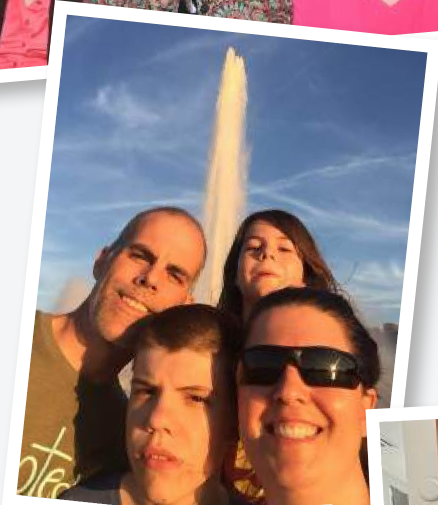
PARENTS AND RESEARCHERS INTERESTED IN SMITH-MAGENIS SYNDROME

WITH YOU
then. now. always.

prisms

ANNUAL REPORT 2023


prisms
I AM SOLVING MY BROOK WAY!
BEAUTY IS STAR




PANTHERS


fireflies
30


prisms


prisms

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PARENTS AND RESEARCHERS INTERESTED IN SMITH-MAGENIS SYNDROME

prisms

MISSION

PRISMS IS DEDICATED to providing information and support to families of persons with Smith-Magenis syndrome (SMS), sponsoring research and fostering partnerships with professionals to increase awareness and understanding of SMS.

VISION

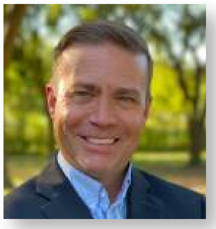
PRISMS IS A LEADER of the Smith-Magenis syndrome community and engages, inspires and supports families, physicians, educators, researchers, and others so they can improve the lives of everyone affected by SMS.

VALUES

- ✓ COMPASSIONATE
- ✓ EMPOWERING
- ✓ CONSCIENTIOUS
- ✓ INCLUSIVE
- ✓ EXCELLENCE

A Message from the President

Brandon Daniel, President



Wow! What a year PRISMS had during its 30-year anniversary. This year PRISMS had its most successful fundraising campaign ever with the Inspire Match campaign. A special thank you goes to our anonymous donor who contributed $100,000 to make this campaign a reality. These funds are vital to continuing PRISMS' mission not only into 2024 but as a solid financial foundation to start the next 30 years.

As I type this message, I have country singer Tim McGraw's song in my head, "My Next Thirty Years." What does PRISMS look like 30 years from now? Indeed, the organization has grown substantially from its early beginnings in 1993. Today, PRISMS has a full Board of committed and caring parents and professionals making decisions for the organization not only for the upcoming 2024 Conference year but for future years. PRISMS has a dedicated and distinguished Professional Advisory Board to guide the scientific and research priorities for the years ahead. PRISMS is led today by our new Executive Director, Michelle Larscheid, who has brought much drive and energy to the organization. PRISMS has a dedicated group of Regional Representatives, regional volunteers, and donors who give their time and finances to allow the organization to grow and provide additional resources to the SMS community. As we start "the next thirty years,"PRISMS is as strong as it has ever been. This is a testament to every donor and volunteer that has ever helped along the way. Thank you!

This annual report shows a culmination of the work PRISMS was able to complete in 2023 for the SMS community. On behalf of the PRISMS Board of Directors, thank you for your ongoing commitment and financial support of PRISMS! I look forward to seeing you all at the 2024 PRISMS Conference "Leading the Way Together" and, as the song says, where we can "spend precious moments with the ones that I hold dear."

Thank you,
Brandon Daniel

30 prisms
1993 | 2023
WITH YOU THEN. NOW. ALWAYS.

prisms | 30th Anniversary Retrospective

1982 Smith-Magenis syndrome was first reported in medical literature by Ann Smith.

1993

On February 4, 1993, Parents and Researchers Interested in Smith-Magenis syndrome (PRISMS) was formed and officially recognized as a nonprofit organization.

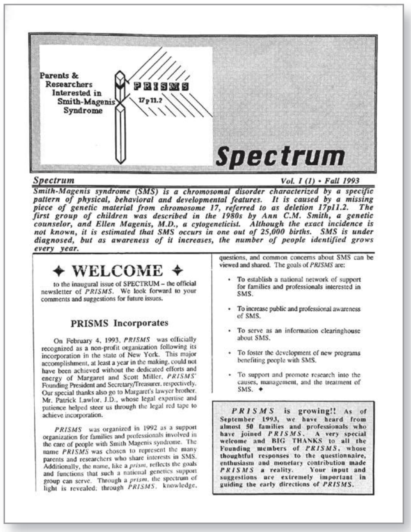
2000
PRISMS Professional Advisory Board was established.

2001
PRISMS was invited to participate with Rick Guidotti, founder of Positive Exposure.

2001
PRISMS Professional Advisory Board authored the first GeneReview publication of SMS.

1980 / **1990** **2000**

1993
PRISMS published its first newsletter, SPECTRUM.



Parents & Researchers Interested in Smith-Magenis Syndrome

PRISMS 17p11.2

Spectrum

Spectrum *Vol. I (1) • Fall 1993*

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 was described in the 1980s by Ann C.M. Smith, a genetic counselor, and Ellen Magenis, M.D., a cytogeneticist. Although the exact incidence is not known, it is estimated that SMS occurs in one out of 25,000 births. SMS is under diagnosed, but as awareness of it increases, the number of people identified grows every year.

✦ WELCOME ✦

to the inaugural issue of SPECTRUM – the official newsletter of *PRISMS*. We look forward to your comments and suggestions for future issues.

PRISMS Incorporates

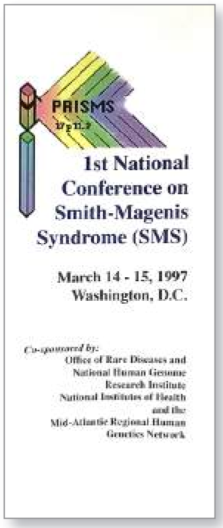
On February 4, 1993, *PRISMS* was officially recognized as a non-profit organization following its incorporation in the state of New York. This major accomplishment, at least a year in the making, could not have been achieved without the dedicated efforts and energy of Margaret and Scott Miller, *PRISMS'* Founding President and Secretary/Treasurer, respectively. Our special thanks also go to Margaret's lawyer brother, Mr. Patrick Lawlor, J.D., whose legal expertise and patience helped steer us through the legal red tape to achieve incorporation.

