FOUNDATION FOR PRADER-WILLI RESEARCH

Working Toward an Independent Future

FIRST STEPS

A PARENT'S GUIDE TO SCHAAF-YANG SYNDROME

First Steps: A Parent's Guide to Schaaf-Yang Syndrome

CONTENTS

A Letter of Hope

First Steps: A Parent's Guide to Schaaf-Yang syndrome was written by parents for parents. We are so glad you found us! This book is a way to share the gift of hindsight: what we wish we'd known when we first received our child's diagnosis, or what we did learn that was so very valuable and helpful as we began our journey with Schaaf-Yang syndrome within our own families.

Most likely, you are the parent or family member of a child recently diagnosed with Schaaf-Yang syndrome. In this moment, two things are true: (1) You have a beautiful child who will bring you great joy and has the opportunity to experience a beautiful life. (2) You are not alone in your journey nor with the challenges of SYS.

The SYS community is fierce in its determination to do whatever we can to provide the best possible care and the brightest futures for our children. While the list of potential challenges associated with Schaaf-Yang syndrome is long and sometimes difficult to consider, the parents whose voices you'll hear in this book are committed to doing all we can to ensure our children live happy, healthy, fulfilling lives.

If there is one piece of advice that most of us would give to families with a new diagnosis of SYS, it is to not believe everything you read about SYS. All too often, the picture painted of SYS is so bleak that it can overwhelm families to the point of despair. If that is where you are, we are here to say that THERE IS HOPE! Our hope is that First Steps will connect you to helpful resources, alleviate some of your fears about your child's future, and empower you as you move forward.

—Susan Hedstrom
Executive Director
Foundation for Prader-Willi Research

A Message from Dr. Christian Schaaf

Welcome to the wonderful group of families and friends who care about loved ones affected with Schaaf-Yang syndrome. I hope you find comfort in having received a diagnosis and getting some answers for your most pressing questions. Most importantly, you are now connected to a group of families who are all on the same journey as you, a group of people you can turn to at any time. This will be incredibly powerful.

There are experts in medicine and biology who understand Schaaf-Yang syndrome - experts who care deeply about the individuals affected with this rare syndrome, who are interested and motivated to learn more about it. This is also a group of physicians and scientists you can turn to and ask any questions you may have. We may not have the answers to all of your questions, but we will try.

We are on this path together. We will work together, trying to better understand SYS, and ultimately, we are trying to find ways to treat it and to improve the quality of life of those affected with this disorder. The syndrome was first described in 2013, and we have come a long way and have learned a great deal about SYS within a relatively short time frame. In part, this is due to a large group of international experts researching the genetics, molecular biology, and pathways affected by SYS.

On the other hand, this is due to the amazing group of families who have shared information about their loved ones, who have been willing to invest time to participate in research studies, and who have gone to great lengths to raise funds to support SYS research.

I look forward to all the things we can accomplish together. I promise you that my team and I will not give up until we find a cure for Schaaf-Yang syndrome. Also, I promise you that we will be at your side. We will do everything that we can to support you, as well as the entire SYS community.

With kind regards,

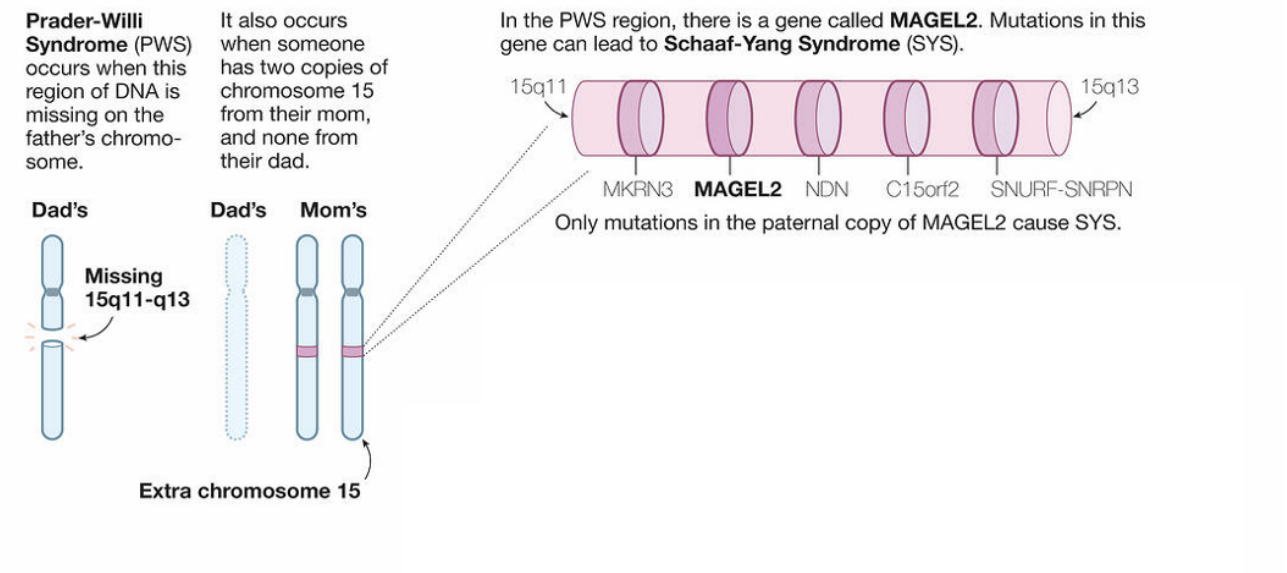
Christian Schaaf
(christian.schaaf@med.uni-heidelberg.de)

