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Dear *|FIRST_NAME|*,

We were happy to see some old friends at Camp Sunshine and meet some new families. As always, a special thanks to our medical team for giving of their time to come to camp and educate us. We also discovered that they can sing too.

We are happy to welcome some new members to our SDSF Board of Directors. Shari St. Hiliare, John Wall, and Tom Dahlman. All are SDS parents. John has retired from his business as a PT and orthotics expert, Tom is the Rector of Emmanuel Episcopal Church, and Shari has been helping us for some time with her design skills.

We also wish to welcome Daniel Leung, MD to our Medical/Scientific Advisory Board. Dr. Leung is a specialist in gastroenterology, hepatology and nutrition at Texas Children's Hospital.

We are happy to have them and look forward to their help.

The patient care manual, which the Registry has been writing, is in the proofreading stage. We hope to be able to print and distribute it within the next few months. It will be a great help in ensuring all SDS patients receive optimal health care.

Summer is coming to an end and school is restarting. While I'm sure parents are happy about this, the kids probably not so much. We have recently sent you some information regarding school. I hope you have all had a chance to read it.

Wishing you all a happy Autumn.

SDS Strong,

Joan Mowery
President
Shwachman-Diamond Syndrome Foundation

How to Communicate About SDS and Our Children's Needs?

by Christian Del Ré



The start of a new school-year can be a time of anxiety in any home. Kids are nervous, parents are nervous, and between battling to get back into school routines, we also have to worry about whether or not our child's teacher knows what SDS is and what special needs have to be met each day . . .

For the past seven years my wife has scheduled an appointment with the school principal, and before the start of the year, she meets with the administrative team as well as Anthony's teachers to give them the SDS 101 talk and to go over what special considerations Anthony needs while in school

I have attached some of the documents we use each year; feel free to adapt and edit them for your own family. Here is also a check-list of important things you may want to do before the start of the school-year

[Click to Read More & See Check List](#)

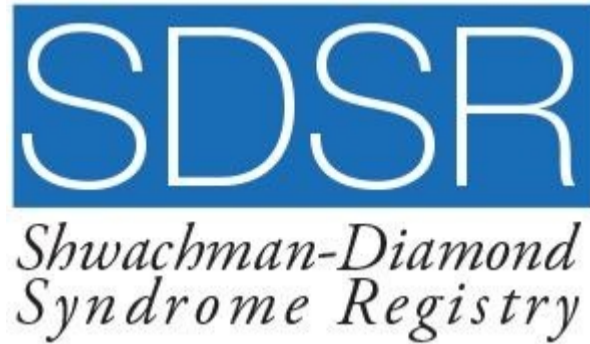
[All Email Blasts \(/emails\)](#)

[School Letter](#)

SDSF Info Packet

Medication List Template

Registry Remarks



The SDS Registry attended Camp Sunshine in July and had a wonderful time meeting and connecting with families! The SDSR would like to share the informative handouts from our physicians' talks at Camp Sunshine. Click the buttons below to access the handouts.

If you are interested in joining the SDS Registry, or if you have already joined the SDS Registry and want to send in new blood/bone marrow samples or new medical records, please visit our website at sdsregistry.org or reach out to our team via email at SDSRegistry-@childrens.harvard.edu.



Hematologic Complications in SDS Handout

Hematologic Issues in SDS and their Management

Blood complications
The types and timing of blood complications can vary in different patients with SDS and require different treatments.

Blood complications can include:

- Low blood counts
 - May need transfusions
 - Low hemoglobin - red cell transfusions
 - Low platelets - platelet transfusions
 - Low neutrophils - G-CSF
- Bone marrow failure (BMF, severely low blood counts)
 - May need bone marrow transplant
- Myelodysplastic syndrome (MDS)
- Acute myelogenous leukemia (AML)

MDS/AML

- MDS is a pre-cancerous (leukemic) condition that is caused by abnormal blood cells
- MDS is characterized by the development of
 - Abnormal cells,
 - abnormal chromosomes or
 - Mutations of genes in the bone marrow often with lower or changes in blood counts
- Leukemia is a cancer of white blood cells - most commonly AML in patients with SDS
- MDS/AML in patients with SDS is difficult to treat
 - Standard therapies typically do not work or are too harmful

Bone Marrow Surveillance

- You CANNOT wait for symptoms of MDS/AML to develop
- MILD OR NORMAL BLOOD PROBLEMS DOES NOT EQUATE TO LOW RISK OF MDS/LEUKEMIA
- Bone marrow surveillance is recommended as blood surveillance does not assess for abnormal changes in marrow cells, chromosomal abnormalities.
- We also do not know the sensitivity of blood surveillance in detecting abnormalities.