PRISMS was organized in 1992 as a support organization for families and professionals involved in the care of people with Smith Magenis syndrome. The name *PRISMS* was chosen to represent the many parents and researchers who share interests in SMS. Additionally, the name, like a *prism*, reflects the goals and functions that such a national genetics support group can serve. Through a *prism*, the spectrum of light is revealed; through *PRISMS*, knowledge, questions, and common concerns about SMS can be viewed and shared. The goals of *PRISMS* are:

- To establish a national network of support for families and professionals interested in SMS.
- To increase public and professional awareness of SMS.
- To serve as an information clearinghouse about SMS.
- To foster the development of new programs benefiting people with SMS.
- To support and promote research into the causes, management, and the treatment of SMS. ◆

PRISMS is growing!! As of September 1993, we have heard from almost 50 families and professionals who have joined *PRISMS*. A very special welcome and BIG THANKS to all the Founding members of *PRISMS*, whose thoughtful responses to the questionnaire, enthusiasm and monetary contribution made *PRISMS* a reality. Your input and suggestions are extremely important in guiding the early directions of *PRISMS*.

1997
The first PRISMS conference was organized and held.



PRISMS 17p11.2

1st National Conference on Smith-Magenis Syndrome (SMS)

March 14 - 15, 1997
Washington, D.C.

Co-sponsored by:
Office of Rare Diseases and
National Human Genome
Research Institute
National Institutes of Health
and the
Mid-Atlantic Regional Human
Genetics Network

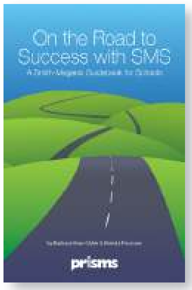
2003
PRISMS produced an SMS video.

2003
A mutation in the RAI1 was identified by Dr. Sarah Elsea and her team of researchers.

2009
PRISMS first edition of the Medical Management Guidelines Published.

2010
The first Research Alliance Meeting (later to become Research Symposium) was held.

2011
PRISMS became an organization member of NORD (National Organization for Rare Disorders).



2014
PRISMS published 1st book dedicated to SMS.

2014
Regional Representative Program launched.

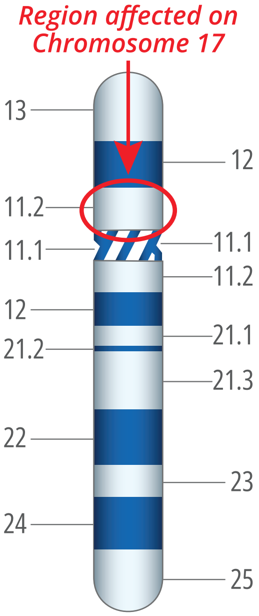
2014
PRISMS partnered with Baylor College of Medicine.

2018
PRISMS partnered with the Brain Donor Project.

2019
Webinar series was established.

2020
PRISMS Clinic and Research Consortium established.

2010 **2020**



2012
PRISMS partnered with Coriell Institute for Medical Research to establish a biobank for SMS.

2013
PRISMS worked with Vanda Pharmaceuticals to recruit patients for an investigational treatment for sleep disturbances in persons with SMS.

2015
PRISMS hired and onboarded its first Executive Director.

2016
PRISMS launched an International Partnership Program.

2017
Patient Registry established.

2017
PRISMS funded a two-year Post Doc position at Michigan University.

2021
PRISMS and Vanda Pharmaceuticals collaborated on a commercial that debuted on SMS Awareness Day 2021.

2023
PRISMS celebrated its 30th Anniversary.

1993 30 2023
prisms
WITH YOU THEN. NOW. ALWAYS.

What Does the Professional Advisory Board Do for PRISMS?

Maggie Miller, Vice President

Our work at PRISMS would not be possible without the expertise of our outstanding Professional Advisory Board, also known as the PAB.

The PAB consists of ten individuals representing the diverse needs of the Smith-Magenis syndrome (SMS) community. It is an internationally recognized group of scientists, educators, and clinicians with expertise in SMS, genetics, molecular research, education, speech, development, and caregiver and sibling concerns. Most PAB members are actively involved in research and/or clinical activities related to Smith-Magenis syndrome. The PRISMS PAB performs many roles that go beyond the extent of most scientific panels. The PAB not only participates in a broad range of SMS research, but also authors and reviews all of the resources that PRISMS shares with the community. The PAB gives stringent review over research funding requests, IRB approved research projects and helps to expand our collaboration with academic and research institutes and industry partners. Their work outshines the limited scope of other such panels. We are truly indebted to the PAB and their dedication to the global SMS community.

The PAB advises the PRISMS Board of Directors on medical, scientific, educational, and therapeutic matters and assists in the education and research activities of the organization.

All of the resources that we publish are reviewed by these experts in SMS to ensure that they are of the highest quality. This is how you know you can trust information from PRISMS.

Some of the key online resources our PAB has contributed to are:

- Smith-Magenis Syndrome GeneReviews
- PRISMS Medical Management Guidelines & Treatment Recommendations
- Our Living with SMS articles
- *On the Road to Success with SMS* book, available for purchase through the PRISMS Store.

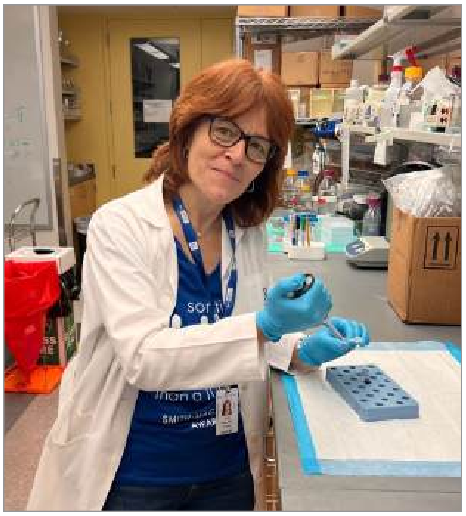
In addition to this, if you've ever asked us a question through the Contact Us form, we probably checked with an advisory board member before we gave you the answer!

The PAB also provides input behind the scenes on PRISMS' programs, developing questionnaires for the SMS Patient Registry, reviewing grant applications, and reviewing potential research opportunities for the SMS and PRISMS community. The PAB looks forward to the 2024 PRISMS Research Symposium this summer where current research related to SMS will be presented and research goals for the community discussed in an open format.

The PRISMS Professional Advisory Board is an incredible asset to our Smith-Magenis syndrome community, and we are so fortunate to have these professionals serving alongside the PRISMS Board of Directors. They are a key component in making PRISMS a leading resource for families and professionals looking for information about SMS. We could not do this work without them!

PRISMS Board Visits Elsea Lab at Baylor College of Medicine

The PRISMS Board and staff met in Houston in October for an in-person planning meeting. They were excited about the opportunity to tour Dr. Elsea's lab and ask questions about her work. Touring the lab and understanding more about the lab operations and how her staff and students work together was a highlight of the meeting.



