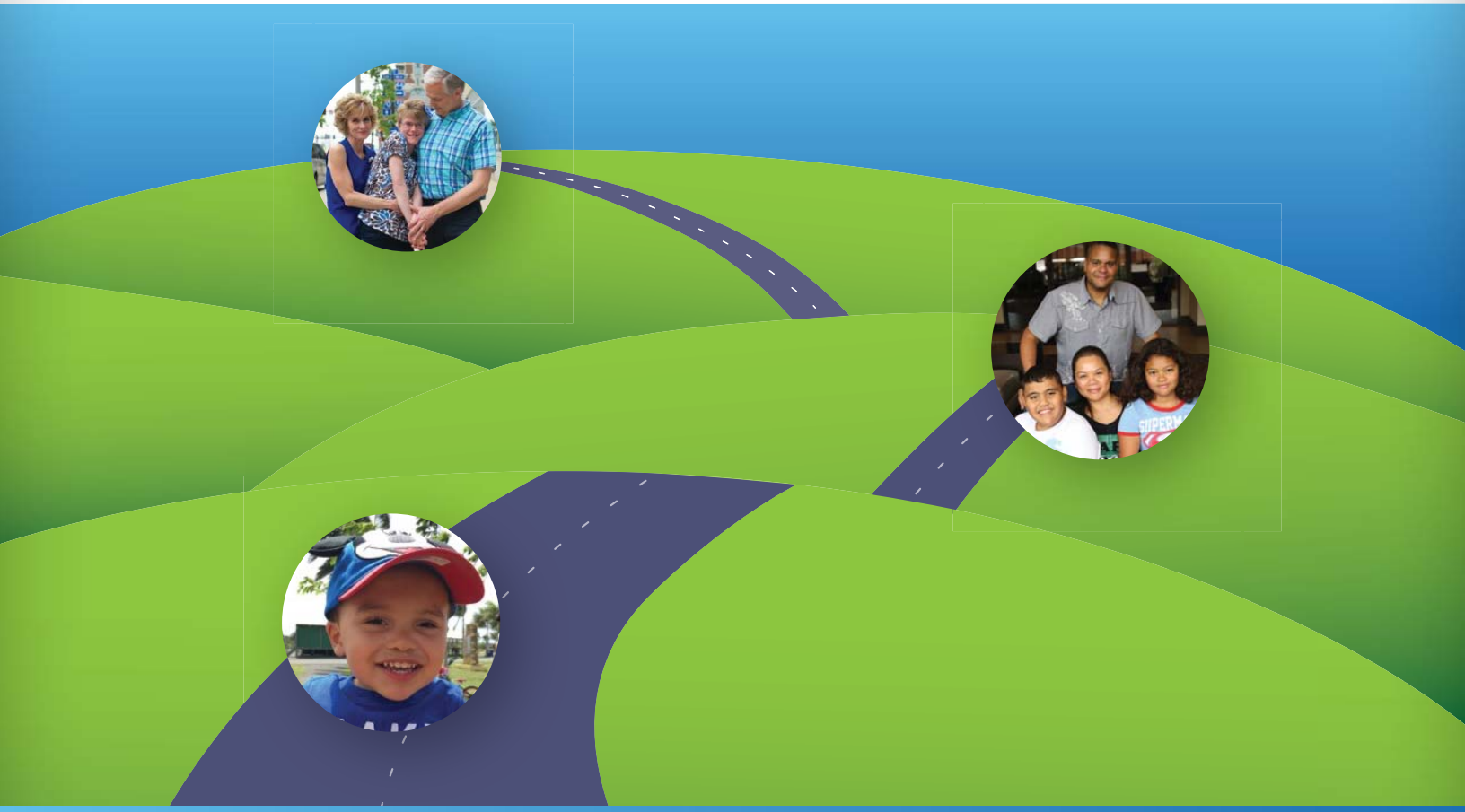
A Guidebook for Families Newly Diagnosed With Smith–Magenis Syndrome



prisms

Parents and Researchers Interested in Smith-Magenis Syndrome

A Guidebook for Families Newly Diagnosed With Smith–Magenis Syndrome

Purpose

This guide has been organized to assist families gather, understand and prioritize information they will need after receiving a diagnosis of Smith-Magenis Syndrome (SMS). This knowledge can empower parents and caregivers in the ongoing support of their child with SMS and develop the skills they will need to become confident and effective advocates.

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

With Heartfelt Appreciation

Funding for the production of this guidebook is made possible through the generous gift of John and Debora Mayer, the parents of Charley Mayer. On behalf of the entire community of families, we are grateful for your support!

Table of Contents

Welcome Letter

Dear Families,

Welcome to PRISMS, Inc.! Upon receiving the news that your child has been diagnosed with Smith-Magenis Syndrome (SMS), you may be feeling overwhelmed and even physically and emotionally drained. The good news is that you are not alone and you have found a family of support in PRISMS. Many families find PRISMS to be their lifeline with a wealth of information, resources and connection. Nothing compares to the feeling of knowing that there are many other families around the world who have gone through what you are experiencing.

PRISMS is the acronym for Parents and Researchers Interested in Smith-Magenis Syndrome. It is a non-profit organization founded in 1993 by the co-discoverer of SMS, an SMS researcher and the parent of a child with SMS. PRISMS can connect you with others, or provide you with many resources via our website at www.prisms.org. You will find updated information including the latest research, publications, past issues of our Spectrum Journal, access to PRISMS Shop, PRISMS blog, the Regional Representative program and more. I encourage you to take some time to navigate through the website and identify the programs and services you find most helpful for you and your family.

We recommend that you sign up to be a member of PRISMS via the website. Membership is free. It is important that you fill out the form completely. As a new member, you will be added to our mailing list, receive our latest Spectrum Journals, access current information on education and awareness, and learn about research opportunities available to members of the SMS community. PRISMS has launched a new SMS Patient Registry, and we encourage you to enroll. The goal of the registry is to pull together information to help families learn more about therapies and treatments. With collective information gleaned from our registry, PRISMS will be more effective in advocating for the needs of those with SMS and their families. Be assured that we do not share your personal information with anyone without your explicit permission.

This Newly Diagnosed Guidebook contains information gathered from the experiences of families who have children with Smith-Magenis Syndrome and professionals. In it, you will find an array of perspectives meant to encourage, support and provide suggestions for you now that you have received the diagnosis. Every day there are new educational, technological and scientific advances that improve the lives of people with SMS. You have every reason to feel optimistic about your child's future.

We hope that being connected to other families through PRISMS will help you to find your way. I know this may be a difficult time for you and your family, but know that PRISMS is here for you. I want to welcome you personally to PRISMS and hope to meet you at one of our conferences or regional meet-ups. Please do not hesitate to call me at 972-231-0035 or email me at efields@prisms.org. I would love to hear from you.

Warm regards,

Emily

Emily Fields, Executive Director
PRISMS, Inc.

A NOTE FROM THE WORKGROUP

The goal of this Newly Diagnosed Guidebook is to assist families in gathering the critical information they need upon first receiving a diagnosis of Smith-Magenis Syndrome (SMS). While the target audience is parents, the information provided is also relevant to siblings, relatives, guardians, caregivers, support staff, professionals and all other interested parties.