Surveillance Recommendations

- Blood counts
 - Approximately every 3-4 months (quarterly)
- Bone marrow examination should include
 - Morphology (how cells look)
 - Cellularity (marrow structure)
 - Cytogenetics (chromosome abnormalities)
 - FISH (specific chromosome abnormalities)
 - Flow cytometry (cell maturation patterns)
 - Loss of heterozygosity (LOH)
 - Microarray
 - Somatic mutation analysis
- Could defer bone marrow exams until 2 years old if blood counts are not concerning
- Bone marrows are typically performed annually

Bone Marrow Transplant for SDS

- Indications for BMT include:
 - Very low blood counts (aplastic anemia/BMF)
 - Need for regular transfusions
 - Recurrent infections despite G-CSF
 - MDS/AML
 - High-risk features with expert consultation
- BMT for Patients with SDS
 - BMT for BMF have good outcomes with expert care
 - Patients with SDS and MDS/AML have worse outcomes compared to patients with BMF likely due to:
 - More sensitivity to standard chemotherapy
 - Relapsed disease
- Decision to undergo BMT should be made with providers experienced with treating patients with SDS

SDS GI Issues Handout

COMMON GI AND PANCREAS ISSUES IN SDS AND TREATMENT STRATEGIES
AMIT GROVER, MD

ISSUES WITH THE INTESTINE IN SDS

- More than half of patients with SDS have abnormalities by endoscopy

GROWTH ISSUES

Growth can be impacted in patients with SDS due to:

- Feeding difficulties
- Impaired digestion
- Poor absorption in the intestines
- Lack of essential fats/vitamins/minerals
- Impaired liver function
- Endocrine abnormalities
- Behavioral feeding issues

LIVER ISSUES IN PATIENTS WITH SDS

Abnormalities in infants and young children may include:

- Liver enlargement
- Abnormal liver function tests

Other abnormalities include:

- Chronic liver disease
- Scarring (cirrhosis) of the liver

Long-term outcomes related to the liver are not known.

Many patients with liver issues may be asymptomatic.

Current guidelines recommend:

- Follow liver function tests annually or more often as indicated

CURRENT APPROACH CONSIDERATIONS

- Try to identify GI providers with experience in seeing more than one with SDS or expertise in pancreatic disease
- Multi-disciplinary management is important and may include providers from these disciplines:
 - GI
 - Hematology
 - Nutrition
 - Feeding specialist
 - Endocrinology
 - Psychology
 - Social Work
 - Genetics

GI SPECIFIC ASSESSMENTS

- Age-relevant pancreatic testing
 - Isoamylase >3 years old
 - Trypsinogen <3 years old
- Liver function testing
- Fat soluble vitamins (ADEK)
- Nutrition consultation should be considered early on

GI SPECIFIC THERAPIES

- Pancreatic enzyme supplementation
 - Coated or uncoated formulations
 - Oral, NG-tube feeds, G-tube feeds
 - Dosing is based on age
 - Ensure enzymes are being administered correctly
- Vitamin supplementation
 - ADEK
- Feeding issues
 - Consider seeing feeding specialist
- Behavioral issues/oral aversion
 - Consider seeing behavioral specialist or feeding therapist

Also remember patients with SDS can also get common GI issues including:

- Constipation
- Abdominal pain
- Irritable Bowel Syndrome
- Acid reflux
- Lactose intolerance

Neurocognitive Considerations Handout

Neurocognitive Considerations in SDS

How to find a neuropsychologist

- Talk to your primary care provider
- Local children's hospital
- State child development center

What can I do for academic concerns?