Smith-Magenis Syndrome Patient Registry

The Smith-Magenis syndrome Patient Registry (SMSPR), first launched in 2017, continues to welcome new participants and evolve as a resource for the SMS community. One of the goals during PRISMS' 30th anniversary year was to shine a light on this research endeavor and, thanks to those efforts, the enrollment rate doubled in 2023 compared to 2022. There are now 132 participants enrolled, with 116 completing at least 1 of the 10 surveys. Participants come from diverse backgrounds from around the world (see SMSPR Participant Map).

This table describes the demographic information for participants in the SMSPR. This map visualizes where the SMSPR participants live.

Two peer-reviewed publications using data from the SMSPR were also

Demographics of the SMSPR (N=116)

	n	%
Sex		
Male	58	50%
Female	58	50%
Diagnosis (n=91)		
17p11.2 Deletion	76	83%
RAI1 Variant	15	17%
Age		
1 - 11 years	41	35%
12 - 17 years	33	29%
18 - 73 years	42	36%
Race		
White	103	89%
Asian	3	2.5%
More than one race	10	8.5%
Ethnicity		
Non-Hispanic	107	92%
Hispanic	9	8%

SMSPR Participant Map

SMSPR participants hail from 10 countries across 5 continents!

North America	104
Europe	5
Australia	3
South Africa	2
Asia	2

submitted in 2023 and are, or will be, available for families to read. Both of these studies focus on various aspects of speech, language, and hearing. More collaborations to share data from the SMSPR are planned.

1. Brennan C, Smith ML, Baiduc RR, O'Connor L. "Speech, Language, Hearing, and Otopathology Results from the International Smith-Magenis Syndrome (SMS) Patient Registry." J Speech Lang Hear Res, 2024 Mar 11;67(3):917-938. doi: 10.1044/2023_JSLHR-23-00179. Epub 2024 Feb 7. PMID: 38324273 (https://www.prisms.org/speech-language-hearing-and-otopathology-results-from-the-international-smith-magenis-syndrome-sms-patient-registry/)
2. Brennan C, Baiduc R. (under review) "Subtle differences with overlapping hearing, language, and communication profiles in individuals with Smith-Magenis Syndrome (SMS) who have a genetic deletion versus the RAI1 variant."

Finally, new measures are being developed to ask questions about lipid metabolism, including cholesterol and liver function tests, as well as Birt-Hogg-Dubé syndrome. The goal is to learn more about these conditions in SMS so that screening and treatment recommendations can improve. Be on the lookout for your invitation to take part in these new surveys through the SMSPR in the months ahead.

If you haven't joined the SMSPR yet, please consider registering today!

https://www.prisms.org/research/sms-patient-registry/

Highlights from PRISMS' 2023 Webinar Series

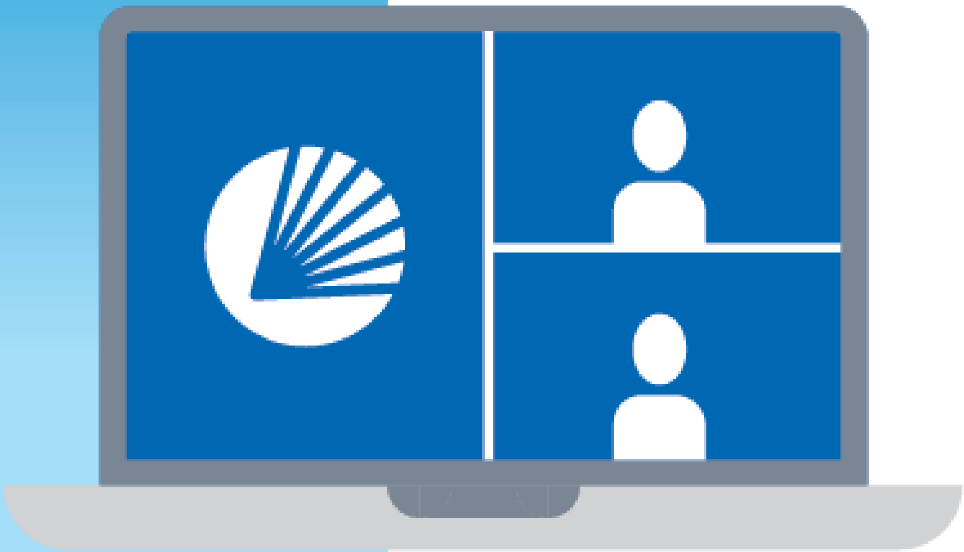
Allison Stephanouk, Education Chair

PRISMS hosted a series of enlightening webinars throughout 2023 aimed at sharing critical information with families affected by SMS.

1. Treatment for Nighttime Sleep Disturbances in SMS

Date Presented: May 2, 2023

PRISMS commenced its 2023 webinar series with a focus on addressing a pervasive challenge faced by individuals with SMS and their families. The webinar, titled, "Treatment for Nighttime Sleep Disturbances in SMS," was presented by Vanda Pharmaceuticals' representatives, Mallery Mayo, PhD, Senior Director of the Medical Affairs Department, Frances Fritz, BSN, RN, Director of Health Education & Advocacy and Samantha Nash, MSW, Senior Manager of Case Management. The webinar provided attendees with valuable insights into this groundbreaking treatment approach.



2. Adult Living—Residential and Vocational Options

Date Presented: May 24, 2023

In the pursuit of fostering independence and empowerment for individuals with SMS as they transition into adulthood, PRISMS presented the webinar “Adult Living—Residential and Vocational Options.” Led by Daniela Morse, M.Ed., founder and Executive Director of the Shared Living Collaborative, attendees were introduced to a myriad of non-traditional housing options, vocational training, employment opportunities, and social resources. The webinar emphasized the importance of person-centered, self-directed planning, highlighting its role in cultivating a desirable quality of life for individuals with complex challenges.

3. A Day in the Life of the SMS Clinics

Date Presented: July 12, 2023

PRISMS continued its commitment to fostering holistic well-being for individuals with SMS through the webinar “A Day in the Life of the SMS Clinics.” Presented by Barbara Haas-Givler, M.Ed, BCBA, and Madeline Williamson, MS, CGC, of Geisinger’s Autism and Developmental Medicine Institute, the webinar provided attendees with an immersive look into the comprehensive and compassionate care provided by SMS clinics. Through real-life insights and experiences, participants gained invaluable understanding of the clinic’s multi-specialty approach to patient care, consultation with treatment specialists, and development of personalized care plans for each patient.

Thank you to all our wonderful 2023 webinar speakers for providing these valuable resources to families affected by SMS. Thank you also to our attendees who always strengthen our webinars with thoughtful questions.