This guidebook was conceived and written by parents and professionals, all of whom brought their personal voice, style and perspective to the text. We offer wisdom and advice based on our experiences. Our intent is to provide you with useful and practical information.

We believe that given the proper supports, there are unlimited possibilities for someone with SMS. Capacity and resources vary greatly across the country and around the world. Each person, family and living situation is different. Each child is unique. A wide degree of variation exists in how the characteristics of SMS affect each person. Additionally, education and environmental factors play a significant role in a child's development. Making choices that best meet the needs of the individual with SMS involves personal decisions for each family to determine.

PRISMS does not endorse any particular model or process for living life after the diagnosis. We do not make judgments about the choices that each family makes. We find that within PRISMS, the community and network we share is invaluable. This guide is a "living resource" that will be updated as families provide us with new information. We provide this guide with the hope that each reader finds something of value that can help when you receive the diagnosis of Smith-Magenis Syndrome.

The Workgroup — Leah Baigell, Barclay Daranyi, Melissa Haley and John Mayer

What Is Smith–Magenis Syndrome?

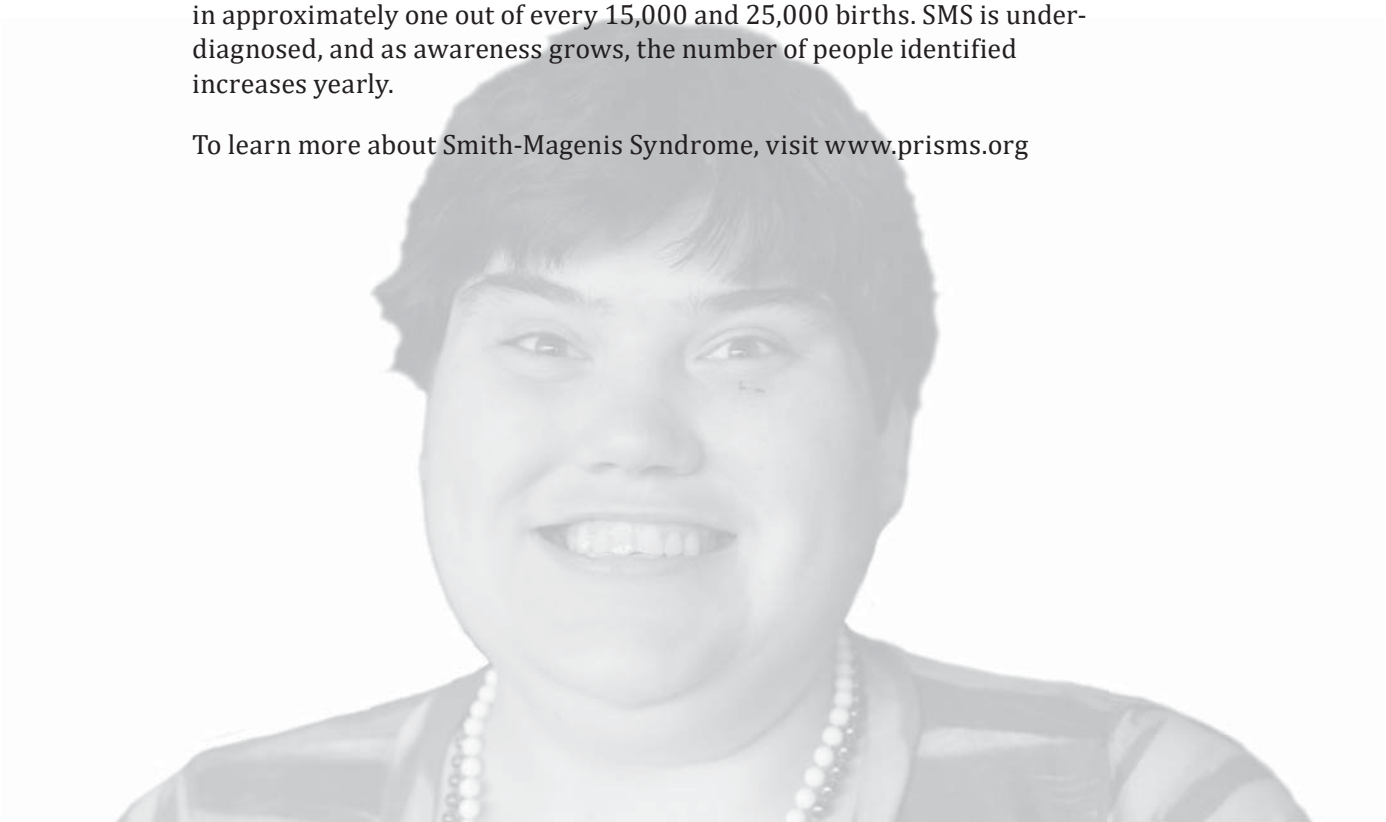
Smith-Magenis Syndrome (SMS) is a chromosomal disorder characterized by a recognizable pattern of physical, behavioral and developmental features.

Common characteristics include variable levels of intellectual disability, developmental delay, early speech/language delay, distinctive facial features, low muscle tone, middle ear problems, skeletal anomalies and decreased sensitivity to pain. The syndrome includes a distinct pattern of neurobehavioral features characterized by chronic sleep disturbances, arm hugging/hand squeezing, hyperactivity and attention problems, prolonged tantrums, sudden mood changes and/or explosive outbursts and self-injurious behaviors.

Despite these challenges, people with SMS respond well to educational, behavioral and environmental supports designed to meet their needs. Many individuals with SMS have engaging and endearing personalities, along with a well-developed sense of humor. They generally appreciate attention and respond well to positive reinforcement. Adults with SMS have been described as eager to please, communicative and affectionate. Most adapt readily to structure and routine.

Although the exact incidence is not known, it is estimated that SMS occurs in approximately one out of every 15,000 and 25,000 births. SMS is under-diagnosed, and as awareness grows, the number of people identified increases yearly.

To learn more about Smith-Magenis Syndrome, visit www.prisms.org



How Will I Deal With This Diagnosis?

Below are two vignettes—one from a family who received a diagnosis of SMS at 15 months, and one from a family who received a diagnosis at 15-years-old. While the immediate reaction to getting the diagnosis was very different, both families found acceptance and answers, and had to learn to live with their new "normal."

JOEY

When Joey was diagnosed with SMS at 15 months old, it felt like my world came crashing down. My precious little baby would now have life-long struggles and my dreams for him were shattered. I worried. Will he walk? Will he talk? Will he be able to read? Will he have friends? Will he be happy? Will he be okay? Will I be okay?

My husband and I spent months visiting doctors, meeting with specialists, scheduling therapies and talking with the school district. It was hard. It was overwhelming. I am not going to lie. I cried…a lot.

In his short nine years, I have watched Joey work extra hard to achieve milestones that come so naturally to typical kids. He has now eliminated all my fears of him not walking or talking. After the 100th time of him asking if he has school in the morning, I often reflect of the days when all he could do was sign the word for "cracker." He has worked so hard and he has come so far. I could not have imagined that when we first learned about his diagnosis.

I, too, have worked very hard to accept that this is our "normal." I have come to accept over the years that I cannot change Joey's diagnosis. Finding joy in the little things and gratitude for the support we have has helped me live each day as it comes. I will never understand "Why Joey?"

