prisms

spectrum

PARENTS AND RESEARCHERS INTERESTED IN SMITH-MAGENIS SYNDROME

INSIDE THIS ISSUE

PRESIDENT'S MESSAGE

GOOD WISHES TO ALL, AND WELCOME TO THE FALL 2017 PRISMS NEWSLETTER.

Along with the change of the seasons and the approach of the holidays, I am finding there is much going on with the family, my work and with PRISMS. It is an exciting time for me—and I hope it is for you, too.

The PRISMS Board just completed our October face-to-face meeting. Twice a year we meet in person to conduct our work. I return home from these meetings exhausted, but inspired by our plans for the future. We met in Pittsburgh to review our location for our 2018 conference and to explore resources in the area for our families. The city seemed great, and I have every confidence Pittsburgh will be an excellent location for our gathering. Don't miss out on the 2018 conference—make your plans to join us July 19 - 21, 2018! Ready or not—Pittsburgh here we come!

I am delighted to highlight a few other topics.

The PRISMS Patient Registry launched less than a month ago and in that time, around 40 people have enrolled. It is a great start – but we need your participation. Our hope is that the registry will grow and include information about a large number of people with SMS. It is very easy to enroll via a simple on-line questionnaire. The process begins

SMITH-MAGENIS SYNDROME PATIENT REGISTRY

by contacting the registry (actually the PRISMS genetic counselor, Dianne Samad) through a portal at prisms.org. As the registry builds and more information is gathered, this will become a powerful resource for researchers focused on improving the lives of our family members. We have enrolled our son Charley; I hope you will enroll as well.

In 2018, PRISMS will be celebrating our 25th year of serving the SMS community. To mark the occasion we have been collecting information and developing a history of our organization. Throughout 2018 we will share our story in various ways. There are many people to recognize who, over the years, gave much to help our families. Needless to say, we owe much to the insights and passions of Dr. Ellen Magenis and Ann Smith, both of whom the syndrome is named for. Looking back at the very early years of PRISMS, we have steadily developed our ability to serve our families over those 25 years.

25 Years prisms 1993-2018

And finally, as the end of the year approaches, along with appeals from many other organizations, you will receive a request from PRISMS to make a financial gift as part of our annual appeal. I hope you will join me and make a year-end donation. Support from our community is what keeps us going. Your contribution truly makes a difference in advance. We all thank you for this support.

I look forward to a great year in 2018. Best wishes to you all!

John Mayer
President, Board of Directors

Pittsburgh will host the 2018 PRISMS International Conference.

EMPOWER THE SMS COMMUNITY WITH PRISMS!

> "CONSIDER SUPPORTING PRISMS IN YOUR END-OF-YEAR GIVING AND HELP US TO EMPOWER THE ENTIRE SMS COMMUNITY"

It's hard to believe that 2017 is nearing its end. On behalf of PRISMS, I wish you and your family a happy and healthy holiday season! I know that during this time of year many organizations compete for your attention to support their charitable efforts. I ask that you consider supporting PRISMS in your end-of-year giving and help us to empower the entire SMS community!

Nearly 25 years ago, a group of parents and researchers formed the nonprofit Parents and Researchers Interested in Smith-Magenis Syndrome (PRISMS) to educate, raise awareness and support research to improve the lives of those living with this rare condition. As you know, there is a tremendous need to educate doctors, teachers and social service providers about SMS. PRISMS makes a real difference in the lives of so many people.

Over the last few years, the scope of services PRISMS is able to offer to our community has expanded greatly. We work harder every year to try to best serve families and individuals with SMS find a path to thrive in life as opposed to just trying to survive. In 2017, PRISMS raised awareness providing information on SMS to more than 350,000 people around the world, hosted an International SMS Research Symposium, launched an SMS Patient Registry and devoted nearly $200,000 to advance SMS research.

We plan to grow even more in 2018, when we will celebrate 25 years of leadership and a future of hope for the SMS community!

Make a Donation Today!

PRISMS Launches SMS Patient Registry

PRISMS is happy to announce the launching of its first-ever patient registry! The Smith-Magenis Syndrome Patient Registry (SMSPR) is an online tool used to collect, organize and store detailed information about Smith-Magenis Syndrome. This information will be used to help improve the clinical management and quality of life of all individuals with SMS.

WHY DO WE NEED A REGISTRY?

A patient registry will allow researchers to more accurately determine the natural history of Smith-Magenis Syndrome by collecting data (medical, clinical and genetic information) and making that data available to researchers across the globe. Access to this data will lead to a deeper understanding of SMS and improve care pathways. An SMS Registry will accelerate research, which may lead to treatment trials and improved therapeutic options, and will more accurately ascertain the prevalence of SMS.