If you missed any of our 2023 webinars, you can find our entire webinar series on PRISMS website under the education tab. Please plan to join us for future PRISMS webinars and subscribe to our YouTube channel: @smsprisms.

Spreading Awareness

Michelle Lee, Awareness Chair

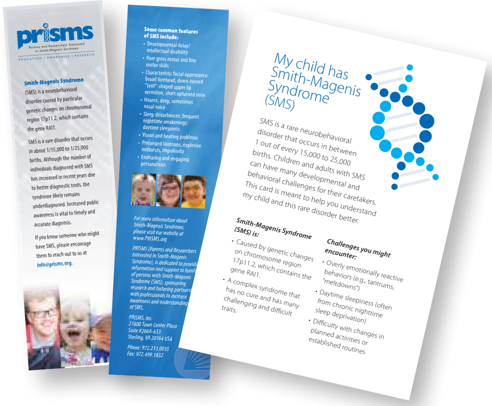
It was another busy year spreading awareness about Smith-Magenis syndrome! We continued to utilize our social media to spread awareness and reach many new SMS families. Our Facebook support page has 3,571 members and is growing weekly. The PRISMS Facebook page now has 2,681 followers and continues to post helpful resources and important information for the PRISMS community. Our Twitter account has more than 667 followers and the PRISMS Instagram account has over 1,386 followers. If you haven't joined, followed, or "liked" PRISMS, please do so! Liking, commenting, and sharing help our pages get seen.

We continued our "Sharing More Smiles" series this year. This campaign features a photo and fun facts about an SMS individual. We shared stories as they came in and are always searching for more smiles to share, and would love to share a story about your SMS star. Send your stories to info@prisms.org.

This year, PRISMS turned 30! We enjoyed celebrating PRISMS' 30th Birthday on social media by sharing pictures of SMS families. The PRISMS board enjoyed a 30th birthday cake during their in-person planning meeting to celebrate. In addition, PRISMS shared 12 Throwback Thursday posts on social media as well as a timeline graphic showing our history and growth. It was great to see PRISMS celebrated all over the country.

Once again, our community came through on Smith-Magenis Awareness Day (November 17th) by sharing on social media. PRISMS provided an Awareness Day calendar with fun events, activities, and posts to share starting November 1st through November 17th. Many people helped raise awareness by sharing photos of them wearing awareness shirts, using our coloring pages, hosting a Facebook fundraiser, and posting daily SMS facts. We had over 16,500 Facebook views, as well as over 5,500 participation events with likes, comments, and shares. Instagram was also utilized to spread awareness with over 8,000 people reached! Your participation in SMS Awareness Day is greatly

SMS community members wearing their SMS Awareness Day shirts.



prisms
Parents and Researchers Interested in Smith-Magenis Syndrome
EDUCATION | AWARENESS | RESEARCH

Smith-Magenis Syndrome (SMS) is a neurobehavioral disorder caused by particular genetic changes on chromosomal region 17p11.2, which contains the gene RAI1.

SMS is a rare disorder that occurs in about 1/15,000 to 1/25,000 births. Although the number of individuals diagnosed with SMS has increased in recent years due to better diagnostic tools, the syndrome likely remains underdiagnosed. Increased public awareness is vital to timely and accurate diagnosis.

If you know someone who might have SMS, please encourage them to reach out to us at info@prisms.org.

Some common features of SMS include:

- Developmental delay/ intellectual disability
- Poor gross motor and fine motor skills
- Characteristic facial appearance: broad forehead, down-turned "tent"-shaped upper lip vermilion, short upturned nose
- Hoarse, deep, sometimes nasal voice
- Sleep disturbances: frequent nighttime awakenings; daytime sleepiness
- Vision and hearing problems
- Prolonged tantrums, explosive outbursts, impulsivity
- Endearing and engaging personalities

For more information about Smith-Magenis Syndrome, please visit our website at www.PRISMS.org

PRISMS (Parents and Researchers Interested In Smith-Magenis Syndrome), is dedicated to providi information and support to famil of persons with Smith-Magenis Syndrome (SMS), sponsoring research and fostering partners with professionals to increase awareness and understanding of SMS.

PRISMS, Inc.
21800 Town Center Plaza
Suite #266A-633
Sterling, VA 20164 USA

Phone: 972.231.0035
Fax: 972.499.1832

My child has Smith-Magenis Syndrome (SMS)

SMS is a rare neurobehavioral disorder that occurs in between 1 out of every 15,000 to 25,000 births. Children and adults with SMS can have many developmental and behavioral challenges for their caretakers. This card is meant to help you understand my child and this rare disorder better.

Smith-Magenis Syndrome (SMS) is:

- Caused by genetic changes on chromosome region 17p11.2, which contains the gene RAI1.
- A complex syndrome that has no cure and has many challenging and difficult traits.

Challenges you might encounter:

- Overly emotionally reactive behaviors (e.g., tantrums, "meltdowns")
- Daytime sleepiness (often from chronic nighttime sleep deprivation)
- Difficulty with changes in planned activities or established routines

appreciated and we love your enthusiasm in spreading awareness.

PRISMS continues to provide awareness tools to the SMS community, including brochures, behavior cards, wristbands, bookmarks, and awareness/compassion cards. The PRISMS online store prismsstore.org continues to sell apparel and other awareness items. You can now purchase the *On the Road to Success with SMS* book through the PRISMS store.

We also continue to work with major national organizations, including the National Organization of Rare Disorders (NORD) and Global Genes, to help celebrate Rare Disease Day and raise awareness of rare disorders.

Thank you for your tireless efforts to help us raise awareness about Smith-Magenis Syndrome. Educating your community of friends, neighbors, family, physicians, educators, and more is exactly how we bring understanding and support to our rare community.

Members of the PRISMS Board and staff proudly wearing their SMS Awareness Day shirts.

Financial Overview

Percy Huston, Treasurer

PRISMS' 2023 financial year was definitely a success, and somewhat of an unexpected one at that. The year began with modest fundraising expectations but, due to a number of factors, ended with a "bang."

First and foremost, some existing PRISMS board members took it upon themselves to get much more involved in our development efforts. Most notably Jackie Fallenstein, Development Chair and Diane Erth, Secretary, spearheaded efforts to improve all levels of fundraising with both enthusiasm and professionalism. Then, in April, an anonymous donor came forward offering a $100,000 match for any new fundraising efforts moving forward through December, which provided a whole new spark for potential donations. Thus, the PRISMS Inspire Match campaign was launched, and the rest was history. Before the year had ended, the campaign smashed its goal, raising over $137,000, in addition to the donor match!