What Is Schaaf-Yang Syndrome?

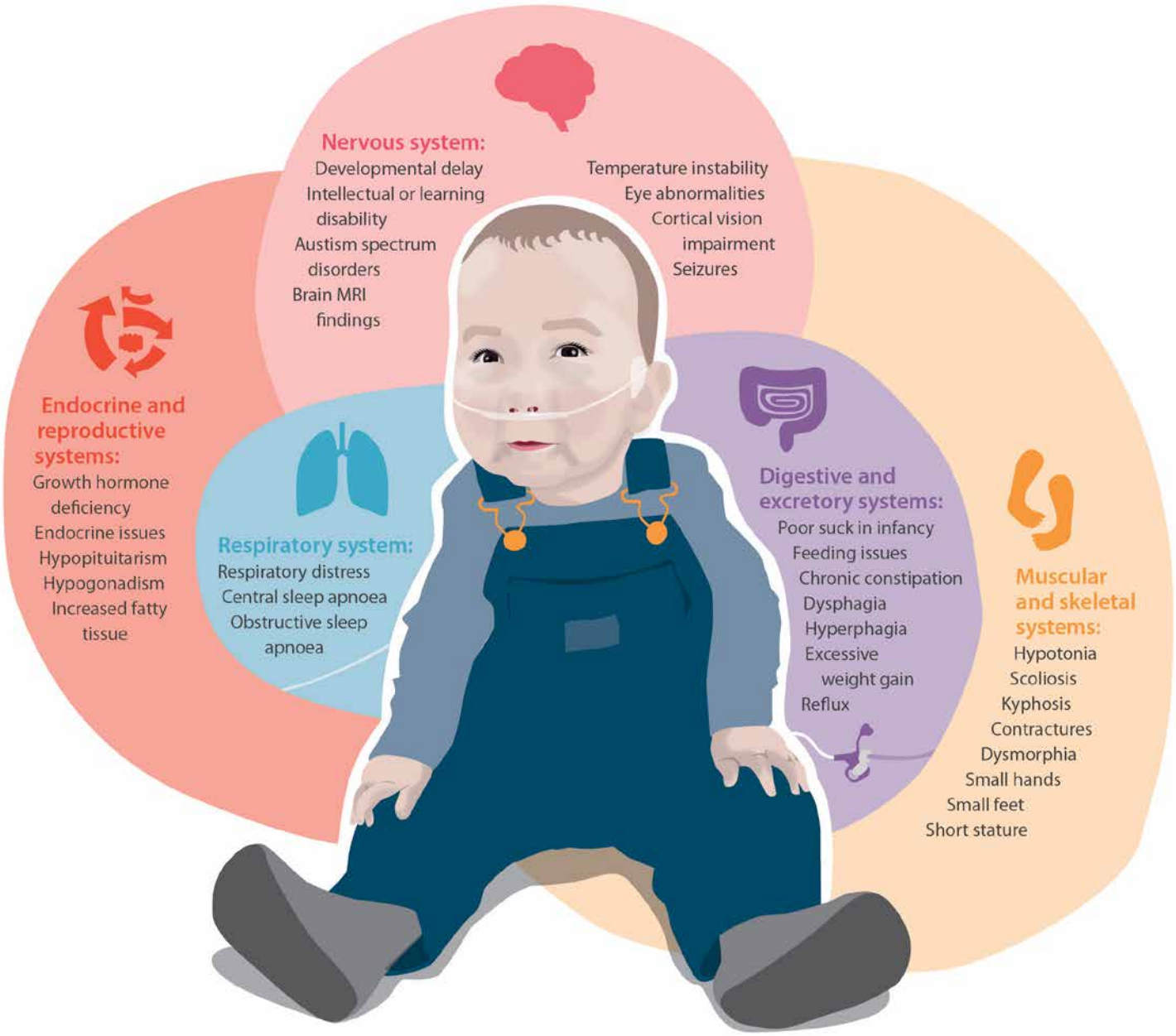
Schaaf-Yang syndrome (SYS) is a rare genetic disorder caused by a disruption of the MAGEL2 gene on the paternal copy of chromosome 15.

