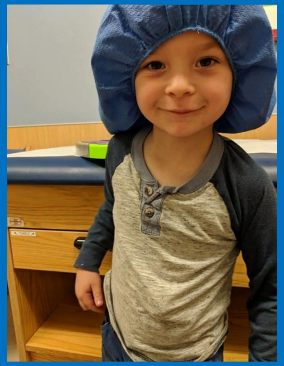
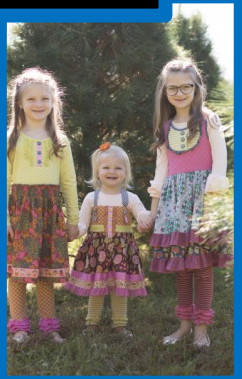
Family to Family: What I wish I'd known about SDS





From our SDS Family to Yours

Dear Parent,

Knowing your child has the diagnosis of Shwachman Diamond syndrome can be scary and overwhelming. We hope that the SDS Registry can help you find the experts to care for your family and connect you to resources that can help.

SDS is a difficult disease, there's no denying that. Most children and adults living with SDS THRIVE, but only with the right combination of medical care, diligence, support, and optimism.

A rare disease like SDS can be isolating. No matter where you are, facing a rare disease can affect your perspective and the way that you experience the world around you. The advice in this book was gathered during a family conference from a group of experienced families who have faced an SDS diagnosis. Many were enrolled in the SDS Registry, but not all. The following pages contain advice from real families, based on of their actual experiences facing SDS. The pictures in this book are of real SDS patients who have become part of our greater SDS family. This booklet was created to help those starting out on this journey, so that they may be more prepared and know that they are not alone. We express our sincere appreciation for the families involved in this project, as there is nothing more powerful than kind words from a family who has already trodden the same path, to one who is just beginning it.

Best,
Kas & Akiko

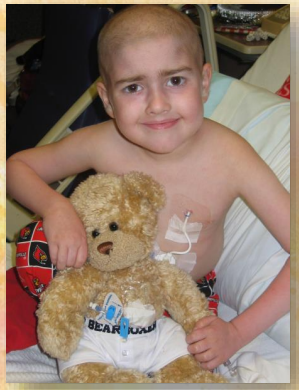
SDS Stories

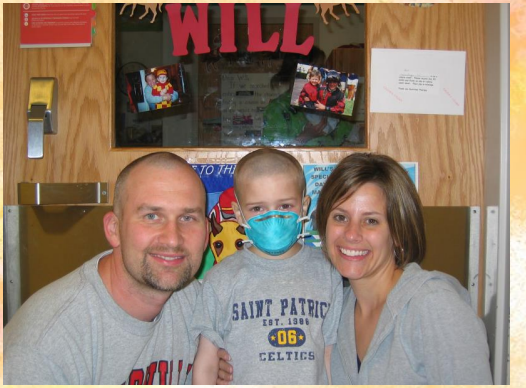
Will

Although symptoms of SDS started when I was 3 days old, I did not officially get diagnosed until I was 3 years old. Some of my symptoms were low counts, infections, and being small.

From the age of 3 on, I had regular follow ups with GI, Hematology, Endocrinology and Orthopedics. And at one point started Neupogen injections. My marrow eventually started to fail and eight days after my 7th birthday, I received a bone marrow transplant from my brother Jeb. My transplant was a success with very few bumps along the way.

Since the transplant, I have had regular follow ups with all of my specialists. I have had several hip surgeries, knee surgery, an appendectomy, gall bladder removal and several spinal surgeries. I also was on growth hormone for a period of time.





What have I learned from all of this ?

First of all, **get used to uncertainty**. Embrace it as you would a roller coaster ride. You don't know when the next fall will be, but hold on tight, scream, cry, laugh, whatever you need to do to get through that ride.

I have also relied on my faith over the years and it gives me comfort during the tough times.

So where am I now?

I am in college with the intent to work in the healthcare field when finished. With all I have gone through, I feel that my calling is to help people. Who knows, maybe I will find a cure for Shwachman Diamond Syndrome one day.

Makenna

Makenna was first put on Hematology/ Oncology's radar at ten weeks old when it was discovered that her hemoglobin levels were critically low. Ten months later, she was tested for SDS and genetically confirmed.

Today, Makenna is a sweet and spunky toddler who loves to play with her friends, play outside, and talk. Makenna's proud parents are Derek, an engineer and Tracy, a teacher. They hope to model a spirit of resilience and grit for Makenna so that she grows to know no limits.

