

 Share This Email

 Tweet This Email

 Pin This Email



[Visit our Website](#)

[Donate Online](#)



Dear Pamela,

I'm sure you're as happy as I am that Spring has finally arrived. Winter seemed very long this year.

I want to make you aware that two members of our Medical Advisory Board have retired from their medical facilities. Dr. Blanche Alter, who has been with us many years and has been an invaluable medical advisor, has left her position at the NIH. Also, Dr. Garry Tobin has retired as Clinical Director of the Diabetes Center at Washington University Medical Center. Both physicians have thankfully agreed to continue on our Medical Advisory Board.

We had hoped to sponsor a week at Camp Sunshine this summer. We miss seeing all of you. That said, COVID has once again made that impossible. Camp, who always considers the health and welfare of patients first, has made a decision that extreme precautions must be put in place for the safety of all. This will prohibit us from having the large groups for the physicians' presentations. We feel it important that all patients and families have access to all available SDS information to help them deal with SDS. For this reason, we are planning a virtual camp and have asked our medical advisors' help and coordination. This is an ambitious endeavor as we will need to work around the doctors' other commitments. We will definitely keep you updated as plans are finalized.

I ask for continued prayers for all of our patients, especially those who are dealing with difficult problems right now.

My very best to each of you.

Joan Mowery
President
Shwachman-Diamond Syndrome Foundation

COVID-19 Vaccine Information

We have had many questions about whether or not the COVID vaccine is safe for our SDS patients. We asked one of our medical advisors, Dr. Kasini Myers of Cincinnati Children's Hospital and this is her recommendation:

"The SDS registry continues to support vaccination and we are not aware of any particular concerns with the vaccine or complications specific to SDS but of course recommend patients and families discuss any concerns with their primary doctor who knows them best."

So you can feel safe getting the Covid vaccine for your child if they are over 12 and your primary doctor has no concerns. Stay safe out there, everyone!

We Support You!

Please let us know if your child is going to be transplanted. We would like to send them a gift to let them know we care.

CARE PACKAGES
for families going through
TRANSPLANT



Send us an email when you go to transplant
so we can share hope
info@shwachman-diamond.org



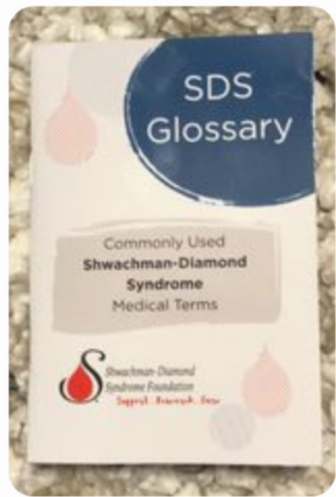
Contact Us

Did You Get Your Copy of the SDS Glossary?



SDS Glossary

An in-depth glossary for families and patients with definitions of the most common Shwachman-Diamond Syndrome terminology



Thanks to a generous community grant this is a **Free Resource**

However, donations to cover shipping costs are appreciated if possible.



Celebrating Our Own

The Superina Family SDS Story

by Lisa Superina



On March 16th, 2020, our sweet, beautiful, Nora Leigh was born. . . . Having three daughters prior to Nora's birth, I was no stranger to motherhood, which is why I began to have some concerns when she wasn't hitting all of her milestones and was having difficulty gaining weight. . . . At Nora's 6 month checkup with her pediatrician, Nora was labeled "Failure to Thrive" Over the next few months Nora had more blood draws and doctor appointments than any baby should. No one had answers for us, and we kept getting referrals to different specialty doctors At two of her ER visits her bloodwork showed that she was neutropenic and the doctors said this was typical for sick babies and did not think anything of it. I was frantic for answers . . . so I dove into my own research. During the day I was a mom and high school teacher, at night I became a doctor and researcher. I spent hours upon hours searching for a diagnosis for my baby. . . . It

was June 28th, 2021, when we finally got answers. . . . Nora's geneticist called with the results of her Whole Genome Sequencing. "We have a diagnosis for your daughter." We didn't know it then, but that one sentence would change our lives forever. With that sentence, Shwachman-Diamond Syndrome, a disease that we had never heard of before, had instantaneously become a significant part of our life. . . . Having a diagnosis for Nora has been a blessing because we now know what we are up against. Like the saying goes, knowledge is power. . . . Sometimes life throws you a curveball. Sometimes life throws you two. Four months after Nora's diagnosis we received a phone call that the genetic testing for my other daughters . . . Kayla, my sweet 3-year-old, chubby faced, happy girl, was positive. What did I miss?