WHO IS ELIGIBLE TO PARTICIPATE?

All individuals with a confirmed diagnosis of Smith-Magenis Syndrome.

HOW DO I ENROLL?

If you are interested in enrolling, or have questions about this study, please contact the PRISMS registry staff at **prisms.registry@bcm.edu**.

To learn more about the Smith-Magenis Syndrome Patient Registry, please visit the SMS Patient Registry on the PRISMS website.

PRISMS Awards $150,000 for Innovative SMS Research at University of Michigan

PRISMS is honored to announce the award of $150,000 to Dr. Shigeki Iwase and Dr. Michael Sutton at the University of Michigan to support their research into the "Roles of RAI1 in Translating the Histone Methylation Code into Synaptic Plasticity."

Earlier this year, PRISMS opened up an application process to research institutions around the world to apply for funding to support a post-doctoral fellowship for a research project related to SMS at $75,000 per year for two years. PRISMS received several applications, all of which were reviewed and scored by PRISMS Professional Advisory Board, taking into account the significance and innovation of the proposed research, the methodologies, timeline, cost feasibility and the opportunity the proposed research presented for future SMS research. Upon review, the workgroup was delighted to fund Dr. Iwase and Dr. Sutton's innovative project.

RAI1 mutations are responsible for two distinct neurodevelopmental disorders, Smith-Magenis and Potocki-Lupski syndromes (SMS and PTLS). Individuals with SMS and PTLS are characterized by abnormal adaptive behavior, which requires synaptic rewiring by experiences. The molecular and cellular roles of RAI1 in the brain remain unclear, which makes it difficult to design rational therapeutics for these conditions.

With the support of this funding from PRISMS, we aim to uncover what RAI1 does in neuronal cells at molecular and cellular levels. More specifically, we will test our prediction that RAI1 "reads" specific chemical marks that are placed on chromatin, DNA-containing materials, in neurons and by doing so, RAI1 controls gene expression and rewiring of neuronal connectivity. Our hope is to pinpoint molecular and cellular roles of RAI1, thereby generating a solid ground for the future therapeutic design. Since this is an early phase exploratory project, it would be challenging to obtain federal funding. PRISMS fund is highly instructive for our collaborative team to jump start this exciting and important project.

Drs. Iwase and Sutton

This research initiative will involve the co-mentoring of a post-doctoral fellow in the Sutton and Iwase labs, which will enable this individual to become an independent scientist focused on RAI1 biology and related brain disorders.

"PRISMS could not be more proud to have the opportunity to fund this research on behalf of the SMS community," says PRISMS Executive Director Emily Fields. "Investing in this project is an investment in building future researchers with an interest in and commitment to SMS research. Additionally, we are excited about the opportunity this research presents to guide future therapeutic developments for SMS".

i have sms

Poem by Claire Woo, mother of Chloe

I am a child born
Into a loving home.
I have a disorder
Called Smith-Magenis Syndrome.

It is a very rare disorder
And widely unknown.
But mummy and daddy love me.
I'll never be alone.

It is very complex
And also very rare.
Sometimes my behavior
Will cause people to stare.

There are many common features,
Although no two are the same.
Sleepless nights and meltdowns
Drive my mummy insane.

I hit my face hard
And repeatedly bang my head.
I bite my fingers so much,
Often they have bled.

I can't help doing it,
I'm really not being bad,
But I can tell by mummy's face,
It makes her really sad.

I don't sleep at night,
I wake often to play.
My body doesn't understand
The difference from night and day.

Mummy gets worried;
I get into things at night.
I touch things I shouldn't,
I break everything in sight.

I cannot talk to mummy
And tell her what I need.
I try to sign, it's really hard
But I'm determined to succeed.

I want to tell mummy I love her;
That would make her smile.
She tells me every single day;
To say it back would be worthwhile.

My name is Chloe,
I am like any other child.
I'm obsessed with Peppa Pig
And running about going wild.

I am a very caring girl,
Loving, sweet and funny.
Affectionate and kind,
In fact, just like my mummy.

Please do not judge
Those with special needs.
Yes, we are different,
But beautiful indeed.

CALLING ALL SMS BLOGGERS

By Emily Fields, Executive Director

PRISMS is hard at work creating a refreshed website as a service for the entire SMS community. Through this new site, families and professionals will have an opportunity to access the latest resources and information on Smith-Magenis Syndrome, contribute to ongoing initiatives and share their own stories through a new blog, #SMSScoop.

The #SMSScoop will be the official blog for PRISMS. PRISMS staff and friends will share information of interest to the SMS community on topics related to research, behavior management, traveling with SMS, experiencing the International SMS Conference and more.