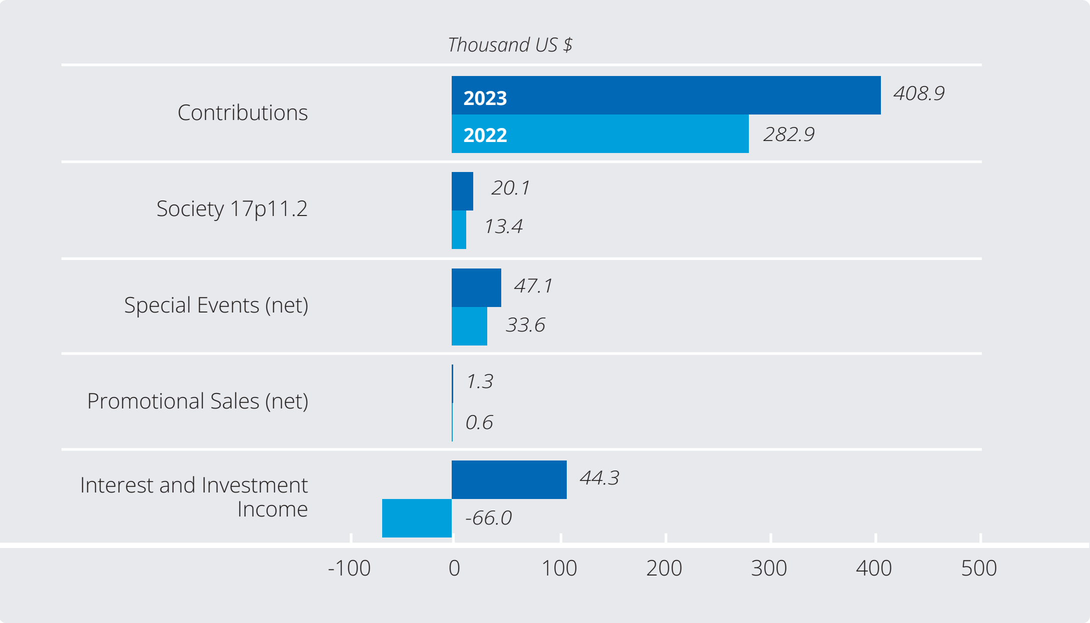
prisms
$100K INSPIRE MATCH

Heading into another conference year, these fundraising efforts were vital in replenishing funds necessary to successfully fund an upcoming conference year. Total assets grew by nearly $100K and net income for the year exceeded $150K. In addition, our investment portfolio rebounded nicely as market conditions improved.

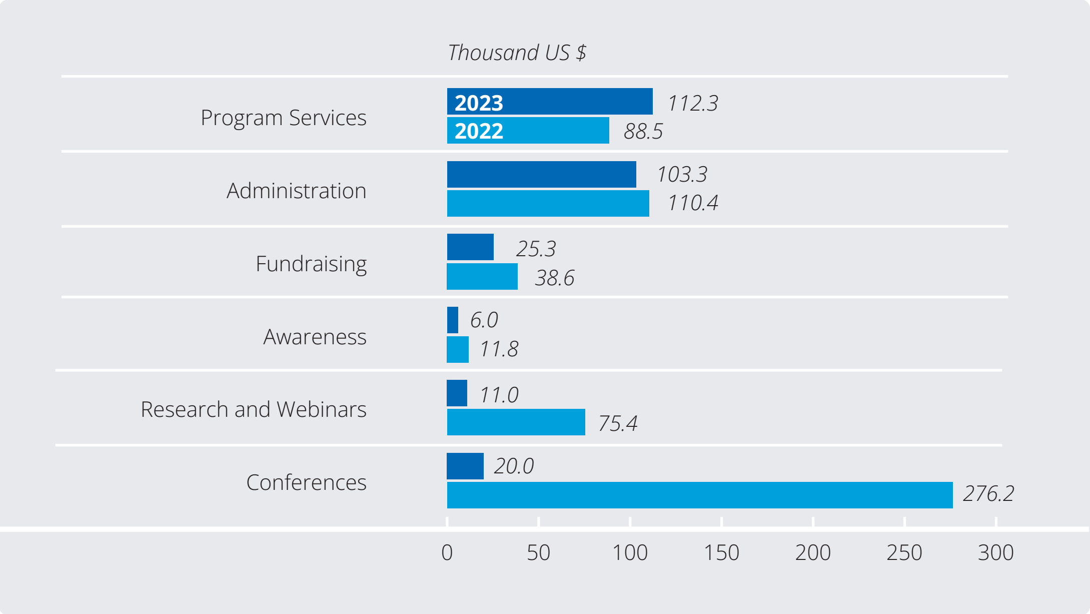
As we turn to 2024, I am excited about another conference year and look forward to reuniting with our PRISMS community at a great venue in Dallas. Please consider coming to Dallas July 11-14, especially if you have never been to a conference. I also want to take this opportunity to say thank you to Brandon Daniel as he steps away from his presidential responsibilities after 5 years in this role. While we are happy he remains on our board, we will miss his leadership and steady hand at the helm. As I follow his footsteps in the presidential role, I know they will be hard ones to fill, but I am excited for the opportunity and the trust our board has shown in supporting me in my new role. As I step away from the treasurer position, I could not be happier to be welcoming Phil Ruedi back to our board who will again serve as our treasurer after a previous 10-year stint in this position. I am truly blessed to be surrounded by dedicated and hard-working board members who work together for the common good of our PRISMS family.

We are so very grateful to all our donors who made this campaign a success but most notably our anonymous donor whose efforts showed how successful this model of fundraising could be. It is their hope that this will "inspire" other PRISMS supporters to come forward to lead future campaigns by offering a large match.

Support and Revenue, 2022-23



Expenses, 2022-23



	2023	2022
Ending Cash and Investments	**$652,218**	**$569,292**
Ending Total Assets	**$689,985**	**$594,963**

One of the ways you can help make sure PRISMS is around for the next 30 years is by making a planned gift by naming PRISMS in your will or life insurance.

All you need to do is give this information to your attorney: Our legal name **PRISMS, Inc.**, and common name for reference **PRISMS**, our **EIN: 54-1652029**, and our mailing address: **PRISMS, Inc., 205 Van Buren Street, Suite 120 #1027, Herndon, VA 20170**.

Fundraising Update

Jackie Fallenstein, Development Chair

PRISMS' many programs and resources are only possible due to the generosity of our donors and sponsors. Here are some of this year's fundraising initiatives:

The Inspire Match

On May 17, PRISMS launched the $100,000 Inspire Match campaign. This dollar-for-dollar Match was offered to PRISMS by a very generous community member who has seen firsthand the impact of PRISMS in the life of their loved one with SMS and the family that supports that individual. The Match was offered through December 1, and with some incredible efforts from our SMS community, we were able to reach our goal by the deadline. In total, the Inspire Match lead to $237,000 in giving to support PRISMS. Thank you to all who participated!