If anyone reading this can take anything out of Nora and Kayla's story, it would be that a mother's instinct is always right. Trust yourself, you know your child. Do the research and don't be afraid to make demands from doctors and ask for the extra tests and bloodwork. If you finally get a diagnosis and it turns out to be a rare disease, like SDS, do more research! Spread awareness, fundraise, and align yourself with like-minded people who have similar goals. You are your child's biggest advocate and you will make a difference in your child's life! Last, but not least, SHARE your story because you never know who is staying up late researching their child's symptoms in hopes of possibly stumbling across something that can help them get answers. You can make a difference.

[Click to Read The Superina's Full Story](#)

All In - Research Update

The ALL-IN campaign was such a successful venture for securing research that will impact every SDS family. Dr. Warren aims to create a drug that will treat Shwachman-Diamond Syndrome at the root of the problem - the ribosome.

Since our initial donation, research has been moving at a rapid pace. Dr. Warren and his team have recently developed a mouse that has advanced research into a drug for SDS therapies. The creation of this mouse model allowed researchers to observe that SBDS may play a bigger role in producing productive ribosomes than we once thought.



Without ribosomes, and the key role that they play in maintaining cellular function, homeostasis would not be possible. Homeostasis is the ability of a cell or body to maintain stable function amidst changing conditions. Proteins are used in cells for everything from creating antibodies and building muscle to facilitating all biochemical processes in the body, as well as transporting the oxygen that keeps cells functioning.

Dr. Warren observed the relation of having mutated or depleted SBDS in the cell and the effect it has on our body creating a working ribosome. By studying flies, yeast, and the mouse model he created to mimic SBDS deficient bone marrow, he is working to create a therapeutic drug that will restore the function of the ribosome.

We can't wait to see this research continue - We are very hopeful for this project to provide therapeutic benefits to every patient affected with SDS!

SDS Live



Thank you everyone for helping make SDSF Live such a success.

Make Sure to Tune in This Summer:

- SDSF Live will continue this year to bring informative and interactive live shows in place of Camp Sunshine lectures since we unfortunately will not be attending camp this year. Stay Tuned!

Our upcoming scheduled shows include:

- June 5, 2022 at 7:30 pm EST - Be the Match
- June 26, 2002 at 7:30 pm EST - Wills

We welcome any suggestions for future shows. Please send any ideas to SDSFLive@shwachman-diamond.org

Click for SDS Live Videos



Registry Remarks



Updates from the North American SDS Registry (SDSR), April 2022

Every patient's experiences and samples hold vital clues to advance our understanding of SDS and to accelerate the pace of research for new treatments. SDS is rare so every patient counts. The North American SDS Registry (abbreviated SDSR) enables patients to share medical information and samples with clinical and laboratory researchers working to cure SDS.

This spring, thanks to survey responses from SDS Registry participants and families in the summer of 2021, the registry has published a new report describing the experiences of patients with SDS with COVID-19 in the journal *Pediatric Blood and Cancer*. This important work is the first to provide information on the experience with COVID-19 for patients with inherited bone marrow failure syndromes.

The key findings of this study were:

- Patients with SDS and COVID-19 reported a short clinical course without significant complications or cytopenias.
- COVID-19 vaccines were well tolerated by patients with SDS with minor side effects.

Additionally, our registry collaborator, Dr. Jane Koo, an Instructor of Pediatrics at Cincinnati Children's Hospital Medical Center was awarded a poster presentation describing a study of neurocognitive outcomes in SDS

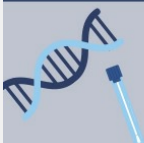
SHWACHMAN-DIAMOND SYNDROME REGISTRY

Directors: Dr. Akiko Shimamura & Dr. Kasiani Myers

WHY IS THE SDS REGISTRY (SDSR) NEEDED?

SDS is a rare condition, so information about SDS is scarce. More information is needed to improve the diagnosis and treatment of children and adults with SDS.

The SDSR provides a way for any patient with SDS or an SDS-like condition to share their information, experiences, and samples with researchers working to find a cure for SDS.



WHAT DOES THE SDSR DO?

By studying medical records and biological samples from the same patients over time, the SDSR works to enhance SDS diagnosis, improve treatment, and advance our understanding of risk factors for severe complications.

Samples for the SDSR are collected when they are obtained for clinical care, so no extra visits or needle pokes are needed.

WILL MY IDENTITY BE SHARED?

Data and samples are de-identified for research. None of your personal information will be shared outside the study team.



WHO WILL HAVE ACCESS TO THE SDSR?

SDSR databases are password-protected behind a firewall and can only be accessed by designated members of the SDSR study team. Only de-identified data and samples are provided to qualified investigators who will study these to improve the lives of people with SDS.

