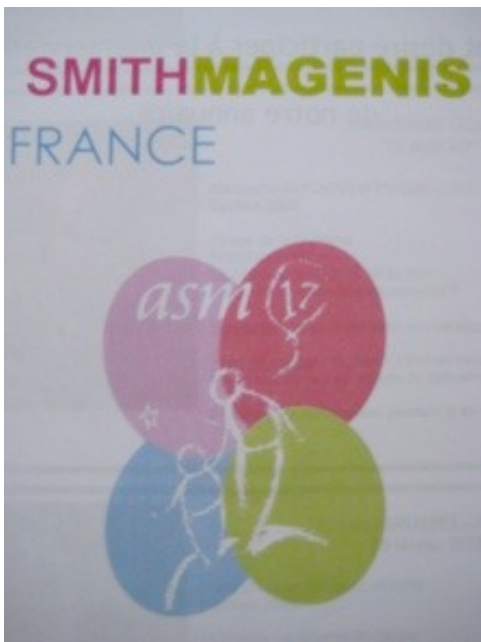
Building on the unique scientific expertise available at the National Institutes of Health (NIH), an inter-disciplinary SMS Research Team of clinical and basic science researchers was established to conduct pioneering, state-of-the-art research to further our understanding of this complex rare microdeletion syndrome. Ann C.M. Smith, M.A., D.Sc. (hon.), co-discoverer of the syndrome, serves as the principal investigator of the NIH research protocol entitled, "Natural History Study of the Clinical and Molecular Manifestations of Smith-Magenis Syndrome (SMS)" (01-HG-0109). The primary goal of the NIH SMS research study is to gain a better understanding of the range and type of medical problems that occur in SMS and how they change over time. The NIH study has a special focus on defining the underlying speech, language and behavioral aspects of SMS, as well as trying to understand its natural history. Go to PRISMS website to read more about this program, or send an

email to Ann C.M. Smith (acmsmith@mail.nih.gov) for further information. In addition to these clinical aspects, the NIH research includes a basic science component to study the SMS deletion interval of 17p11.2. To this end, a SMS Research Registry and Tissue Bank has been established at the National Human Genome Research Institute at NIH. The SMS Tissue Bank permits ongoing and future collaborative studies to elucidate the gene(s) and mechanism(s) underlying SMS.

Baylor College of Medicine

Clinical and molecular research on SMS continues to be active at Baylor College of Medicine and the Texas Children's Hospital. The first genetically engineered mice with SMS were born in August 2000. Hopefully these mice will provide a better understanding of the genes in this region. While patient enrollment for SMS clinical research at Baylor is now limited to those individuals with small or unusual-sized deletions of 17p11.2, Dr. Lorraine Potocki is actively recruiting all SMS patients (and their parents) for the purpose of molecular (DNA) analysis of the SMS region. If you would like to participate in their research studies, please visit the Baylor web site, contact Dr. Potocki via email (lpotocki@bcm.tmc.edu), or call her at (832) 824-4292.





On Sunday, May 3, 2009, forty-five French families of children with SMS welcomed me as their keynote speaker at their ASM17 conference in Paris. Organized by SMS parent Valerie Bedard, L'Association SmithMagenis France (ASM17) is the support network in France that connects over 90 SMS families. Donning headsets, French SMS families and clinicians attending the conference heard my talk translated instantly from English to French in "real-time." It was like being at the United Nations with everyone wearing headsets to understand my English and I wore a headset to understand all their questions in French. Dr. Helene DeLeersnyder, who is well known to PRISMS and American SMS families because of her research to understand the inverted melatonin rhythm, also spoke about medical management of SMS. Dr. DeLeersnyder serves as a physician advisor to the ASM17 group and follows several French children with SMS in her pediatric practice in Paris. During the afternoon, concurrent breakout workshops focused on managing behavior and medical support issues for younger and older children generated discussion and provided support for attending families. Coffee breaks in the morning and afternoon were

An American in Paris

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

the place to be, with French pastries and great shared conversation. I enjoyed tasting the special box of candy given to me by a young man with SMS who works in a chocolate factory that makes the delicious chocolate treats!

At the end of the day, the children and adults with SMS regaled everyone with their parade of "hats" marching onto the front stage for the conference finale of French songs — everyone with SMS wanted a turn holding the microphone — that closed the conference. Helene and I were both "showered" with thanks, strong SMS hugs, lovely flower bouquets, and two very special posters made with the loving hands (and handprints) of all the children (see inset)! The kids were amazed to hear me sing Frère Jacques (Are You Sleeping) in French. What better theme song in French/English for SMS! Clearly, there is no language barrier to understand the universal story of SMS. Like PRISMS' upcoming conference in Sept. 2009, the ASM17 conference provides persons with SMS and their families, caregivers, primary care providers, health professionals and researchers the opportunity to gather together for support, education, research related updates and advocacy!

During my short week stay in Paris I enjoyed the special Parisian hospitality offered by Helene DeLeersnyder and her husband, Jean Marc, staying in their lovely Paris home. The week provided the opportunity to con-

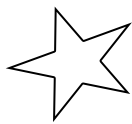


Ann Smith, M.A., D.Sc(hon) and Dr. Helene De Leersnyder

tinue our ongoing research collaboration AND to enjoy PARIS with walks along the Champs-Élysées to take in the sights and delicious cuisine - bon appetite!



SMS SuperKid!