PRISMS 17p11.2 Society

Recognizes donors committing a minimum of $17 in monthly giving. This ongoing support helps PRISMS sustain income throughout the year. Society donations grew 10% in 2023, and efforts will continue to increase in 2024. To learn more or to join the 17p11.2 Society, go to https://www.prisms.org/get-involved/17p11-2-society/.

Double the Donation

Thousands of companies match employee charitable contributions at 1:1 or greater. Millions of dollars are available through these matches, but often employees aren't aware of the match or how to request it. PRISMS has partnered with Double the Donation, an organization that helps donors identify and submit their company matches, streamlining the processing and increasing donations. On PRISMS' donation page, donors can click the tool to learn more about matching donations, https://www.prisms.org/donate/employer-match/

CAF Canada

This organization supports our Canadian donors by allowing them to receive a charitable tax receipt for their donations to PRISMS. Canadian donors can go to https://www.cafcanada.ca/ and select PRISMS from the menu to make a tax-deductible donation.

A sincere thank you to all our donors and sponsors. Your contributions are essential for PRISMS to continue its work supporting all those impacted by SMS!

17p11.2 Society Members

Danielle Bier
Kristine & Glen Braden
Kristine Braden
Lauren Carney
Sharon Cook
Kevin Daly
Barclay Daranyi
Patty Davis
Dennis Dillon
John Doherty
Angela Eaton
Sarah Elsea
Diane Erth
Jackie Fallenstein
Sylvia Farber
Manuel Faria
Cheryl Huber
Percy Huston
Paula Jump
Bonnie Krautheimer
Michelle Larscheid
Josephine Lawlor
Allison & Mike Leatzow
Etienne Lecompte
Michelle Lee
Patty Loyer
Lisa & Michael Mariano
Tina & Charlie McGrevy
Jason Michaud
Alicia & Scott Miller
Scott Miller
Trinity & Mike Miodunski
Karin Monahan
Kevin O'Connor
Amy Pereira
Denien Rasmussen
Gail & David Reiner
Cherisse Rodriguez
Jeremy Rude
Eric Schaller
Caitlin Seldon
Allison Stephanouk
Steven Tanenbaum
Todd Tanenbaum
Stephanie & Mark Tonsoni
Osman Umarji
Derek Waldeck
Debi Waters
Mary Ann Zimmer

PCRC

PRISMS Clinic and Research Consortium

PRISMS Clinical Research Consortium (PCRC) Update

Maggie Miller, Vice President

PRISMS Clinical Research Consortium (PCRC) continues to serve the SMS community throughout the United States and has become a vital extension of the resources that PRISMS provides for families. Clinics within the PCRC provide multi-specialty, comprehensive and compassionate care for patients with SMS. The clinics provide families an opportunity to receive medical and clinical care and/or treatment recommendations that they can carry home to their local providers of care that address the challenges and health concerns associated with Smith-Magenis Syndrome. The clinics' approach to care includes consultation with other treatment specialists, as needed, while developing a plan of care for each patient that strives for health and well-being.

The PCRC is not only responsible for providing a continuum of care across each clinic site, but also responsible for the development of resources that can be shared not only with clinic patients, but is available on the PRISMS website resources. The experience of the clinic providers gives key insights into the challenges of SMS patients. At their regular meetings, the PCRC can discuss and share insights about patient scenarios and learn from their peers. Members can brainstorm and share possible treatment plans and unravel questions or concerns that come up during clinic visits. The PCRC also invites guest speakers to present during these meetings and expand the circle of expertise and interested clinicians. Laleh Ardeshirpour, MD, FAAP, Pediatric Endocrinologist at Connecticut Children's Hospital, was a recent guest lecturer at the PCRC meeting.

PRISMS is working to establish additional clinics throughout the U.S. Families can assist with this effort by informing PRISMS of any doctor, especially those affiliated with a hospital, who has demonstrated a consistent interest in SMS. PRISMS' staff and advisors will then follow-up with the doctor to determine if they might be a good applicant to the PCRC.

The PRISMS SMS Clinic Travel Fund Reimbursement Program supports families who need to travel to one of the clinics within the PCRC. This fund will help defray costs for travel (airline or mileage), lodging, and other miscellaneous costs attributed to traveling to a clinic. This reimbursement is available to a family only one time. If you are planning a clinic visit and need financial assistance, please email info@prisms.org for an application.

Prisms Expands Its Global Partnerships

In 2023, PRISMS continued to expand its global partnerships with other advocacy and research organizations. One of the most significant driving forces behind progress in rare disease research is the formation of strong and collaborative partnerships.

Collaborative partnerships allow stakeholders from various fields—including researchers, clinicians, pharmaceutical companies, patient advocacy groups, and government agencies—to pool their resources and expertise. At PRISMS, we recognize the importance of partnerships in supporting our SMS community and have strived to build these partnerships during our 30 years of service to the global SMS community.

COMBINEDBrain & Biorepository

In 2022, PRISMS became a member of COMBINEDBrain.

The Consortium for Outcome Measures and Biomarkers for Neurodevelopmental Disorders is a non-profit organization devoted to speeding the path to clinical treatments for people with rare, genetic neurological disorders by pooling efforts, studies, and data.

As a member of COMBINEDBrain, we join forces with other rare, neurodevelopmental disorder organizations to advance research by engaging with pharmaceutical companies, research institutes and industry representatives. When we collaborate with other neurodevelopmental disorders, we accelerate research by widening the scope of new researchers and unlock undiscovered data. It is imperative that SMS is not studied in a vacuum or only in isolation. Valuable research requires many tools (basic science, clinical and longitudinal data) and research is better served when studied in a consortium and in collaboration with other disorders.

In 2023, PRISMS recruited SMS participants to contribute to COMBINEDBrain Biorepository. The biorepository is a bank where human samples of blood, urine and tissue can be stored and safely managed. Think of this as similar to the SMS Patient Registry (SMSPR) that PRISMS has managed since 2017; but the biorepository collects these particular samples and keeps them in a library of sorts. Participants' parents/guardians must go through a review and consent process to participate in the biorepository and COMBINEDBrain set-up home collections to retrieve the samples. The process has been efficient and we appreciate all of the families that participated. Interested researchers that would like to request samples from the biorepository must have an approved IRB (Institutional Review Board). These samples can be used to identify biomarkers, understand the process of the disorder (examining the disorder at different ages) and provide a pathway to possible treatments and interventions. PRISMS' participation in the biorepository is a huge step in advancing research which allows for new research opportunities for Smith-Magenis syndrome.

The Brain Donor Project

In 2018, PRISMS partnered with The Brain Donor Project to raise awareness about the critical need for this type of donation to research neurological disorders and brain disease. While this is a very difficult topic to consider and discuss, it is important that brain donation be included as a part of the research of Smith-Magenis Syndrome.