The disrupted gene causing Schaaf-Yang syndrome is also missing in another, more common genetic disorder, Prader-Willi syndrome (PWS). PWS is caused when MAGEL2 and several additional genes on chromosome 15 are missing or inactivated.

Schaaf-Yang syndrome shares many clinical features with Prader-Willi syndrome. Those include hyperphagia, hypotonia, feeding difficulties during infancy, global developmental delay/ intellectual disability, hypogonadism, obesity, and sleep apnea. However, people with Schaaf-Yang syndrome also have a relatively higher prevalence of autism spectrum disorder, and almost all of them manifest joint contractures, typically affecting the finger joints, but in more severe cases also involving knees and elbows.



What Is Schaaf-Yang Syndrome? (continued)



Body systems affected by Schaaf-Yang Syndrome

Above:

The body systems involved in SYS, with possible symptoms. This is a range of symptoms that have been observed in people with SYS, but most people will not experience all of them.

Frequently Asked Questions

How common is SYS?

We aren't quite sure! SYS has only recently been identified and described in the medical literature. Our current estimate of the incidence rate is 1 in 100,000 births. This estimate could change as we learn more about the syndrome and more people with SYS are diagnosed.

Is SYS inherited?

Yes and no. Some cases of SYS are de novo, meaning new genetic changes that occurred by chance. These cases are not inherited. In other cases, the father is carrying a silent chromosome 15 that has a MAGEL2 defect. When this is the case, the chance for each future child to inherit the mutation and to be affected with SYS is 50%.

What causes SYS?

SYS is caused by a disruption of the MAGEL2 gene on the paternal copy of chromosome 15.

How is SYS diagnosed?

A suspected diagnosis of Schaaf-Yang syndrome may be confirmed through whole-exome sequencing or through MAGEL2 sequencing.

Is there a cure for Schaaf-Yang syndrome?

Currently, there is no cure for Schaaf-Yang syndrome. The Foundation for Prader-Willi Research is interested in advancing research toward understanding and treating specific aspects of the syndrome, with the goal of an eventual cure.

Will my baby meet typical developmental milestones?

There is great diversity amongst people with SYS. However, research has established some average ages at which certain milestones may be reached. It is important to remember that 'later' is not the same as 'never' and that you will celebrate your child's achievements with just as much joy (or maybe more!) when they do occur.

Milestone	Average time	Range
Sit independently	18 months	8-60 months
Crawl	31 months	12-84 months
First words	36 months	6-132 months
Walk	50 months	14-132 months
Two-word sentence	40 months	17-60 months

Parents' Stories

Love. The love you will have for your loved one with SYS may be filled with a wide variety of emotions. At times, it may feel like you are not doing enough for them. My husband and I struggled with blaming ourselves, depression, and tons of anxiety. At first, we had no idea what was wrong with our baby, which caused what should have been a moment filled with love and happiness to be kind of dark. After she was finally diagnosed, things seemed to fall into place. She is our little ray of sunshine. My daughter is strong and always up for a challenge. I never thought I would have such a wonderful child. Savanna has offered so much to our family and I cannot picture my life without her. If you find yourself wondering 'why' and 'how,' trust that your sweet child will give you the strength to push forward and that the word *love* will not compare to the connection you will have to your child.

Communication. Communication may be different with your child than what was expected, but you will learn to understand what your child with SYS needs just like you learned how to care for your newborn. You and your child can take advantage of many different therapies, including speech therapy. This therapist will help determine what works for your family and may help you navigate the world of communication devices. As your child grows, they will work toward using sign language and words.