I recently heard a quote that has stuck with me. "You won't know until you're there that you're okay." Well, I am there. SMS is hard and there will still be days when I feel inadequate, damaged and defeated. I will have those sleepless nights where my fears play hopscotch, frantically jumping from worry to panic and back to normal again. Then, I know I will have those moments with Joey—those magnificent perfect occasions that I once thought were impossible. Those precious moments offer me hope and possibilities, when I can say to myself, "I am here and I'm okay."

Amy Pereira

ZACH

Zach was diagnosed when he was 15-years-old. In one moment, with one phrase, all the years of work and energy we spent fighting for his rights were validated. For us, it was the biggest sigh "heard round the world." We now had answers and information. We were relieved, not shocked; we were not necessarily happy, but we were gratified to have an explanation.

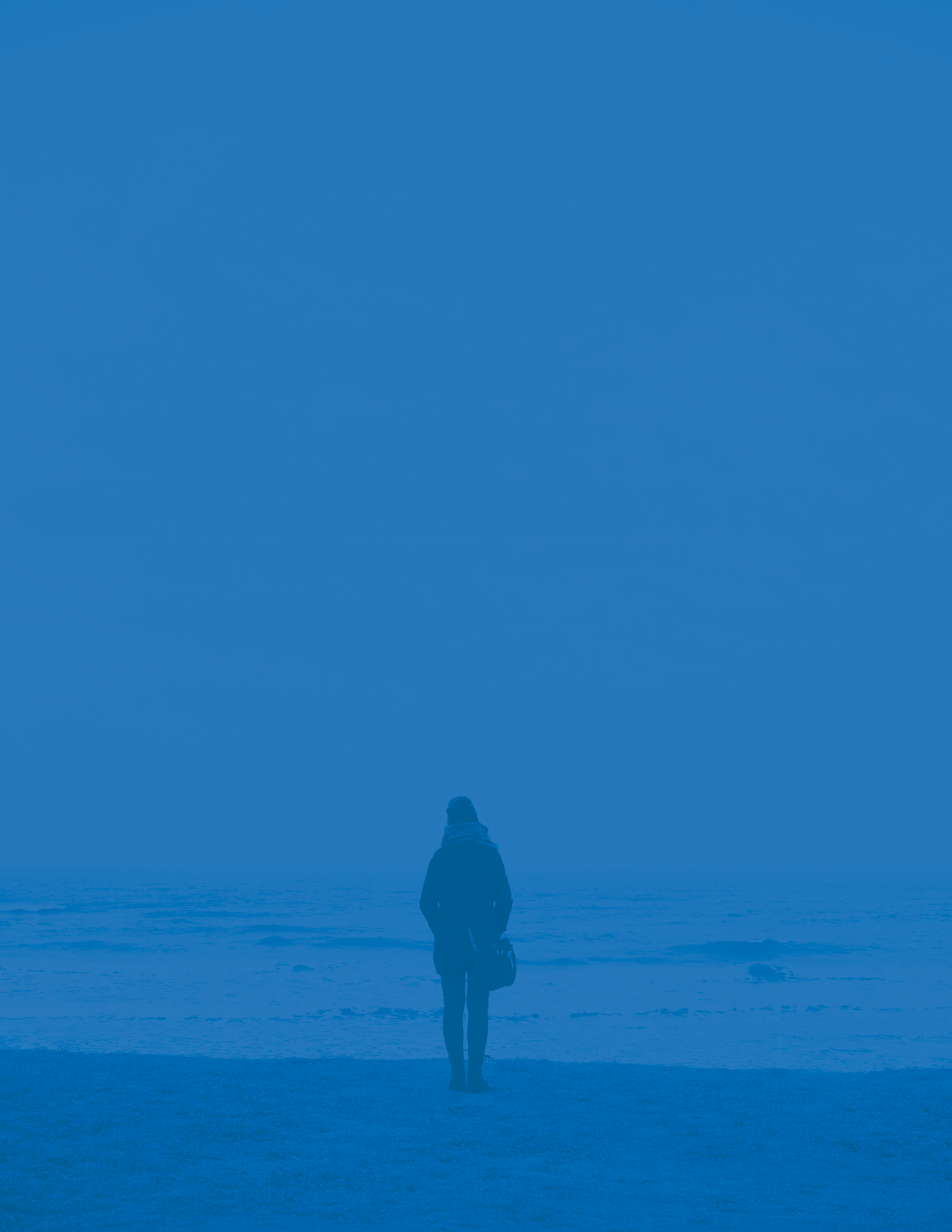
In the years prior to Zach's diagnosis, we knew he had intellectual and developmental delays, and, of course, he never slept. We made accommodations as needed but we did not put limits on him. We expected him, with modifications, to participate in family events and vacations, to learn how to ride a bike, swim, hike, etc. He had every therapy you can imagine on a regular basis from the time he was one-year-old. With the diagnosis of SMS came a way of thinking about how to best support Zach in terms of academics and life skills learning. I do not mean we stopped doing things with Zach or lowered our expectations; rather, we were now in a position to focus our energy in a more productive and supportive way for him. Keeping in mind that he was 15, we shifted from straight up education to include more life skills lessons.

Once Zach was diagnosed, we felt vindicated in how throughout his life we insisted on services and best practice for him. We were validated in that we knew what was best for our son, and now providers had to really listen to what we were saying about how to achieve success with Zach. We were glad that even without a diagnosis we listened to our instincts and pushed the system as hard as we did. Once we got the diagnosis, we had a map. We could move forward and support our son in the best way possible.

Leah Baigell

You Are Not Alone

Receiving the diagnosis of Smith-Magenis Syndrome (SMS) will likely bring on a range of emotions for you and your family. Parents and caregivers can feel both sadness and relief when their child is diagnosed. For many, it has been a long journey looking for answers or the right path; for others, the diagnosis comes when their child is very young. With a confirmed diagnosis of SMS, you have places to go to for information. More information can lead to more questions: how to best advocate for your child, provide for and fulfill their needs, should my child go to a special class or school, who will pay for that, what about doctors, who do I see, what does the future hold and so many more questions.



Emotions You Might Experience

When you learn that your child has a diagnosis of SMS, you may be filled with conflicting emotions: shock, relief, heartache, grief, validation. Many years ago, a parent of a child with SMS asked several families to share their first thoughts upon receiving the diagnosis of SMS, marking their child as likely needing support for the rest of their lives. The most common responses were:

Whatever emotion you are feeling now, know that you are not alone. Wherever you are on this journey, there are hundreds of families walking in front, alongside and behind you sharing that same trip. You know it well - the one you did not sign up for - the one you just got. Do not fear your emotions, take proper time and care to process them. There is support and guidance available. There are people who can answer your questions or guide you towards the support you need. The following offers some advice that we found helpful when experiencing a few of the above reactions.

Psychiatrist Elizabeth Kubler-Ross developed a model known as the *Five Stages of Grief* that she defined as denial, anger, bargaining, depression and acceptance. While Kubler-Ross' model has roots in medical illness, the stages in her model, as well as other elements of coping, lend themselves to non-medical situations such as receiving a diagnosis of SMS. While this theory is based upon observations and not scientific research, we found some of her concepts to be applicable to the grieving and loss that we experience as parents and caregivers. For our purposes, we will touch on elements of coping including sadness or grief, anger, denial, loneliness and acceptance. An individual can experience and move between these five emotions for any length of time and in any order. At times, you might find yourself back in a place you thought you worked through. It is helpful to recognize what you are experiencing and work towards acknowledging the challenges you might be facing.