The Brain Donor Project is working with PRISMS to further the science of brain disease. "BDP" works as a conduit for potential brain donors by raising awareness of the urgent need for this precious resource and by simplifying the process to donate. The Brain Donor Project was developed exclusively to support the NeuroBioBank of the National Institutes of Health (NIH) in making available high-quality, well-characterized donated post-mortem brain tissue to neuroscientists.

Since 2018, the Brain Donor Project has been widely recognized as an advocate partner with many organizations like PRISMS and is dedicated to the advancement of research in neurodevelopmental disorders. The Brain Donor Project has made the process as easy as possible and all arrangements can be made years in advance. PRISMS recognizes that it can be distressing to even consider such a donation but PRISMS is dedicated to sponsoring all research and the research landscape should include this type of donation. For more information: https://www.prisms.org/research/active-research/brain-donor-project/

PatientWorthy

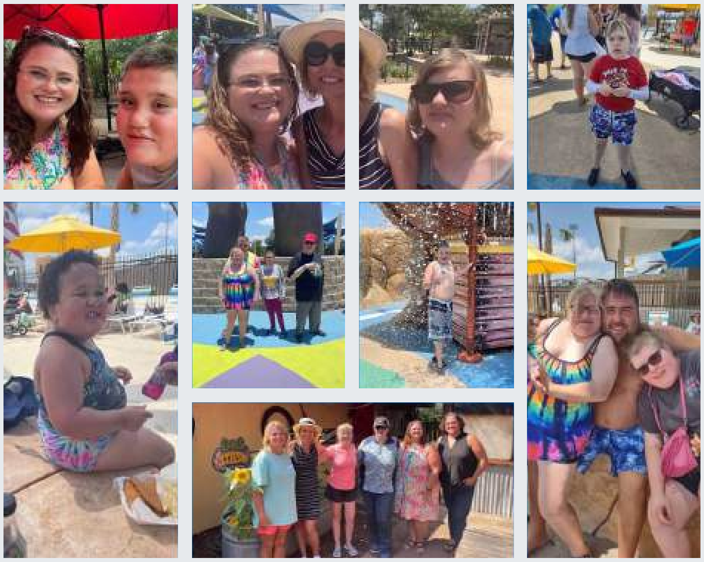
In October of 2023, PRISMS became an advocacy partner with PatientWorthy. PatientWorthy's mission is to amplify the voices of rare disease families and the advocacy groups which serve them. PatientWorthy has published more than 20,000 articles on rare disease and shared them across all the primary social media platforms. This includes sharing patient stories, publicizing new developments in treatments, genetic testing or other advances, and advertising your webinars, patient meetings, and scientific conferences. They reach over 50,000 people every day on their website and more that 2.2 million every week on social media. Approximately 42% of their readers are from the global community as rare disease knows no boundaries. PatientWorthy has over a hundred patient advocacy groups who join as partners in teaching and reaching other families, researchers, physicians and industry.

PRISMS reached out to Kathy Devanny at PatientWorthy to inquire about partnering with them so they could help amplify our voice and help raise awareness of Smith-Magenis Syndrome. PatientWorthy partnered with us for SMS Awareness Day and increased our social media presence in volumes and shared our website and resources with new families and professionals. The impact of their work has increased PRISMS outreach and has expanded our circle of advocates. We look forward to more endeavors with PatientWorthy!

Rare Revolution

PRISMS became a charity partner with RARE REVOLUTION in October of 2023. RARE REVOLUTION is an international not-for-profit media company specializing in rare disease content. RARE REVOLUTION has a digital platform that allows rare disease organizations to share information via online content, social media, video, and online as RARE REVOLUTION Magazine. PRISMS' partnership with RARE REVOLUTION allows us to expand our reach across the globe and share valuable resources about SMS to every corner of our community. In November of 2023, RARE REVOLUTION Magazine featured a story from one of our PRISMS families (https://rarerevolutionmagazine.com/shining-a-light-on-smith-magenis-syndrome/) who wrote about their sweet daughter, Kaydence, who has SMS. The Beecher family shared their journey to diagnosis, their challenges and the wonderful moments and people they have encountered along the way. Their story was filled with honesty and hope and we were delighted that RARE REVOLUTION chose to spotlight SMS. We know this collaboration will lead to more opportunities to raise awareness of SMS and increase PRISMS' presence as a leader in the SMS community.

Family Meet-Ups



Texas

Cecilia and Gordy Poole hosted their annual meet-up at Morgan's Wonderland/Inspiration Island in June in San Antonio, TX. It was a great turnout with 11 SMS families attending (a total of 46 people). That was the biggest turnout they have had since they started organizing the event.

The weather was a perfect 93 degrees and families enjoyed getting wet at one of the four colorful splash pads. Afterward, several of the families headed to Two Brothers BBQ and enjoyed the outdoor live entertainment, playscape and shaded picnic tables. Karen and Josh Dixon came from Iowa to join in the fun. Josh touched and inspired the group when he read a special poem. Special thanks to Brenda Dickerson for snapping most of our photos and capturing the fun memories.

Minnesota

PRISMS Board member, Jackie Fallenstein, organized a meet-up for Minnesota SMS parents in July and welcomed guest speaker Ann Smith, MA, D.Sc. (Hon), CGC PAB Chair Emeritus. The 11 families in attendance enjoyed refreshments, conversation, a presentation by Dr. Smith on "Smith-Magenis Syndrome - a 40-Year Journey Since Discovery" and an informative Q&A session.

Northern California

Laurie Bellet hosted a Northern California Family SMS Picnic in Walnut Creek, California on Sunday, September 3rd. There were 35 people who gathered to enjoy food, fellowship and the playground. Even Merlin the therapy dog enjoyed the afternoon! The group was also spontaneously joined by two families from the playground—a nurse and a special education teacher. A big thanks to Laurie and all the families in attendance!

Southern California

PRISMS Board member and California Regional Representative, Diane Erth, organized a meet-up for many Southern California families. They had a beautiful day for a picnic by the water and eight individuals with SMS were in attendance. The group also donated $225 towards the Inspire Match campaign.

PRISMS is able to provide some administrative and financial support for meet-ups. If you are interested in organizing a meet-up in your state, please reach out to info@prisms.org or contact your Regional Representative.