Siblings. It is normal for typically developing children to have different feelings about their sibling with disabilities. Some children may experience jealousy of the time parents spend with their sibling, feelings of embarrassment or guilt, or stress about changes in the home. To reduce these concerns, it's important to prioritize scheduling one-on-one time with your other children, keep an open and safe forum for kids to communicate their feelings with you, and reduce pressure to feel responsible for their sibling. Although parents often share their concerns about their other children, there are benefits to raising a child alongside a sibling with disabilities. Many siblings grow up with a greater level of maturity, a better understanding of social behaviors like empathy and gratitude, and a higher acceptance of diversity.

First Steps After Diagnosis

After you first receive your child's diagnosis, it may feel like the whole world has shaken around you, and the list of questions and concerns for your child will grow quickly. To help guide you into this new journey, we have created a suggested "to-do" list in the first few months/years to help prioritize the care needed ahead.

Love Your Baby! Your little one needs attention and affection from the start, as does any baby. Provide stimulation and valuable input into your baby's sensory system by holding him, bouncing her, swaying her, talking and singing with him, massaging her, and waking your baby at regular intervals for feedings. A fun example of sensory play is to gently tie a brightly-colored balloon around your baby's ankle and watch them enjoy it while they kick around!

Stabilize Feeding & Weight Gain. Most children, particularly infants, with SYS struggle with feeding due to low muscle tone. While some children are able to eat on their own, others may require a feeding tube to help stabilize weight and retain energy. Consider talking with a gastroenterologist, nutritionist and speech therapist, who can help provide additional guidance on a feeding plan.

Build Your Parent Network. Although it's a rare condition, we have formed a tight-knit community of parents and caregivers who are committed to changing the future of our children with SYS. We encourage you to join our Facebook group where we share and celebrate our kids' successes and struggles, support one another, and share information, novel treatments, and best practices to help build our collective knowledge about managing SYS effectively.

First Steps After Diagnosis (continued)

Start Early Intervention Therapies. The first few years of your child's life are the most formative in their development, and implementing early intervention can lead your child on a path of success from the start. Most early intervention therapies include physical therapy, occupational therapy, and speech therapy, all tailored to your own child's needs and pace. Programs are available in every US state and territory. [1] Please refer to the Early Intervention section below for detailed information.

Begin Growth Hormone Therapy. Recent reports have shown there are significant benefits to starting growth hormone (GH) treatment for individuals with SYS. A study dated May 2021 reports that "all parents reported an increase in muscle strength and endurance, and several families noted beneficial effects such as improved cognition and motor development." [2] Consider speaking to an endocrinologist to discuss treatment options. Additional information can be found in the Growth Hormone section below.

Consult Other Specialists. Depending on your child's needs, you may need to be seen by additional specialists, such as a pulmonologist, gastroenterologist, ENT specialist, neurologist, orthopedist, nutritionist, geneticist, or developmental/behavioral pediatrician. Talk to your child's primary care physician about your concerns and they should guide you to the right specialty. Additional information can be found in the Specialists to Consider section below.

1 - https://www.cdc.gov/ncbddd/actearly/parents/states.html#textlinks
2- https://doi.org/10.1111/cge.14000

Take Care of Yourself.

This journey has undoubtedly been very scary and exhausting. Now that you have found a place among others like you, take this reminder to take care of yourself as well. Your child needs a healthy parent/caregiver, so finding a routine that includes care for your own well-being will be crucial.

Specialists to Consider

As you get started on the SYS journey, it is important to build a team of medical specialists specific to your child's needs. This section is set up to include a description of specialists to consider based on other SYS family experiences, what they will check for and/or monitor, tests for consideration, and terms you may hear from that specialist. It is recommended to identify a pediatric specialist in each of these areas due to the complexity of children with SYS. A specialist's focus may vary by country or even by health care system: this is meant as a rough guide to start from. Please note that not all of our SYS loved ones experience all of these subsequent diagnoses!

- **Endocrinology -** This is THE MOST important specialist you need to see. The endocrinologist will check hormone levels and conduct blood tests. You will want to request an assessment for growth hormone deficiency and consider growth hormone treatment. Terms you may hear: hyperpituitarism, hypothyroidism, hypoglycemia, diabetes, temperature instability, hyperphagia, hypogonadism

- **Sleep Specialist -** The sleep specialist may be a sleep medicine doctor or a pulmonologist and will focus on your child's quality of sleep. You will want to request an overnight sleep study to check for sleep apnea. If starting growth hormone, this is strongly recommended and may be a requirement. Terms you may hear: obstructive and/or central sleep apnea, CPAP, BiPAP, supplemental oxygen