Sadness/Grief

It is okay to mourn the loss of hopes and expectations you had for your child. It is all right to feel sad and it is okay to struggle with realizing that the future will look different from what you envisioned. Allowing yourself to feel sadness can help you grow. Crying can be cathartic and healing. It is important to recognize that sadness is different from depression. Some people can fall into depression and if so, it is important to get proper help. It could be helpful to find a therapist or someone to help you get through this challenging time.

Anger

You might feel anger directed at friends and family. It is not uncommon for parents to feel resentment or jealousy towards parents of 'typical' children. Anger is normal and a part of the process of acceptance. Anger is your way of communicating with those around you that you are feeling hurt. Sometimes friends and family do not believe what you are telling them. They, too, need time to process. Many parents report being told "he or she will outgrow it [the behavior]." Direct people to the PRISMS website for information as it is very likely they do not have an understanding of the syndrome.

Denial

You may find that there are times when you refuse to believe the diagnosis for your child. This is a natural response to coping. You might avoid accepting facts that relate to your child's diagnosis, and you might find reasons why your child does not fit the SMS diagnosis. Be careful that your denial does not get in the way of your child receiving appropriate treatment. Physicians and therapists might give you information that is difficult to hear about your child. Try considering the information when you have had some time to calm yourself.

Loneliness

After the diagnosis, you might feel alone or isolated. You are in a new situation, one that can feel alienating. It can be very hard to relate to friends and family. You are never alone; PRISMS is filled with support and information for you and for your child with SMS. As you get accustomed to having a child with SMS and find a community of parents with children with special needs, the hope is you will begin to feel less alone.

Acceptance

In this frame of mind, you accept and embrace the diagnosis. You will continue to face challenges but you recognize the reality of the diagnosis and take steps to move forward with renewed expectations.

It is important to process the diagnosis in your own way and in your own time. Make every effort to move forward in support of your child, and do so with awareness that some days will be harder than others. It may be beneficial to focus on the here and now, taking one step at a time and being patient with yourself.

Sharing the Diagnosis

In this section, we will talk about how to talk to others about Smith-Magenis Syndrome. Before we get to that, there are two things to think about. First, it is our job as parents to educate and inform others about SMS, particularly health care professionals and teachers who, chances are, know nothing about the syndrome. Given that SMS is rare, we are all unwitting ambassadors, and spreading the word is important. Second, it is important to think about and be aware of how we use language when we talk about the syndrome and our child. When we talk about our child with SMS, we want to be sure to place the child before the syndrome. In other words, try not to say "my SMS kid". Rather, say "my child with SMS." Words matter.

There are a variety of people in your lives for whom having information about SMS is important, including siblings, extended family, friends, colleagues, medical professionals and educators. What do we share, with whom do we share, when is a good time to share, who needs to know, what if they have a reaction I'm not prepared for, and what if they don't understand? How can we respond to the onslaught of questions when, at the early stages of diagnosis, we might not have the answers ourselves?

Let us start with family members and close friends. How do you talk to family members and friends about your child with SMS? It is important to keep in mind that as new as you are to the diagnosis, and as challenging as it was for you to wrap your mind around your new "normal", it will be doubly challenging for those around you to grasp what you are experiencing. They are not living your life and have no template against which to compare the information you are sharing. People are accustomed to standard diagnoses and medical issues, and know how to respond to those. For SMS they might feel uncomfortable and grasp at things to say. For example, you might hear, "She'll outgrow it", "If you just discipline him, he'll be fine" or "No baby ever sleeps." People mean well. They just do not know what to say, and it can be exceedingly frustrating when you try to explain how you feel. One might say that Smith-Magenis Syndrome is not a "casserole illness." People know what to do if you break a limb, need to replace a knee or have chicken pox. But with a rare genetic syndrome, they do not know how to respond or what to say and do.

So what should you tell people about SMS? Tell them as much as you are comfortable telling them. The more you share, the more

others can begin to understand and even research on their own. People follow your lead. If we are embarrassed or ashamed, they will likely not ask too many questions or know how to help. It is important to normalize having a discussion about SMS, and that includes being open to it ourselves.

How do you talk to your other children about the SMS diagnosis? As with family and close friends, it is best to share as much information as you can in age appropriate language. Experience shows that siblings want to know how they will be affected. It is important to let them know they cannot "catch" SMS, and if they "had" the syndrome, you would already know. Some individuals with SMS have significant medical issues requiring multiple emergency or surgical interventions. This can be scary for siblings, especially if they are not well informed. If you do not offer information, they might imagine something worse. Siblings want to know if their brother or sister is going to be okay. They need reassurance that you are doing all you can to help their sibling. Kids roll with things more than we give them credit for. By talking openly with them, you are providing knowledge. Siblings need to know they can come to you with any question. Your home environment is the home they know so chances are, regardless of the challenges their sibling presents, they will be comfortable with him/her.

For educators and doctors, as with the groups above, the more information they have the better able they are to work with your child. Transparency is key. Provide information, come in with print outs on SMS, direct them to the PRISMS website, offer to come in for meetings to present and respond to all their questions.

How about when people you do not know look at you funny, or say things like "Can't you control your kid", or "Keep your kid quiet." What do you say to these people? Try to come up with a phrase you can reliably pull up that you are comfortable saying. For example, you can say, "My child has a rare genetic syndrome and this is how he/she expresses him/herself." Several families have printed small cards with a brief description of SMS that they hand out at such times. That saves you from having to repeat the story over and over. It also serves to educate the community.

Bottom line is to be open to sharing information, take on questions, answer as honestly as you can, and support your children. If people in your life do not "get it", do not blame them, and do not spend time getting upset. Instead, choose how you want to engage. Take care of yourself and your family.

How Do I Get My Child the Help He/She Needs?

A diagnosis of Smith-Magenis Syndrome will introduce you to new information about a wide range of topics and to a network of support available from professionals and other families managing the syndrome. SMS is a life long genetic condition, but the long-term outcomes for each child affected are highly variable. There are systems, interventions, therapies and resources you and your family can take advantage of that make extraordinary differences for your child and his/her development.

EDUCATIONAL AND THERAPEUTIC PROGRAMS TO SUPPORT THE GROWTH OF YOUR CHILD

Depending on the age and needs of your child – you will experience a variety of educational settings or situations. There is a lot of information out there and it is important to learn and begin to process it as early as possible.

Early Intervention (EI)

Children with Smith-Magenis Syndrome typically face delays in specific developmental areas. Early intervention has proven successful for many individuals with SMS and their families. It is highly recommended to secure these services as soon as possible.

Early intervention is a systematic program of therapy, exercises and activities designed to address developmental delays that may be experienced by children with SMS or other disabilities. Research shows that early intervention treatment services can greatly improve the development of basic skills in a child with SMS. Services include therapy to help children talk, walk and interact with others. The most common early intervention services for infants and toddlers with SMS are speech & language therapy, occupational therapy and physical therapy.