The best news is that anyone can contribute. Sharing your story will be made easy through the new site. You'll simply submit an interest form, and a member of the PRISMS team will reach out to you to support you in crafting your post.

We are actively searching for blog posts to launch with the new site in early 2018. Interested in sharing a success story of your SMSer? Experience a challenging moment with the school? Have some words of wisdom to share with fellow SMS families? If you feel inspired to connect and share, don't hesitate to reach out.

Blog posts should be no more than 1500 words. And you can feel free to share photos, as well. Contact us at info@prisms.org to share your story.

Super Kids

By Dennis Dillon

Spectrum is proud to recognize the following SMSers for their accomplishments:

Dalton Lee

Dalton Lee, 14, of League City, TX. Dalton and his brother, Xander, 10, earned their black belts in mixed martial arts. Dalton and Xander worked three hours per day, six days per week for eight weeks—including from 5:30 a.m. to 9 p.m. daily in the final week—to achieve this high honor. Dalton has been training in martial arts since he was 5, and he quickly clicked with his instructor. He impressed the adults in his class with his ability to memorize the curriculum and various kata moves. You don't want to mess with Dalton; he can break a board with a simple strike of his palm.

Chris Pucci, 28, of Middle Island, N.Y. For his birthday, Chris traveled 2½ hours by bus with his mom to New York City to see the Broadway production of "Cats." An avid fan of musicals, Chris previously went to the Big Apple and saw "Aladdin" and "The Lion King." According to his mom, Liz Rickards Pucci, Chris "enjoys the shows immensely. That entails lunch, shopping and closing with a Shake Shack milkshake."

Ash Downey, 19, Calgary, Alberta. A 2016 graduate of Olds College, where she took a veterinary technical assistant program, Ash has been volunteering for the Alberta Animal Rescue Crew Society at a shelter that provides care for rescue dogs and cats.

Cedar Rearick, 18, Farmington, ME. When one of his training wheels broke off accidentally, Cedar quickly ditched the second one and started riding his mountain bike on a single-track path in Carrabassett Valley, near the Sugar Loaf ski area. "Off he went, like a champ," says his dad, Dave Rearick. Cedar is a senior at Mt. Blue High School.

If you have an SMSer who deserves recognition for his or her achievement, email ***ddillon923@gmail.com.***

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Parents and Researchers Interested in Smith-Magenis Syndrome

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FIND US ON SOCIAL MEDIA

Host a fundraiser or awareness event in your area

Contact info@prisms.org for support and ideas to make your event a success.

Join the PRISMS community by becoming a member for 2018

Learn more about membership by visiting our website at www.prisms.org.

Make a donation

Support PRISMS' mission to increase awareness, education, and research for the SMS community by visiting our website and making a donation today at www.prisms.org.

Share your story with us

We want to hear from you. Reach out to PRISMS Executive Director, Emily Fields, at efields@prisms.org.

EASY SUPPORT FOR PRISMS

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AmazonSmile is the same Amazon you know—same products, same prices, same service. Follow these simple steps to support PRISMS while you shop!

1. Go to www.smile.amazon.com
2. Type "PRISMS, Inc." in the pick your charitable organization text box. Click Search. (Note: type PRISMS in capital letters.)
3. Select "Prisms, Inc." There are a few organizations with the same name. Choose the PRISMS in Sterling, VA founded in 1993. Click "about" if you want to confirm you have chosen the correct PRISMS.
4. Save smile.amazon.com to your bookmarks or favorites bar to make it easy to return to AmazonSmile. In order for PRISMS to receive a percentage, you must shop through smile.amazon.com.
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6. Enjoy shopping knowing you are supporting PRISMS!

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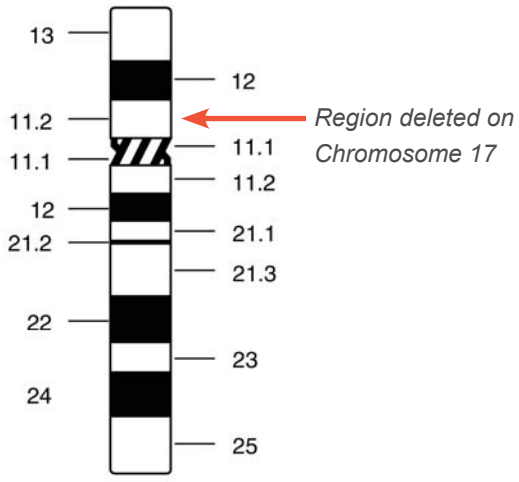
You shop. Amazon gives.

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.



VOLUNTEER OPPORTUNITIES

PRISMS is always looking for helping hands from the community to serve as members of one of our various committees, assist in a program, or coordinate an event. Interested in sharing your time and talents with the SMS community? Contact us at info@prisms.org.

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