- **Neurology -** The neurologist will conduct a neurological exam of brain and spinal cord systems and may conduct additional blood tests or suggest additional testing. You may request an EEG test for seizure screening or an MRI for abnormal activity. Some tests they perform may also be related to vision. Terms you may hear: hypotonia, seizure, temperature instability, visual evoked potentials

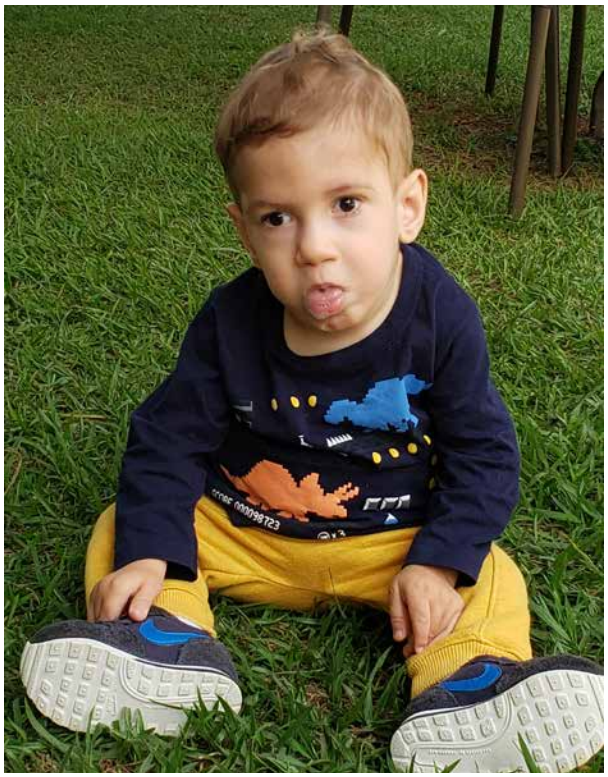
Specialists to Consider (continued)

- **Pulmonology -** The pulmonologist will conduct an assessment of your child's respiratory system, such as the lungs and windpipe. Low muscle tone may cause breathing problems that require monitoring, oxygen, or assistance breathing. Terms you may hear: pulse oximeter, intubation, mechanical ventilation, tracheostomy, endoscopy, aspiration, bronchomalacia

- **ENT (Otolaryngology) -** The ENT will focus on the ear, nose, and throat. Children with SYS may have enlarged tonsils and/or adenoids that affect breathing or sleeping. The ENT may discuss ways to control excessive spit or gagging. Feeding issues are common, and you may request a swallow study. They may also refer to a hearing test and/or recommend ear tubes. Terms you may hear: tonsillectomy, adenoidectomy, aspiration, penetration, endoscopy

- **GI (Gastroenterology) -** The GI will focus on the digestive system. If there is a need, this specialist will advise on the transition to a PEG feeding tube. Other common GI issues that may be addressed include food intolerances such as dairy or eggs, constipation, reflux, and slow motility. You may request a stool sample, pH probe study, a GI motility test, or a swallow study. Terms you may hear: constipation, NG tube, PEG tube, GERD/ reflux, laxative, endoscopy, fundoplication

- **Urology -** The urologist will diagnose, treat, and manage urinary tract and genital problems. They may look for and treat fluid in the kidneys, inguinal hernias, undescended testicles, or reflux from the kidneys. You may request an ultrasound of



the kidneys or genitals or a VCUG for reflux risk between the bladder and the kidneys. Terms you may hear: hypogonadism, hydronephrosis, reflux

- **Orthopedics -** The orthopedist will take care of musculoskeletal problems including clubfoot, hand contractures, curvature of the spine, and hip dysplasia that may occur with SYS. In addition to the physical exam, they may take x-rays to monitor growth, perform DEXA scans to monitor bone mineral density, recommend treatments such as casting and bracing, or perform various surgeries to correct any issues found. Terms you may hear: AFO (ankle foot orthosis), hand splints, scoliosis, kyphosis, hip dysplasia

Specialists to Consider (continued)

- **Developmental-Behavioral Pediatrician -** This specialist focuses on learning and behavioral differences. They will complete a thorough review of the concerns related to your child's developmental, medical, social, and educational history, as well as physical and/or neurological examinations. Then they will recommend a plan that may include a referral for ABA therapy or other behavioral interventions. Alternatively, a child psychologist may also evaluate for an autism diagnosis. Terms you may hear: developmental delay, intellectual delay, autism spectrum disorder, anxiety, ADHD, ADOS (autism diagnostic observation schedule) or other assessment names