Fundraisers

We had many members of our SMS Community host a variety of fundraisers this year. Here are some examples:

- **Bernadette and Percy Huston** helped organize a fundraiser for PRISMS during the 5K Wine Run at Chaumette Winery. [1]
- **Adrian and Tim Hibbs** hosted a coffee fundraiser in honor of their son, Anderson. [2]
- The **Josh Sewell family** hosted a fundraiser at a brewery as part of an SMS Awareness Day Fundraiser. [3]
- **Kay Chappel** created a t-shirt fundraiser in honor of her grandson.
- PRISMS Board member **Diane Erth, and her husband Joe and son Max,** organized a lemonade stand fundraiser in their Southern California neighborhood. [4]
- **Alana and Anthony DeLeo** organized a lemonade and cookie fundraiser in honor of their niece, Angelina who has SMS. [5]
- The **Jean Bishop family** hosted the 12th Annual Nickels for Nico cornhole tournament. [6]
- **Tabith Dostal's family** hosted a BBQ fundraiser in Hastings, MN. [7]
- **Sylvia Farber and her family** hosted another successful annual fundraiser, Pickleball for PRISMS. [8,9]

Many of our SMS Community members ran social media fundraisers.

Are you interested in running a fundraiser to support PRISMS and the SMS community? Click here https://www.prisms.org/get-involved/fundraising/ and fill out the form. We would be happy to help you host a successful event.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Easy Peasy
Lemonade Stand Fundraiser
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Parents and Researchers Interested

4


Lemonade Stand Fundraiser
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Parents and Researchers Interested in Smith-Magenis Syndrome
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5


Smith-Magenis Syndrome
WICKER FROG
HUG GGED

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HUG or be HUGGED
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IF YOU'RE LOOKING FOR A SOFT

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PRISMS Regional Representative Program

Denien Rasmussen, Regional Representative Chair

PRISMS Regional Representative program is continually growing and evolving. Our Regional Representatives are PRISMS community members who volunteer their time and care deeply about our community. They want to assist others who may have similar needs or issues, or just a safe space to vent to people who get our lives.

We currently have 64 Regional Representative volunteers in our program. The Regional Representatives cover the United States, Canada, Australia, France, Brazil, Italy, Mexico, Puerto Rico, Russia, and the United Kingdom.

PRISMS continues to have our quarterly Zoom meetings where we discuss many things, some of which are:

- Newly diagnosed families and how to support them
- Hetliotz medication for sleep
- Topics for the upcoming 2024 PRISMS conferences
- New SMS Clinics
- Social media
- Resources and state benefits- how to find them in your region
- Awareness material
- Hosting fundraisers
- Ideas for regional get-togethers
- Upcoming webinar topics

The quarterly Zoom meetings have been incredible and we appreciate the participation in the calls. They have also been highly informative to PRISMS by providing an on-the-ground regional view of what families are dealing with and looking for from PRISMS.

Regional reps are there for you! They have been at the beginning of your journey, through your journey, and will be for the remainder of your journey. #ThenNowAlways.

Our Regional Reps are here to:

- Welcome new individuals and families into the PRISMS Community
- Help families navigate through the PRISMS website for specific information
- Guide families to local resources and state benefits
- Encourage families to join the PRISMS educational webinars
- Encourage families to join our PRISMS community
- Encourage families to attend our conferences and meet-ups

I am so proud to work with this amazing group of people and look forward to every opportunity I have in either speaking with them through Zoom or meeting them in-person. To learn who your Regional Representative is, visit https://www.prisms.org/get-involved/get-connected/. If you have any questions about this program, please contact us at info@prisms.org.

2023 Regional Representatives

Alejandro & Delma Aguilar
Leah Baigell
Eliane Barros
Cally Bauman
Mary Beall
Tracie Belcher
Abigail Bell
Laurie Bellet
Jean Bishop
Sabrin Bisiani
Heather Boney
Debbie Brooks
Maria Elena Carrancedo
Ashton Cheramie
Jennifer Comford
Kevin Daly
Barclay Daranyi
Ana Delgadillo
Amanda Downey
Diane Erth
Maria Feagin
Rhonda Franklin
Heidi Graf
Trevor Gritman
Melissa Haley
Stephanie Hanquez
Mary Hards
Sheila Hernandez Vale
Roberta Higgins
Eric & Kim Hoffman
Bernadette Huston
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Carissa Le
Allison Leatzow
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Rhonda Lowney
Patty Loyer
Callihan Marshall
Maureen Monroe
Kevin O'Connor
Mick Pearson
Denien Rasmussen
Remi Robbins
Marni Rolston
John Roseborough
Brianna Ryczek
Natasha Schaller
Caitlin Seldon
Vinginie Sergent
Mark and Theresa Smyth
Randi Tannenbaum
Bela & Alexander Tzetlin
Osman Umarji
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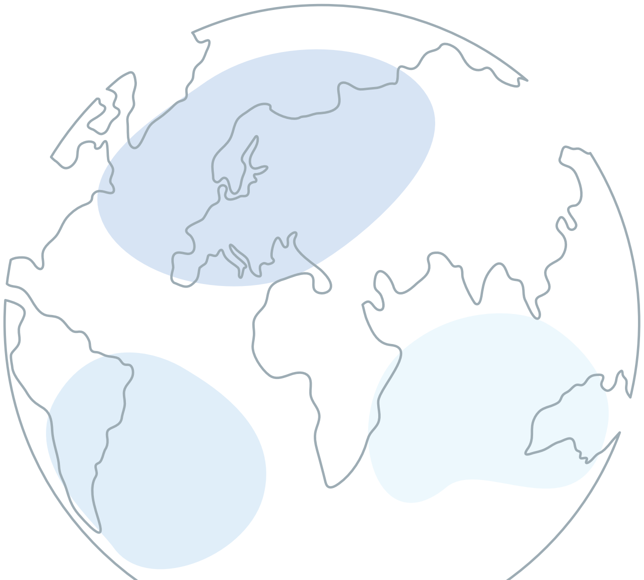
Brandi Wilson
Kim Wirth
Jill Wood
Bill Yates
Michele Zdanowski
Annetta Zidzik



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PARENTS AND RESEARCHERS INTERESTED IN SMITH-MAGENIS SYNDROME

Volunteers

Pat Boschetto

Karen Dixon

Tabitha Dostal

Sylvia Farber

Scott Miller

Kerrie Slattery

Advocacy Workgroup

Leah Baigell

Barclay Daranyi

Melissa Haley

John Mayer

Conference Committee

Kayla Beecher

Pat Brown

Athenna Harrison

Lynda Kilian

Michelle Larscheid

Michelle Lee

Jason Michaud

Maggie Miller

Amy Pereira

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Maggie Miller, Vice President

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Lynda Kilian, Office Manager
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Many photos throughout this annual report are courtesy of ***Rick Guidotti, Positive Exposure***

Designer: ***Mark Hernandez***

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What Is Smith-Magenis Syndrome?

Smith-Magenis Syndrome (SMS) is a chromosomal disorder characterized by a specific pattern of physical, behavioral and developmental features. It is most commonly 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.

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