HOW WILL SHARING MY SDS EXPERIENCES AND SAMPLES HELP?

Your information and samples are critically important to advance knowledge about this rare condition with the goal of finding a cure. Here are some examples of ongoing research made possible by the data and samples collected through the SDSR:

- Understand the medical complications of SDS and their progression over time
- Develop strategies to monitor for and prevent complications such as leukemia
- Guide recommendations for medical care to improve the health of people with SDS
- Elucidate the causes of SDS
- Develop better therapies for SDS



For a rare disease like SDS, every patient makes a difference.



HOW CAN I GET INVOLVED?

If you would like to partner with the SDSR in the quest for a cure to SDS, please reach out to our team at SDSRegistry-dL@childrens.harvard.edu.

If you would like to learn more about the SDSR or have any questions about SDS, you are welcome to contact the Registry team.

INTERESTED IN LEARNING MORE?
CHECK OUT OUR WEBSITE:
SDSREGISTRY.ORG



patients from the SDS Registry cohort data. She will be presenting this work at ASPHO (American Society of Pediatric Hematology/Oncology) conference this coming May in Pittsburgh, PA. . . .

[Click the link below to read the findings of this study.]

Questions about SDS can be directed to the SDS Registry either by sending a message through the SDSR website (sdsregistry.org) or by contacting the SDS Registry (SDSRegistry-dL@childrens.harvard.edu). The SDSR team is also available to speak with physicians seeking information about SDS, such as questions about somatic mutation testing or bone marrow exams.

The SDSR continues to contribute to collaborative studies led by other researchers around the world. These are posted on the SDSR website (sdsregistry.org).

[Click to Read the Results of the Study](#)

In Memoriam

Although we fight daily to save the lives of SDS patients and search for a cure, we must acknowledge those for whom a cure did not come in time.



Birthday Celebration!



[Click to Send an Email to SDSF with Your Birthday](#)

Help Us Find a Cure

Gift Card Fundraiser

by Pamela Miller

. . . . I have started a *Gift Card Campaign*. I am asking for people to send gift cards to SDSF that can be used for meals, online shopping, or anything else that could be useful for our families who are facing transplant. This can help alleviate one specific need that the foundation funds. . . .

As many like to have a specific thing to see their donations going towards, rather than just going to a "General Fund," I thought this would be a perfect way to ask for help for SDSF. Many grocery stores offer points for purchases, I know mine can sometimes offer 4x fuel points for gift card purchases. So, the purchase of a gift card will help you and help the Foundation!

Specifically, we can use these gift cards for meals for out of town families who are facing long hospital stays or transplants. . . .

Your gift of a gift card to SDSF can directly help these patients and parents. We have all been in a position where we are overwhelmed, scared, and just beyond our limits. Please help us help our families during these difficult times. . . .

Mail Your Gift Card to:
Shwachman-Diamond Syndrome Foundation
P.O. Box 6723
Florence, KY 41022

[Click to Read More](#)

SDS Awareness Ribbons



The long awaited SDS Awareness Ribbons are custom made double layer ribbons to bring awareness to SDS. Each ribbon will arrive assembled with a gold pin attaching them.

Ribbons are \$5 per ribbon, or 3 ribbons for \$12. Domestic shipping is included.

There is only a limited quantity of these - act fast!

These are great to spread awareness! Fill out the form below to place your order.

[Click to Order SDS Awareness Ribbons](#)

Online Fundraisers

We know that this is a challenging time for so many. We are asking that if you set up an online fundraiser for a birthday or special event or if you order from Amazon Smile, please set Shwachman-Diamond Syndrome Foundation as your charitable organization. Every little bit helps us fund research so help find a cure for SDS!

Welcome New SDS Families

Kosovo
France
Sydney, Australia
Pakistan
Sweden
Mokena, Ill
Lincolnton, NC
Wallalong, Australia
Elgin, Ill
England
Georgia
St Joseph, MO
Pinjarra, Western Australia
Philadelphia, PA
Apex, NC
Barrack Heights, NSW, Australia
Bremen, Germany
Obregon, Mexico City, Mexico

Thank You For Your Donations

Donations received between January 23, 2022 - April 27, 2022

Tommaso Muir
Allackie Gathogo
Robert MacKenzie
Janice Roden
Andrea Peterson
Amber Hutchinson
Shanon La Mar
Kasia Roe
Sarah Wagle
Moran Eary
Wanda Huh
Suzanne Nagy
Tranquilina Rios Winick

PayPal Giving Fund
Your Cause
Facebook Fundraisers
United Way Greater Niagara
Benevity
Frontstream
Blackbaud Giving Fund
Charities Aid Foundation of America
Americal Legion Auxillary -
Madison/Florham Park, NJ
Claire Coleman
Kristin Bellmann
Kerry Campbell
Christine Thompson
Sheryl Homan

In Memory of John Olivo - Ann Friedhofer, Thomas Pearson, Kenneth Funk, Patricia Beer

In Memory of Katie Fugiel - Edward Schida

In Memory of Ethyl Davis - Gerald Deters

In Memory of Alex Sheets - Connie Brewbaker, James Blythe, Sherrie Lussem, Bill Bridge,
Randy Stone, Lana Hatten, Vanderpool Construction, Inc.