- **Geneticist -** The geneticist will help you understand results of genetic testing, explain whether your child's SYS is inherited, and may suggest additional tests or treatments. Terms you may hear: whole exome sequencing, de novo, inherited

- **Ophthalmologist -** The ophthalmologist focuses on the eyes and can prescribe eyeglasses or contact lenses. They will complete an eye exam and may recommend eyeglasses, vision therapy, or surgery. Terms you may hear: cortical vision impairment, dilation, strabismus, astigmatism, nystagmus

Pediatrician

Your child's pediatrician can be their best advocate and serve as the center for all of the specialists above. They may become an unofficial specialist on SYS in the process, as you will. Look for someone willing to communicate with and connect all of these specialists, to advocate for your child, and to act as a partner in your child's full care plan including therapists, insurance, or medical programs.

Early Interventions

We are not just genetically determined. Every one of us is born with a certain window of opportunity for how well we can do certain things. For a child with Schaaf-Yang Syndrome, that range is shifted from your average child. These kids struggle more, and they need to work harder to achieve certain things. But it's still a window of opportunity; it's a range. And that's where we come in as physicians, parents, and therapists. Because within that range it's our job to help them develop to the best of their abilities and opportunities.

— Dr Christian Schaaf, 2017 Schaaf-Yang Family Conference

Early interventions should begin as soon as a diagnosis has been made. Early intervention in children under the age of three may encourage the achievement of developmental milestones. Physical, occupational, and speech therapies are recommended for infants with SYS.

When seeking a new therapist you may like to ask for recommendations from other parents in your area whose children have SYS or other special needs, or from trusted specialists you already see. If the therapist isn't familiar with SYS (and most won't be as it is so rare), ask if they are experienced with neurological syndromes, global developmental delay, or particular areas of concern for you. If a prospective therapist is willing to make time to understand and discuss your child's needs, this can be a good sign. It may also be worth exploring the possibility of home visits. Sessions conducted in your home not only allow the therapist to see your child in a familiar and comfortable environment but allow them to give more specific suggestions suited to your home and family life.

Early Interventions (continued)

Physical Therapy (Physiotherapy)

Physical therapy is helpful to improve balance, coordination, and strength. Your physical therapist will work with your child to improve gross motor skills such as lifting their head, rolling, sitting, crawling, walking, and transitioning between different positions. They will advise on positioning, exercises, and play activities to encourage the attainment of motor milestones. During sessions, they may also perform physical assessments, such as monitoring back strength in a sitting position and checking on spine curvature. They will also suggest equipment that will support your child's development and participation in daily life, which may include supportive seating or a walker. If available, a specialized neurological physical therapist (known in some countries as a neuro physiotherapist) will be particularly well versed in the neurological features of SYS, but an experienced pediatric physical therapist will have worked with many children sharing similar challenges to your child and will be a wealth of knowledge.

Occupational Therapy

Occupational therapists (OTs) work to improve your child's functioning in daily tasks. For infants, this can include play, communicating, bathing, travel, feeding, and participation in family and community life. Your OT may work on positioning, gross (large muscles) and fine (small muscles) motor skills, vision, sensory needs and integration, and social skills. They may make suggestions as to how toys and equipment can be adapted to your child's needs; for example, many conventional electronic toys can be 'switch adapted' whereby a large button or 'switch' is added for accessible control of the toy. For some children, oral motor exercises may help improve sucking strength in preparation for pre-speech. Additionally, if your child has a suspected cortical vision impairment (where the issue lies with signals traveling to the brain, and not the structure of the eye itself) a functional vision assessment performed by an OT can be enlightening

Early Interventions (continued)

in conjunction with neurological tests such as visual evoked potentials and may provide more practical information about what and where your child sees best.

Speech Therapy

A speech pathologist can help your child to develop their communication skills and help them to learn safe swallowing of food and drink. Many speech pathologists are able to provide therapy for both domains, or you may choose to see one specialized in communication and one specialized in feeding.

Early assessment and intervention are critical to the development of functional communication. If your child is receiving a diagnosis later in childhood, a speech and language assessment should be arranged as soon as the diagnosis is made. The speech pathologist will work to support your child in the acquisition of the oral-motor skills necessary for babbling and speech. In therapy sessions this involves a lot of singing, reading stories together, and play centered around communication.