Early intervention can begin any time shortly after birth and typically continues until the age of three. The predominant goal of early intervention programs is to enhance development by building on a child's strengths and building functional skills.

Each state has its own set of regulations governing early intervention services. The first step in accessing services is to obtain a referral. Your child's physician can help you determine the need for therapy or you can have your child evaluated by a therapeutic service provider. Evaluation is provided free of charge, and you can request an evaluation through a local program directly.

Visit the Early Childhood Technical Assistance Center at http://ectacenter.org/ for a list of contacts by state.

Individual Family Service Program (IFSP)

The Individual Family Service Program is a written treatment plan for early intervention services for young children with developmental delays. The IFSP only applies to children from birth to 3 years of age. The plan details your child's current levels of functioning, specific needs and goals for treatment and is a family-based approach. As a parent, you are an integral part of the development of this document. Other members of the planning team may include a neurologist, occupational therapist, physical therapist, speech-language pathologist and more.

Individuals With Disabilities Education Act (IDEA)

When your child reaches the age to enroll in the local school system, their rights are protected by the IDEA – Individuals with Disabilities Education Act. This is a Federal Law, enacted in 1975, that ensures your child has access to "free and appropriate" education. Until they graduate, these rights are protected.

The law also specifies that children with various disabilities are entitled to early intervention services and special education. It does not, however, prescribe the supports or settings that will benefit your child. Parents, teachers and school administrators will negotiate these details. Working collaboratively and with shared goals is the ideal. Sometimes this process can be contentious and may require legal assistance or support from an advocate.

Free and Appropriate Public Education (FAPE)

In each phase of mandated educational services, an individualized plan is developed by the parents and professionals involved with the child's development. The name of this document varies by states, though commonly called an Individual Education Plan (IEP), or Individual Family Support Plan (IFSP). This plan becomes the legal document describing the mandated educational services required for the child. Parents have the right to challenge and advocate for mandated services in a plan to meet the educational needs of their child. As a parent, you are entitled to be treated as an equal partner with the school district in developing an education plan for your child. Your child is entitled to an education that is tailored to his or her special needs and a placement that will allow them to make educational progress.

Establishing an "appropriate" education is a collaborative process that may require considerable negotiation in order to secure appropriate services and assure that these services are meeting your child's needs. It is important to remember that the word "appropriate" does not always mean "best." You may find that programs outside of the public school setting are a better fit for your child. Parents can utilize advocacy, negotiation, formal appeal and legal assistance to secure appropriate services for their child.

Least Restrictive Environment (LRE)

A school district is required to educate a student with a disability in regular classrooms with his/her "typically" abled peers. The child must be educated in the school he or she would attend if not disabled, to the maximum extent appropriate, and supported with the aids and services required to make this possible. This does not mean that every student must be placed in a general education classroom, but rather they are placed in as natural a learning environment as possible within his/her local community.

Individualized Education Program (IEP)

At no time should your child be subject to a setting or a program that you do not support. Building an understanding of your child, their learning style and their personality will inform the educational strategies. Being open to understanding through evaluations and assessments from a variety of specialists can provide confidence about the benefits of certain methods or therapeutic supports. A child receiving special education services must have a documented Individualized Education Program (IEP). That is the law. The IEP documents your child's learning needs, the services the school will provide and how progress will be measured. Children from age three through high school graduation (or a maximum age of 22) may be eligible for an IEP.

As a parent, you are an integral part of the IEP process and the creation of this important document. The process will include teachers, paraprofessionals, counselors, school psychologist, therapists and others who support your child.

By law, here are some aspects an IEP must include:

- Statement of your child's present level of performance
- Annual educational goals for your child
- Special education supports and services the school will provide to help your child obtain these goals
- Any modifications or accommodations the school will provide to help your child obtain these goals
- How and when the school will measure your child's progress and report back to the parents
- Transition planning for your child for life after high school

THERAPIES

Applied Behavior Analysis (ABA)

Applied Behavior Analysis, or ABA, is used by therapists to teach communication, play, social, academic, self-care and community living skills and to reduce 'negative' behaviors. There are alternatives to ABA, which should be considered if your child does not respond to a particular behavioral approach. Do not hesitate to pursue alternative behavioral therapies if your child struggles with or is not happy with ABA or another behavioral program.

Speech-Language Therapy (S/L)

Onset of speech is often delayed in children with SMS as articulation can be affected by muscle tone. Once speech has been acquired, children often experience difficulties with processing information. Speech-Language Therapy can minimize or remediate these deficits.

Please visit PRISMS Speech and Language page at https://www.prisms.org/about-sms/living-with-sms/speech-language/ for more information.

Occupational Therapy (OT)

Children with SMS often have visual-spatial deficits and difficulty with fine muscle control. Occupational therapy focuses on fine motor skill development in children and can be a source of help with feeding issues, sensitivity to textures and more.

Please visit the American Occupational Therapy Association in your state for more information.

Sensory Integration Therapy (SI)

Children with SMS can become over stimulated by senses including touch, movement, sight and sound. Sensory Integration Therapy can assist your child in processing these senses in a way that calms your child, reinforces desired behaviors, and helps to transition between activities.

Physical Therapy (PT)

Children with SMS can exhibit issues with balance and weak muscle tone. Physical therapy addresses gross motor skill development.

Please visit American Physical Therapy Association for more information.

Medical Management

Because SMS is a genetic syndrome, there is a multitude of abnormalities that can manifest to different degrees. In other words, everyone with SMS is different and there is not one particular way to help your child.

It is important to find a primary care doctor who is sympathetic to the needs of your family and available to guide medical care. When looking for a physician, ask about their interest in supporting patients with special needs. Be prepared to switch to a doctor with whom you are comfortable and who is able to meet the needs and challenges of your child.

The needs of your child will change as he or she ages. At different moments in his or her life, there will be new priorities. Your capacity to address these needs will vary as well. Be prepared to adjust your focus to a new priority, be comfortable with doing the best you can and take things as they come.

PRISMS MEDICAL MANAGEMENT GUIDELINES AND FINDING PRACTITIONERS

PRISMS has prepared Medical Management Guidelines at https://www.prisms.org/about-sms/living-with-sms/medical-management-guidelines/ as an outline for the medical needs of a person with SMS. Be sure to share these guidelines with your doctor and others on your medical team. Work together with all medical practitioners to review the needs of your child as they relate to the Guidelines. You will find a checklist of medical procedures and evaluations that are pertinent in understanding the overall health of your child. GeneReviews also publishes these guidelines in a more comprehensive, in-depth review of SMS. These are exceptional starting points from which parents, guardians, providers and professionals can get background information on what is known thus far about the possible medical manifestations of Smith-Magenis Syndrome. Each person with SMS is an individual who needs caring and committed parents, guardians and providers who are able to think outside the box to ensure timely and accurate diagnosis of any medical conditions and secure resulting treatment or mitigating interventions.

Whether your family member with SMS has many medical needs, or few, you need to determine when to add medical professionals to the team and how to find them. Your primary care doctor can help identify which specialists you may need. There may be times where you have a gut feeling that your child needs further care in

some area. Trust your gut and share your concerns. Remember that you are the best advocate for your child with SMS, and in most cases, you have choices about their care. Medicine is not an exact science. Often, more than one intervention needs to be tried. Do not be afraid to interview providers, ask questions, seek second opinions and ask for accommodations that you or your family member might need in any given medical setting.