Rachel Kirklin



Another SuperKid has emerged. Last fall, Rachel entered artwork in the PTA Reflections Program at her middle school. The Reflections Program is a national PTA program designed to enhance the arts in education and to allow opportunities for students to express themselves creatively. Each year the program has a theme that students are required to interpret through their choice of six areas of expression: dance choreography, film production, literature, musical composition, photography, and the visual arts (which includes art forms such as drawing, painting, print making, and collage).

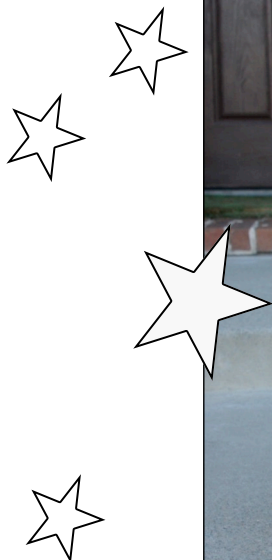
The theme for the 2008-2009 school year was, "Wow!" Rachel decided to express the theme by depicting a large group of birds flocking in a tree and its surrounding area. She was inspired by the unique way in which grackles, large blackbirds, flock in enormous groups in the landscape trees that line the parking lots of retail centers. At around sundown, the flocks can be unnerving and reminiscent of *The Birds*. She remembered how our family has been "wowed" by this fascinating sight on many occasions. Her depiction took the form of an acrylic painting that consisted of a combination of washes, brushwork and sponge painting on illustration board.

The entries are judged by a panel of objective and qualified individuals from the community. The judges are unable to see the names of the artists. They are only able to see the name of the artwork and any artist statement that has been provided. The judging criteria are that 50% of the score is based on interpretation of the theme, 25% of the score is based on creativity, and 25% on artistic merit. The artwork in the top 20% of all the categories combined moves to the Council Level.

Since this is a PTA program, there are no accommodations for Special Needs. I am proud to say that Rachel's artwork competed right alongside artwork of typical kids her age. When judged at the school level, Rachel's painting received a top score of excellent, moving her artwork to the Council Level along with five other students.

At the Council Level, Rachel's painting received an Honorable Mention. Way to go, Rachel. "Wow!" We are proud of you!

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



September 17-20



reston 2009
building
bridges
of hope

PRISMS 6th INTERNATIONAL CONFERENCE

Reston, Virginia

Come to learn about SMS, past, present and future, and to share information and strategies with other families.

Sessions on diagnosis, behavior, therapeutic interventions, medications, school issues, estate planning, caregiver issues, research updates and the Silent Auction, Parent Connect and more!

CONFERENCE SCHEDULE OVERVIEW:

Thursday, September 17

- 2:00 – 4:00 pm – Parent Connect
- 4:00 – 7:30 pm - Registration
- 5:00 – 7:00 pm - Welcome Reception

Friday, September 18

- 7:00 – 8:00 am - Registration & Continental Breakfast
- 8:00-5:00pm - Conference Sessions

Saturday, September 19

- 7:00 – 8:00 am – Continental Breakfast
- 8:00 am – 5:00pm - Conference Continuation

Sunday, September 20

- *8:00 - 11:00 am – Continental Breakfast and Conference continuation.
- ***Note** – conference ends promptly at 11:00 am.

REGISTRATION FEES:

Members	Up to 9/4/09	After 9/4/09
Adult (15+)	\$175	\$225
Child (14 & under)	\$75	\$100
SMS Individual	\$75	\$100
Professional	\$250	\$300
Professional In-Training	\$200	\$250

You can register online for the conference at www.prisms.org. If you need a registration form mailed to you, please call the PRISMS office at 972-231-0035.

All of the details of the conference are available on the PRISMS website at www.prisms.org.

HOTEL COSTS:

At the Reston Hyatt Hotel, PRISMS has a special room block rate available at: \$125 single/double per night, (Thurs-Sat only), plus applicable tax. This special rate is available only until 9/4/09. Please reserve your room early by calling 1-800-233-1234.

Hotel registration can be made on-line at: <http://resweb.passkey.com/go/PRISMS>



CHILDCARE:

Corporate Kids will provide childcare on site for SMS children and their siblings, ages 4-18 years. Corporate Kids has provided high quality children's programs and youth services for over seventeen years. This year's camp will provide a highly structured environment and a 1 staff to 2 children ratio. All staff will be experienced childcare professionals with experience working with special needs children. Daily activities will be scheduled based on your child's particular needs and comfort. Upon completion of your childcare registration form, a representative of Corporate Kids Events will contact you to review your child's needs and answer any questions you may have in regards to the upcoming program.

The subsidized price of \$150 per child (for two days) does NOT include the child's conference registration fee of \$75. Childcare will be available on a first come first served basis. Registrations for Corporate Kids must be made before **Sept. 4th**.

Program dates are September 18-19, 2009. **Fee: \$150** per child for both days. Space is strictly limited. Register via the link www.prisms.org. Or you can call 1-800-757-3580 and ask for Michael Whittle, PRISMS event childcare manager.

We hope you will attend, whether it is your first conference or your sixth. Come meet new friends and visit with old friends. You will also have the opportunity to meet with the researchers and professionals who are spearheading the latest research on SMS. Where else can you go and have immediate access to our Professional Advisory Board and dedicated professionals!

Be a part of the dynamic partnership with our professionals and our families as we ALL "build the bridge" to a better understanding, acceptance, and quality of life for persons with SMS.



*Parents and Researchers Interested
in Smith-Magenis Syndrome*

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