In Memory of Nico Hernandez - Felix Hernandez, Angie Hernandez

In Honor of Keller Wilson - JcfKiss, Candace4his, JohncKiss, KWilson0728, Monty Spires,
Lance Spires, Mphilbin, Stacey Spires, Whitney Garrett, Lisa Edick

In Honor of Elizabeth Cole - Melody Davis, David & Camille Abbruzzese Family Fund

In Honor of Henley Johnson - Sheryl Homan

In Honor of Wade Wachter - Community Foundation of the Ozarks

In Honor of Maggie Collins - Sandra Schnell

Special Thank you to the dozens of Anonymous and Unnamed Donors

Special Thank You to the HUNDREDS of donors for our ALL-IN Campaign!!!

Thank you to all those who did Birthday Facebook Fundraisers

The Many Supporters Through Various Other Facebook Fundraisers

Thank you for your monthly donation

Indro Hoffmann
Maria Hall
Keung King Man James
Anthony Porrata
Jason Huttinger

Angela Greiss
Jenny Arnold
Edna Arnold

In Memory of Michele Mowery - Joan & Greg Mowery
In Honor of Camden Kilcrease - Lisa Kilcrease
In Honor of Jonathan and Joseph Wall- Joyce Wall
In Honor of Jake Walden - Julie Walden
In Honor of Wade Wachter - Jennifer Wachter
In Honor of Marissa Avroch - Dianne Moschetta, Rose Cannonico
In Honor of Keller Wilson- Stacy Spires
In Honor of Scott, Meagan, & Jonathan Miller- Pamela Miller
In Honor of Riley Laber - Mary Balint
In Honor of Anthony Del Re- Christian & Gina Del Re

[Donate Today](#)

Ongoing Fundraisers

SDSF would like to encourage everyone to raise funds to support research and the Foundation. We have had families raise money through letter writing campaigns, school dances, corporate matching gifts through their employers, golf tournaments, physical competition events, Super Bowl parties and more.

[CLICK FOR ONGOING FUNDRAISERS](#)

Shop The SDSF Online Store



Did You Get Your SDSF Car Magnet Yet? Don't Have a Car - Put it On Your Fridge or File Cabinet. Spread Awareness - Start a Conversation. Click the Link Below to Get Yours and See What Other SDSF Merchandise We Have! All Proceeds Support Finding a Cure for SDS!

[Shop The SDSF Store](#)

Shwachman-Diamond Syndrome Foundation

FOUNDER: Joan Mowery 1994

BOARD OF DIRECTORS

Joan Mowery, President
Mary Balint, Secretary
Bryan Sample, Treasurer
Christian Del Ré, Communications Chair
Honey Denson, Patient/Family Education Chair
Stephanie Gregoretti, Fundraising Chair
Orva Jurado, Tech Expert
Nicole Shen, Membership Chair
Jenni Wachter, Webmaster
Carolina Cordova
Michelle Grenell
Savannah Lillywhite
Scott Miller

Pamela Miller, Executive Director

Other Amazing Leaders

Joyce Wall - Anna Angel Basket Coordinator
Maura Donahue - Advisor to the Board

ATTORNEY: Ann Bodewes Stephens, Herzog Crebs



Share This
Email

Have an article, update, or fundraising activity you would like to share in a future newsletter? Email your story to:

christiandelre@shwachman-diamond.org.

Sign Up For
Emails

We would love to hear from you and share your stories!

[Website](#) · [Family Support](#) · [Research](#) · [Fundraising](#) · [News & Events](#) · [Resources](#)



**Shwachman Diamond Syndrome
Foundation**

888-825-7373

info@shwachman-diamond.org
www.shwachman-diamond.org

Contact Us

Shwachman-Diamond Syndrome Foundation | P.O. Box 6723, Florence, KY 41022

[Unsubscribe](#) info@shwachman-diamond.org

[Update Profile](#) | [Constant Contact Data Notice](#)

Sent by info@shwachman-diamond.org powered by



Try email marketing for free today!