It may be difficult to locate professionals equipped to partner with you in diagnosing and treating any medical conditions that the person with SMS might encounter. Gently encourage your provider to research and think outside the box in caring for a person with Smith-Magenis Syndrome. For example, one parent of a three-year-old was referred to a sleep specialist. At the first appointment, the highly recommended doctor was not familiar with and had not researched Smith-Magenis Syndrome. The parent requested the appointment be rescheduled until he had time to do this. From then on, that doctor partnered very well with the family, listened, asked questions and considered the parent's input. This physician became one of the fiercest advocates for the child with SMS, as well as an SMS expert. Although not all doctors respond this favorably, do not be afraid to ask for what you need. If the provider is not receptive, move on to another provider.

SMS CLINICS

Consider a visit to an SMS clinic at a university-based Center of Excellence for a comprehensive evaluation. The SMS clinics offer persons with SMS and their families an opportunity to receive targeted medical and clinical care and supervision to address the specific challenges, including health concerns associated with the syndrome. The clinics provide an opportunity for a multidisciplinary team of SMS experts to assess, consult and collaborate on a cohesive plan of care for each patient. For more information on these clinics go to https://www.prisms.org/research/sms-clinics/.

For the families with multiple medical issues and hospitalizations, remember to take it one day at a time. You are the best advocate for your child, but you will need to pace yourself, as it can be overwhelming and exhausting. When talking to a medical provider, it can be helpful to bring someone with you to listen, take notes, and later replay the information that was shared. In some cases, this person can attend to the family member with SMS so that the parent can direct full attention to the conversation with the provider.

PLANNING & PREPARATION FOR MEDICAL VISITS

With a diagnosis of Smith-Magenis Syndrome, your child will likely need health and related services more than most individuals. This means frequent appointments with doctors, specialists, therapists and other professionals who will help to provide the support and care your child needs. These consultations are critical to ensuring the overall progress and development of your child, and therefore it is important for you to get the most out of each visit. This can be a challenge.

Advance preparation can help make the medical appointment or hospitalization go more smoothly. You can set the stage for success at medical appointments by having food, change of clothes, comfort items and activities to distract while

waiting. Schedule appointments at times of day that your child with SMS is most likely to be successful. Request accommodations to decrease wait times. For example, fill out forms or talk to the nurse ahead of time. Many clinics or hospitals have resources online to help prepare a social story to let the child with SMS know what to expect at the appointment, or to make a visual schedule to use at the appointment, if helpful for the patient. One parent has a routine of going to a vending machine to get a favorite snack after doctor appointments. Being prepared, having a predictable routine, knowing what to expect, and getting a preferred activity or reward during and after the appointment are all things that can help things to run smoothly.

Going to a doctor's appointment can be tough. Unlike other children, my youngster with SMS is not afraid of getting hurt (i.e., vaccinations), but she hates the long waits and having to be compliant with doctors on their timetable. A few things help us with this:

We have a bag of toys she only gets to play with at the doctor's office. This helps her to be excited about an appointment and keeps her occupied while we are in the waiting room.

We take breaks to go on a walk or watch a video on her iPad. We insist on breaks whether it inconveniences the doctor or not. My daughter's mental health is as important as her physical health. These breaks often mean the difference between a tantrum or not.

We have a doctor kit that my daughter gets to bring to appointments. She may use a stethoscope to listen to the receptionist's heartbeat or ask the doctor to say "ahh" to look down HIS throat. Having her own set of 'tools' helps her understand the various parts of the exam. In addition, letting her 'perform' an exam allows her to feel empowered and important in the process.

Shannon Duvall

TIPS FOR MEDICAL VISITS

There are steps you can take to ensure each visit is as beneficial and efficient as possible.

Keep a record of what professionals you have visited and keep their contact information handy. This is a lifelong process, and you will need to recall the medical practitioner you visited as well as the information from medical procedures and evaluations. You might also keep a chronology of each test with the date of the procedure, the location and the result.

- Before an appointment, be sure to ask what medical records, if any, you might need to bring with you. Whenever possible, complete the medical forms before the visit.
- Anticipate your special needs when you schedule your visit. Ask the doctor's office for accommodations. Patients with SMS may need more time for conducting the examination. You may find it difficult to wait in a crowded waiting room due to your child's sensory issues or behavioral needs. Feel free to ask the office if there is an alternate space to wait. Schedule the appointments at optimal times for your child's comfort (i.e., first in the morning when things are quieter and delays are less likely).
- Let office staff know if your child gets anxious during medical visits and provide suggestions about how you might work together with them to mitigate any 'negative' behaviors during the visit.
- You may find that you are pre-occupied in managing your child's behavior during an office visit and may not be able to fully attend to the physician or staff during the appointment. Bring another family member or friend to the appointment who can attend to your child while you listen, ask questions and take notes.
- Request that staff write down any plans for care and information on any recommended medical treatments to take with you following the exam.
- You will have the responsibility for coordinating your child's care. Medical staff can assist with the continuity of care by working to improve timely communication with your child's other medical specialists. Your child's physician may also be able to help you make sense of multiple recommendations from different providers.
- Be assertive and do not forget that you are the expert when it comes to your child.
- You and your child should feel comfortable and confident in the professionals working to meet your needs. Sometimes in your dealings with doctors, you may find that you do not feel a connection or a sense of long-term relationship. Know that it is all right to look for another provider.

Community Engagement and Inclusion

Your child with SMS is a valuable member of many communities: family, school, religious, neighborhood and others depending on his or her interests and family values. You will want to prioritize what social groups you would like him or her to be a part of, and advocate as necessary to promote inclusion in that community. For school age children, it may be school and after school activities. For teens and young adults, it may be other social groups, clubs, work or day programs. There are so many opportunities in your communities that your child can be a part of with some creativity, motivation and advocacy.

Many online resources can help in identifying community resources in your area. One example in the United States is a state-by-state resource guidebook published by Autism Speaks at https://www.autismspeaks.org/family-services/resource-guide.

Routines and rituals are very grounding for children with SMS. It is critical that a person with SMS be prepared with details of what to expect at any given time. Anticipating needs, creating familiar routines and structure, and being proactive can minimize or prevent problems before they occur. This is the key to success for a person with SMS in any community setting.

Finding the Support and Resources for Caregivers and Their Families

Caring for a child with Smith-Magenis Syndrome most likely will be the most challenging and rewarding job you will ever have. You might feel as if you are the only person who can do the job and doubt that anyone else could meet your child's needs the way you can. There is only so much you can give before you will begin to feel mentally, emotionally and physically drained. It is a necessity – not a luxury--to find the support you will need to care for you and your family. Remember, you are only human and it is important to take care of yourself physically and emotionally.

TIPS FOR CAREGIVERS

- ***Put Yourself on the To Do List/Take Care of Yourself*** - You cannot pour from an empty cup. To be there fully for your child with SMS and your family, you must take proper time to focus on yourself and the things that bring you joy and calm.
- ***Getting Enough Sleep*** - This is not an easy task with an SMS individual living in the house. Explore ways to catch up on missed sleep. Nap whenever possible. Share nighttime duty. Explore overnight respite services.
- ***Understand That You Have Power*** - It is true, there are some things you cannot control, but there are many that you can. Remember the things you have the ability to affect, including your child's medical and professional team. Speaking up and advocating for your child can empower you and give you the confidence to be a better parent.