To encourage familiarity with and enjoyment of food, your speech pathologist may start by introducing 'tastes' of puree (where a spoon is given a light coating only) or 'munchables' (hard foods such as celery where the child cannot bite anything off). They will also discuss positioning and routines to give your child the best chance to develop these feeding skills. Even if your child feeds through an NG tube or PEG, you will continue to work on swallowing skills so that over time eating by mouth may be an option for some or all nutritional requirements.

Early Interventions (continued)

ABA Therapy

Autism spectrum disorders (ASD) are common among children with SYS. If your child receives a diagnosis of autism, you may wish to seek out a certified practitioner of applied behaviour analysis (ABA). ABA therapy uses positive consequences to reinforce desired behaviors. ABA therapy has an extensive evidence base, and for many children with autism, it is effective in helping them to gain skills that will improve their daily functioning. ABA practitioners may also provide guidance on managing challenging behaviors (although the intent should not necessarily be to suppress all 'autistic' behaviors, as some are important sources of relief for people with autism). Therapy can be intensive, with many contact hours per week, and may involve highly structured or repetitive approaches to learning functional skills. ABA therapy models have been developed for children as young as 12 months old. For very young children, your ABA practitioner may deliver therapy in a play-based way that feels more natural for your child.

Nutrition Therapy

At visits to your accredited dietician or nutritionist, your child will be weighed and their head circumference and body length will be measured so that their growth can be accurately tracked. The dietician will develop a feeding plan to ensure your child is receiving the vitamins and nutrients needed for optimal development, while also considering your child's specific caloric needs. If your child experiences constipation or painful gas, your dietician/nutritionist may be able to advise on dietary adjustments in conjunction with their gastroenterologist.

The detailed growth information collected by your dietician/nutritionist may also be helpful to your endocrinologist when considering growth hormone therapy.

Your child's therapy team

You may notice a lot of overlap between the therapy provided by your therapists, particularly your physical therapist, occupational therapist and speech pathologist. For example, your OT may advise on feeding practices, your physical therapist may introduce songs, and your speech pathologist may make positioning suggestions — but each is bringing their own particular expertise to the task. Some therapists may be open to communicating with each other or even having occasional joint appointments to discuss approaches and share ideas. These are great opportunities to strengthen the support your child is receiving and ensure all therapists are working toward the same goals.

Other Therapies Parents Have Found Useful

While the above therapies are considered the 'gold standard' for children with SYS, some parents have found these additional therapies helpful for their child.

Aqua Therapy (Hydrotherapy)

Aqua therapy is often recommended for strengthening muscles. Sessions may be recommended, or even provided, by your physical therapist. Aquatic therapy is especially beneficial for people who have difficulty with weight-bearing activities. Benefits of aquatic therapy include improved muscle tone and strength, endurance, cardiovascular function, balance, and coordination. Many parents report that their children with SYS enjoy being in the water. The swimming pool may also be an enriching new sensory experience for your child, and group sessions can give an opportunity to meet other children and parents.

Infant Massage

Though not specific to children with SYS or special needs, research shows infant massage is beneficial in improving blood circulation, aiding digestion, enhancing the development of the nervous system, stimulating neurological development, increasing alertness, and improving immune function. Many parents report that their babies with SYS experience gas pain and for some children, abdominal massage may assist (though you should also raise this with your pediatrician, gastroenterologist, or dietician to discuss dietary factors).

View a video demonstration of infant massage and abdominal massage.

Hippotherapy (therapy on a horse)

Hippotherapy is commonly recommended for children ages 2 and above. The horse's rhythmic, repetitive movements work to improve muscle tone, balance, posture, coordination, strength, flexibility, and cognitive skills. The movements also generate responses that are similar to and essential for walking. In addition, adjusting to and accommodating the horse's movements increases sensorimotor integration. You can learn more at the American Hippotherapy Association website.

Growth Hormone Therapy

Short stature is common in children with Schaaf–Yang syndrome. Evidence suggests that similar to PWS, growth hormone deficiency may be a feature of SYS, and people with SYS could benefit from growth hormone (GH) therapy.

In a retrospective study by Hebach et al., GH treatment was observed to increase height, decrease body mass, and increase muscle strength and endurance in children with PWS after 6 months of use. Furthermore, several families also noted additional beneficial side effects such as improved cognition and motor development. Findings from the study suggest GH therapy should be considered a treatment for SYS.

Depending on the country in which you live, access to GH may require meeting stringent criteria. Your child may need to demonstrate low levels of certain hormones and/or a slow rate of growth in order for GH to be prescribed. Additionally, assessment of sleep apnea may be required before commencing treatment, as severe sleep apnea is thought to present a risk during GH treatment. However, you will need to see a pediatric endocrinologist to receive GH, so the first thing to do is to ask your pediatrician for a referral.