They also enjoy raising money for the Registry because they know it is essential to improving treatments for those affected by SDS; **the Registry gives them hope for a cure!**

Tyler

When Tyler's mother was 20 weeks pregnant-she was told that Tyler might not survive after birth due to his short ribs and small chest. However, Tyler was a tough little guy and surprised everyone. Tyler was officially diagnosed with SDS at 10 months old.

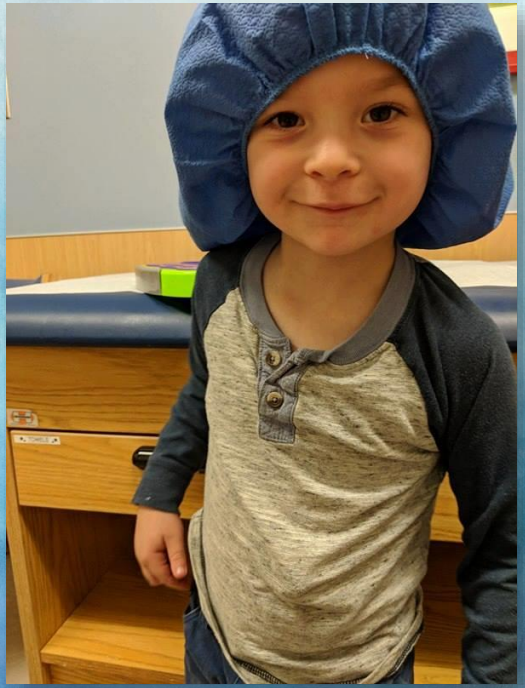
Tyler is now 5 years old and enjoys being a big brother. He doesn't let SDS or the fact that he has a G-tube keep him from being an active kid. Tyler loves running around playing outside with his cousins and little brother and loves to play with his cars.



Nathan

Hi, I'm **Nathan.** What I wish others knew about SDS was how hard it is to look healthy. Every day I receive lots of medicines and I am fed by a feeding tube in my belly. I get shots in my leg every other day to create little army men that defend my body. Without these army men, I get really sick and have to stay in the hospital.

I wish other people knew how hard I try to be a regular kid. I'm usually okay, but sometimes I get really lonely. I wish others could see how sad my sister is sometimes because she feels lonely sometimes too when my mom is with me in the hospital. I wish others could see how super brave I am and that having SDS doesn't scare me.



Scott, Meagan and Jonathan Miller

It's been a journey for the **Miller Family** to reach SDS diagnoses for three of their four children. Scott was diagnosed with pancreatic insufficiency at just 18 months. Initially the diagnosis of SDS was not thought likely because he lacked the SDS mutations, so Scott continued forward. At 6 years old Scott received his first bone marrow biopsy and ultimately reached his diagnosis.

At the same time Scott began his time in hematology/oncology, their son Jonathan, 14 months old, also received a bone marrow biopsy. Jonathan experienced frequent infections leading up to his time of diagnosis but never really presented as a traditional SDS patient.

Meagan, diagnosed with IBS at 3 years old, always had immune system struggles growing up. It wasn't until she enrolled in the SDS Registry, because of her connection to her brothers and their symptoms, that she received her own SDS diagnosis, at 10 years old.

Today, Meagan, Scott and Jonathan are thriving. They attend school and participate in extracurricular activities.

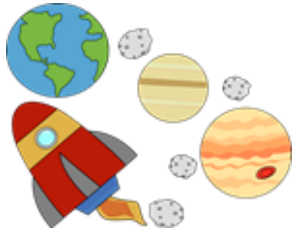
The Millers encourage families to join the Registry because it creates an important community and an understanding that SDS may present differently from patient to patient.

TOGETHER

AJ

Since birth, AJ was labeled as 'failure to thrive'. He spent his first 10 days in the NICU and had his first of several transfusions. He also had 2 bone marrow biopsies before finally genetically confirming Shwachman-Diamond Syndrome several days before turning 5 months old.

We knew the only way to fix the bone marrow issue was transplant and that AJ stood his best chance of doing that before suffering from any illness or infections. So 3 days after diagnosis we were flown to Cincinnati Children's Hospital to start that journey. The team there was/is spectacular. They really helped us take it day by day to focus on getting AJ better and I am so thankful. I truly don't take for granted every moment I get to spend with my cute strong boy!