- ***Develop a Support Network/Be Informed*** - It is important that you take time to understand and process Smith-Magenis Syndrome and its implications for your child and for your family. It is equally important that you are not afraid to ask questions and allow yourself to seek honest and accurate information from a variety of sources. Connecting with other families of children with SMS can be an eye-opening and beneficial experience. PRISMS offers many opportunities and venues to reach out to other parents. Support may also be found through your church, local developmental disability services agency as well as friends and family members. Invite them to share your journey as an SMS caregiver. You cannot do this alone.
- ***Get Physical*** - Research shows that exercise can lower stress, cause you to be less anxious and improve your overall mood. Walking or other aerobic activity just three times a week for 30 minutes can make a huge difference in your life.
- ***Keep a Positive Outlook*** - Seeing the cup as half-full instead of half-empty can help. Your child, like all children, is never done growing and developing. Along the road, there will be challenges and hurdles to

overcome, but there will also be victories. Celebrate each success and take time to acknowledge the accomplishments.

- ***Take Time for Your Other Relationships*** - Your child with SMS will thrive when your other relationships thrive. Remember to take time for one-on-one activities with your significant other, typically-abled children and friends.

MARRIAGE CARE

Couples raising children with SMS face distinctive challenges. It is important that you take regular time to care for your marriage through open communication and attention. While every couple is unique, there are some common issues (see below) that couples raising children with disabilities face. Discussing them openly can be helpful.

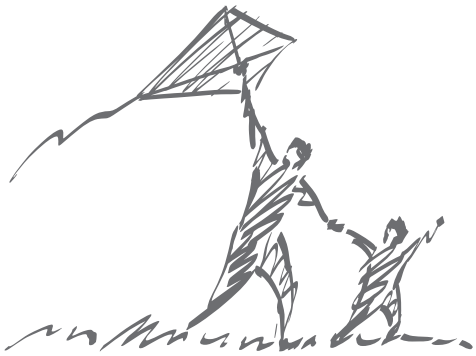
- ***Imbalance in Caretaking*** - You may find that one parent feels as though they are shouldering the full burden of care for their child with SMS. The other parent may feel that they have lost their partner to full-time parenthood. Openly discuss this imbalance and work to create steps for both parents to find a more cohesive balance in sharing responsibilities.
- ***Feelings of Isolation*** - You may find that since the diagnosis you and your partner have begun withdrawing from each other. This is a common reaction when feeling overwhelmed. It is important to recognize if you begin to feel yourself withdrawing; discuss with your partner ways in which you can increase intimacy and connectedness with one another.
- ***Anger*** - It can be difficult to adjust to your child's diagnosis with SMS. You or your partner may feel guilt, denial or even anger. This can be especially difficult if one of you accepts the diagnosis and the other does not. It is all right to have different perspectives and to work through the diagnosis differently. It is important to remain respectful of one another and communicate where you are emotionally.

RESPITE

There will be times when you feel like you cannot go on. Taking care of yourself has to be a priority. Just like on an airplane when you are instructed to put on an oxygen mask before helping another, you are responsible for taking care of yourself before you can care for someone else. Respite is a little time off from the round-the-clock responsibilities of your life. Scheduling a regular weekly break can help reduce stress, depression and exhaustion, and give you renewed energy to keep going.

Sometimes raising a child with SMS can be more than one individual can handle. You are going to need the help of friends and family or other caregivers, especially if you are a single parent. Do not be afraid to share your story with those around you and invite them to join you on this difficult journey. Respite can and should be a regularly scheduled part of your week. It may also be a "date night" for you and your spouse or a special time for you and a sibling of the child with SMS. Respite can be just a few hours or as much as a week. You can also access emergency respite care in a crisis.

So where do you find respite and how do you pay for it? You may be lucky enough to have a grandparent or friend who is willing to step in on a regular basis. Because of your child's special needs and often challenging behaviors, hiring a typical babysitter is not always an option. Caregivers often need specific training to take care of an individual with SMS. A good place to start is through your local or state developmental disability agency to seek funding and resources.



Resources can be found at:

- Autism Speaks respite information - https://www.autismspeaks.org/family-services/community-connections/respite-care-and-autism
- Easter Seals - http://www.easterseals.com/explore-resources/for-caregivers/respite-care.html

Ali was about six-years-old and her sister about four when we hired our first "live-in" babysitter. Ali's violent behaviors had escalated and we could no longer trust the abilities of our high school babysitter to handle her outbursts. We were close to a breaking point and our marriage was also beginning to suffer. We needed regular, scheduled respite and decided to find someone qualified to care for our two young girls for about six hours on Saturdays and a "date night" on Wednesdays. We had an extra room in our home that we could convert to a bedroom, and decided to offer our home, along with meals, in exchange for childcare. Most individuals were with us for about a year or two. All had other jobs outside our home. Some were better than others were. Overall, I cannot imagine having survived those early years without it.

Barclay Daranyi

For information related to stress and caring for the caregiver, please consider the following peer-reviewed publications:

Stress in Families of Young Children With Down Syndrome, Williams Syndrome, and Smith-Magenis Syndrome.
https://www.tandfonline.com/doi/abs/10.1207/s15566935eed1104_2
Fidler, Deborah J.; Hodapp, Robert M.; Dykens, Elizabeth M.

Caring for the Caregivers: An Investigation of Factors Related To Well-Being Among Parents Caring for a Child With Smith-Magenis Syndrome.
https://link.springer.com/article/10.1007/s10897-009-9273-5
Rebecca H. Foster, Stephanie Kozachek, Marilyn Stern, Sarah H. Elsea

Stress and Coping in Families of Children With Smith-Magenis Syndrome
https://www.ncbi.nlm.nih.gov/pubmed/9828063
Hod app RM, Fidler DJ, Smith AC

A Message to Parents About Siblings

Brothers and sisters of children with Smith-Magenis Syndrome often experience a unique set of emotions, responsibilities and opportunities when it comes to their siblings. No doubt, there are moments where they feel jealousy, worry, anger and confusion towards their sibling. They have a right to be upset sometimes. The unique experience of having a brother or sister with a disability has its ups as well. Siblings completely and utterly love one another with a love stronger than most.

TIPS FOR SIBLINGS

- ***Seek Information*** - When ready and at a level appropriate for their age, siblings need to learn about SMS. Children need to be provided with honest and accurate information to better comprehend how this SMS diagnosis affects their sibling. They should be encouraged to ask questions and learn about the syndrome. This knowledge can help them to understand more about their brother or sister with SMS.
- ***They Are Not Alone*** - It is important for your other children to understand that they are not alone. Every family is confronted with challenges, and yes, having a member of the family with SMS is certainly challenging. However, everyone has something difficult to face in his or her families. There is a community of siblings who know just how it feels. Joining a sibling support group may help.
- ***Spend Time with Parents*** - Siblings want alone time with Mom and Dad. Having a sibling with SMS can be time consuming. Each child wants and needs individualized attention from Mom and Dad. Make time to provide some one-on-one time.
- ***Find an Activity*** - Look for an activity that the siblings can do with each other that is time for just them. These activities can be rewarding. The siblings connect by doing something special together that creates a closeness and understanding.
- ***Be Proud*** - Learning to talk about SMS and being open to describing how it affects the family is helpful. Like all siblings, sometimes they will love and other times despise one another. Sometimes they may feel embarrassed by SMS. Assure them that having these feelings is normal and okay. Your children will love each other for who they are and be proud of your family!