Related Videos

FPWR conference presentations by endocrinologist Jennifer Miller M.D.:

Hormone Dosing

Cognitive Benefits

Benefits for Adults

GH Therapy in PWS

Growth hormone therapy has been FDA-approved for Prader-Willi syndrome (PWS) since 2000. It has been shown that in PWS, GH therapy not only significantly increases body height but also improves body composition by decreasing fat mass and increasing lean body mass. More recent studies have shown benefits regarding mental and motor development, as well as an improvement in overall cognition.

Adverse events reported during GH therapy in PWS include worsening of sleep apnea, respiratory tract infections, and peripheral edema.

Reference: A retrospective analysis of growth hormone therapy in children with Schaaf-Yang syndrome (2021)

Getting Support, Getting Involved

Here are some opportunities for you to get and give support in your SYS journey, and to help advance research, awareness, and fundraising—when the time is right for you:

Social Media

Our SYS families use Facebook to connect, make new friends, and exchange support. Join our Schaaf-Yang Syndrome Family Group to connect with others diagnosed with SYS. Visit and like the FPWR Facebook page to stay up to date on PWS and SYS-related research.

SYS Biannual Conference

FPWR's Biannual Schaaf-Yang Family Conference is held in conjunction with our PWS Family Conference and Research Symposium. This powerful experience combines education, networking, and community-building. Learn the latest in Schaaf-Yang research from experts in the field, and have the opportunity to engage one-on-one with the researchers who have a deep interest in finding treatments for our loved ones with Schaaf-Yang. Watch our list of events for details of upcoming events.

Volunteering

FPWR relies on volunteers, like you, to help us accelerate our programs. We invite you to join our team and get involved as we work together to eliminate the challenges of Schaaf-Yang and Prader-Willi syndromes.

Fundraising

Would you like to fundraise for SYS research? 100% of your efforts are earmarked to support SYS-related research. Visit our website to set up a fundraising page or contact susan.hedstrom@fpwr.org to get started. Donations made through our SYS giving page will always be dedicated specifically for SYS-related research. www.fpwr.org/donate-for-sys

Additional Resources

The Foundation for Prader-Willi Research (FPWR)

- www.fpwr.org
- https://www.fpwr.org/about-schaaf-yang-syndrome

Other SYS Organizations

- AESYS: www.aesys.org
- EASYS: http://easys-asso.org/

Dr. Christian Schaaf

- christian.schaaf@med.uni-heidelberg.de
- schaaf@bcm.edu

Video Presentations

- 2020 FPWR Virtual Conference: Presentations by Drs. Schaaf, Wevrick and Potts. Learn more about the SYS research landscape, next steps in SYS research, and the clinical aspects of SYS in these conference presentations.

Peer-Reviewed Publications

- Schaaf-Yang Syndrome Gene Review (2021)
- Schaaf-Yang syndrome overview: Report of 78 individuals (2018)
- The adult phenotype of Schaaf-Yang syndrome (2020)
- Hormonal, metabolic and skeletal phenotype of Schaaf-Yang syndrome: a comparison to Prader-Willi syndrome (2018)
- Expanding the spectrum of endocrinopathies identified in Schaaf-Yang syndrome (2021)
- A retrospective analysis of growth hormone therapy in children with Schaaf-Yang syndrome (2021)

Recursos en Español

- AESYS: Triptico AESYS Separado
- AESYS : Presentacion SYS Noe

We're not just waiting and hoping for new treatments, and a cure, for Prader-Willi and Schaaf-Yang syndromes. We're aggressively doing something about it.

The Foundation for Prader-Willi Research (FPWR) was established in 2003 by a small group of parents who saw the need to foster research that would help their children with Prader- Willi syndrome lead more healthy and fulfilling lives. Today, FPWR is composed of hundreds of parents, family members, researchers, and others who are interested in addressing the many issues related to Prader-Willi and Schaaf-Yang syndromes.

High-quality research will lead to more effective treatments and an eventual cure for these disorders. By working together, we intend to free our loved ones from the burdens of PWS and SYS, allowing them to lead full and independent lives.

Stay in touch! Subscribe to our blog.

FOUNDATION FOR PRADER-WILLI RESEARCH
Working Toward an Independent Future

Foundation for Prader-Willi Research
340 S Lemon Ave #3620
Walnut, CA 91789
888-322-5487