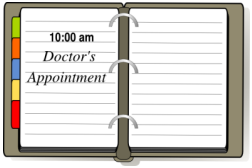


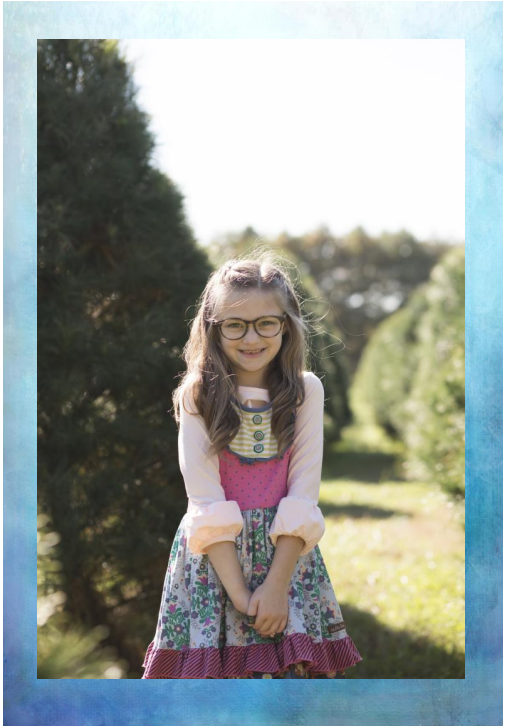
Navigating Clinical Care…

If possible, get your childcare at a **large medical center** so all records are available to all subspecialists and other doctors

Make the doctors talk to your child and each other

Don't be afraid to change doctors if they aren't listening





- Find and go to **the experts**, travel if you have to and are able to. **It's important to have the right medical team**
- **You can seek more than one opinion**
- You can ask for a **genetic test**
- See the **right team** for **bone marrow procedures**
- Put **one doctor** in charge to help with communication
- Have **one hospital** do most of the care if possible

Important Information

- Write down all **medications** and **allergies**
- **Manage information** between doctors
- in a way that works for you (i.e. through an app or a notebook)
- Always have a **safety bag** packed

Ask Questions…

Follow up with the doctors if you are not getting answers

Don't make hasty decisions, **take time to think things through**

Be patient, let information sink in as you learn, you will ask better questions

Ask questions until you understand

HELP AND SUPPORT

- If someone on the **care team** can't provide the answers, ask for someone that can
- Embrace your own ebb and flow, sometimes you want more information and sometimes not
- **Don't be afraid to ask for help** if you need it

Advocate and Educate…

Be your own advocate, trust your gut/parental intuition and if something does not feel right just keep pushing

You can also get an **advocate** to help

Do **research**

Teach your child about their condition so they can one day become their own advocate

Educate your family–it's helpful to have them understand

Practice Self-Care...

Take care of yourself

Continue to live your life, don't put it on hold

Take time to go on vacation and just relax

Add more family time

Take time for your spouse and yourself even if it is only five minutes

Take care of your relationships.

Take one hour a day and indulge your research/tears/read to your kids

Don't be afraid to talk to a counselor if you need it

Connect with other families…

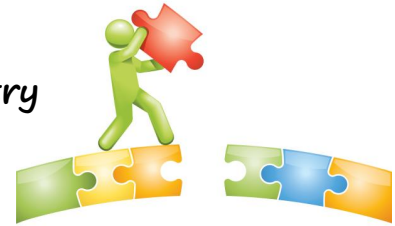
There are great **support groups** and an online community, engage the community

Reach out to other parents, they have been there and understand

Engage in **positive relationships** with other families

It's important for the patient to have a **support system of friends**

Consider enrolling in the **SDS Registry**



Celebrate the Good…

Just because you're different it doesn't define you

Appreciate what you are doing well

Try to make **doctor's visits** and trips to the hospital **as fun and enjoyable as possible**

Make your own normal

Don't focus on what kids can't do and encourage what they can do

Believe in the possibilities, in yourself and that you will persevere

Appreciate the good moments

Be positive and don't dwell too much on the future, embrace uncertainty

Resources

- **Talk with school about resources** that are offered **to accommodate any special needs** the patient may have
- **Family days** offered by institutions
- **Camp Sunshine** allows the kids to experience the community and feel normal

family day

SDS Registry…

The **Shwachman-Diamond Syndrome Registry** (SDSR) is dedicated to accelerating research and treatment for SDS to improve survival and quality of life for all patients with the disease. **The ultimate goal is to cure SDS!** The **SDSR** is centered at Boston Children's Hospital in Boston, Massachusetts and Cincinnati Children's in Cincinnati, Ohio. The SDSR is dedicated to sharing new knowledge with patients, families, and healthcare professionals.

The goals of the **Registry** include:

- Advance research understanding and ultimately cure SDS
- Provide **education** and **resources** to SDS patients, families and the medical community
- **Promote collaboration** among organizations and institutions to improve the lives of patients with SDS
- **Increase awareness** of SDS

SDSR

Shwachman-Diamond Syndrome Registry

We sincerely thank the families and children who generously extended their time, thoughts, words and pictures to this book.

www.sdsregistry.org

Thank you to our community of supporters who have made our outreach and this book possible.

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