When I was younger, I would get frustrated because I did not always understand what my brother was trying to convey. As I got older, I learned who Nick was and how he expressed himself. Our special bond has taught me patience like no other. I credit Nick to helping me be a better person and a better mother, now that I have my own son. He is so excited to be an uncle; it melts my heart!

Tabitha Petkovich

From as early as I can remember, Laura has made herself the center of attention. It does not matter if we are at home, at church, at the grocery store or even at the PRISMS conference, Laura finds a way to make herself noticed. Growing up, I began to resent my sister for always drawing attention to our family. Not because I personally wanted the attention, I was never the attention-seeking type, but because sometimes I simply wanted to blend in. I could not help but feel like everything was all about her. Yet, despite, all of the hard times, I know without a doubt that I would not be the person that I am today if I did not have Laura for a sister. Having someone with SMS in our families gives us resilience unlike any other. I learned at a very early age what really matters in life, and how to not let a little bit of wind blow me over. But what I think I have learned the most is how to laugh. My family can testify that more than anything else, Laura makes me laugh.

Jenny Beall

For more information, please consider the following peer-reviewed publication:

Siblings of Individuals With Smith-Magenis Syndrome: An Investigation of the Correlates of Positive and Negative Behavioral Traits
https://www.ncbi.nlm.nih.gov/pubmed/22672270
Moshier MS, York TP, Silberg JL, Elsa SH

I Am an Advocate

To advocate means to speak on behalf of someone or for some cause - it is a synonym for the word "support."

As a parent of a child with special needs, we are placed in the position of advocate – speaking on behalf of our child and their interests over many years and in a variety of settings. We do not ask for this role – it is not really a choice. But who better can support your child? No one knows your child or will have the same commitment to help them as you do.

In this sense, advocacy is a heightened level of parenting when you have a child with special needs. We advocate for our child because they may not have the ability to make themselves understood. As their parent and advocate, we face the challenge of understanding their hopes and needs, envisioning the future that is possible and working with the resources available to achieve our goals. And we do this with courage, patience and grace.

Acting as advocate requires several essential qualities and skills.

- Advocates are knowledgeable and informed about the issues.
- Advocates have a vision, a plan and a purpose for what they are doing and trying to accomplish.
- Advocates are problem solvers, good listeners and diplomatic. We use the same qualities and values that we believe are essential for our children, and apply them to how we work with others.
- Advocates are organized. We keep records of our meetings and the many reports that show the growth and capacity of our children.

The information in this guide is intended to help you be an advocate, build your confidence and increase your skills so you can work to achieve what is best for your child.

For more information about advocacy go to:

- www.Wrightslaw.com — search for "So You Want To Be an Advocate…."
- https://themighty.com — search for " How To Become a Parent Advocate for Your Child With Special Needs
- www.Parentcenterhub.org — a support organization for Parent Centers nationwide

Resources for More Information

While resources vary from location to location, these national organizations can provide you with valuable information and connect you with local agencies and resources in the United States.

PRISMS is the leading SMS organization and serves as the international clearinghouse for information regarding Smith-Magenis Syndrome. Resources and publications can be found at:
https://www.prisms.org/education/publications-and-resources/

The Arc is a national advocacy organization for people with developmental disabilities and their families with local chapters throughout the United States.
http://www.thearc.org/who-we-are/position-statements/life-in-the-community/family-support

Autism Speaks is a national organization advocating for people with autism.
https://www.autismspeaks.org/

Centers of Excellence/Association of University Centers on Disabilities is a membership organization that supports and promotes a national network of university-based interdisciplinary programs to assist people with disabilities and their families.
https://www.aucd.org

> Here is the link to the interactive map to find a Center of Excellence for each state.
> *https://www.aucd.org/directory/directory.cfm?program=UCEDD*

National Disability Rights Network is a nonprofit membership organization for the federally mandated Protection and Advocacy (P&A) Systems and Client Assistance Programs (CAP).
http://www.ndrn.org/en/ndrn-member-agencies.html

National Association of Councils on Developmental Disabilities is the national association for the 56 Councils on Developmental Disabilities (DD Councils) across the United States.
http://www.nacdd.org/home/

Centers for Medicare and Medicaid Services (CMC) provide a state-by-state resource for public program information and funding options:
https://www.medicaid.gov/about-us/index.html
(click on Resource by State tab, scroll down for specific state search).

Get Connected to PRISMS

Founded in 1993, Parents and Researchers Interested in Smith-Magenis Syndrome (PRISMS) is a non-profit 501(c)3 organization dedicated to providing information and support to families of persons with Smith-Magenis Syndrome (SMS). PRISMS supports research and fosters partnerships with professionals worldwide to increase awareness and understanding of SMS.

PRISMS is governed by a ten-member Board of Directors, each of whom has a child with SMS, allowing for a unique perspective and understanding of living with Smith-Magenis Syndrome. A multidisciplinary Professional Advisory Board supports the work of PRISMS by providing medical guidance for families as well as reviewing and sharing information about the most up-to-date research.

PRISMS offers a variety of programs and services dedicated to education, awareness and research on behalf of the global SMS community.

To learn more about PRISMS, visit: http://www.prisms.org

To address questions or comments about this resource, please contact PRISMS at info@prisms.org.

PRISMS SMS PATIENT REGISTRY

The PRISMS SMS Patient Registry is a collaboration between families and researchers that work together to improve our understanding of SMS. This secure database is where we collect medically relevant information on individuals with SMS. Consider enrolling your child into the Patient Registry and help us to better understand the natural history of this condition. With more understanding, PRISMS and professionals can better serve SMS families with supports and potential therapies to improve the life of your child.

Publications

PRISMS creates comprehensive resource publications on a variety of topics to best meet the needs of the SMS community. To date, PRISMS has released publications related to SMS in the school setting and in residential settings as well as a set of medical management guidelines to provide a starting place for medical professionals to support individuals with SMS. PRISMS will continue to create publications to best serve the SMS community at https://www.prisms.org/education/publications-and-resources/.

On the Road to Success with SMS: A Guidebook for Schools
Smith-Magenis Guidebook: Exploring Adult Residential Living

Acknowledgments

This resource guide was developed with the help of many people. PRISMS is grateful for the contributions from the members of the Advocacy Workgroup including Leah Baigell, Barclay Daranyi, Melissa Haley, and John Mayer; Emily Fields, PRISMS Executive Director, who provided leadership and direction; Mark Hernandez, designer of this publication, Pat Boschetto, as proofreader and Deborah Wehrlen as editor.

Most importantly, PRISMS thanks and appreciates the families who shared their personal experiences and wisdom to create this resource.

prisms

Parents and Researchers Interested in Smith-Magenis Syndrome