- Meet with school team
 - Talk about an educational plan, such as IEP or 504

What can I do for executive or attention concerns?

Books to consider:

- ADHD
- TEEN
- ADHD

What can I do for mood or behavioral concerns?

- Consider therapy
 - Individual and/or family-based
 - Psychology Today
 - <https://www.psychologytoday.com/therapists>

When should evaluations occur?

- At regular intervals throughout childhood:
 - Prior to or during kindergarten
 - At key transition points (3rd/4th grade, middle school, high school, early adulthood)
 - During periods of relative medical stability
- Other times to seek evaluation:
 - Bone marrow transplant
 - Neurological changes (seizures, head injury)
 - Difficulties in real life (school, home)

Plan Common accommodations include preferential seating, extended time, reduced assignments

- May need specialized instruction or support from an intervention specialist

Helpful resources:

- Wrightslaw (<https://www.wrightslaw.com/>)
- Your state's department of special education

Other resources:

- Project Learnet (<http://www.projectlearnet.org/>)
- Harvard University's Center on the Developing Child (<https://developingchild.harvard.edu/guide/a-guide-to-executive-function/>)
- Children and Adults with Attention Deficit/Hyperactivity Disorder (<http://www.chadd.org/>)
- National Resource Center on ADHD (www.help4adhd.org)

Other online resources and books:

- American Academy of Child and Adolescent Psychiatry (<http://www.aacap.org/>)
- National Institute of Mental Health (<https://www.nimh.nih.gov/health/topics/child-and-adolescent-mental-health/>)
- National Library of Medicine's Medline Plus (<https://medlineplus.gov/childmentalhealth.html>)

Psychosocial Corner: Open Dialogue

We are happy to introduce a new component to the SDSF e-Newsletter, The Psychosocial Corner, in which Nancy Cincotta, LCSW, MPhil will write a regular column. Nancy has been connected to the SDS community for many decades, having met our Founder Joan Mowery in the early 1990s.

Ms. Cincotta has become a fixture in the inherited bone marrow failure and childhood cancer communities and has witnessed the growth of information and understanding of these conditions for over forty years. Nancy received her first Master of Science degree (the one in Social Work) in 1979, from Columbia University, after having been a Child Life Specialist. Her career has been focused on the issues families face when they have a child diagnosed with one of these illnesses.

Nancy brings with her a remarkable professional commitment to our population, with a unique understanding of the specificity and scope of the journey encountered in dealing with Shwachman-Diamond Syndrome. One of her interests is exploring avenues of support that enable individuals to adapt and thrive within the context of the SDS journey, understanding what is unique to SDS and what is similar to other illness communities.

Nancy has been both hospital and community-based and has collaborated with a number of illness-related non-profit organizations, working with individuals, families, and groups, and in retreat settings. She has always been interested in hope as a concept in inspiring individuals and their family members as they face unique medical challenges.

Many of you have already met Nancy at regional conferences, at Camp Sunshine, on Zoom, or at meetings held with Dr. Shimamura. We are fortunate that, as she has transitioned out of her role as Psychosocial Director at Camp (after 35 years), she has made an even stronger commitment to her work with the SDS and inherited bone marrow failure communities. She is currently on the SDSF Medical Advisory Board and has been involved in educational, and SDS Congress meetings. She has a particular interest in how individuals cope and live with SDS, issues siblings face, educational issues, transitioning to adulthood, and the impact of a lifelong illness on affected individuals and those who love them.

In her column, Nancy will focus on topics that of interest to those with SDS and their family members. At the same time, she would like to develop a psychosocial research component that examines issues the SDS community over time. Please reach out to her (nancycincotta@gmail.com) with topics you are interested in. The inaugural column of the Psychosocial Corner: *Open Dialogue* will appear in the next SDSF newsletter. We are grateful for Nancy's ongoing commitment to the SDS community, and we look forward to our future endeavors.

"SDS has been an illness of interest to me since I first met Joan Mowery. Her enthusiasm and commitment have inspired mine. Watching people on their journey grow up with SDS, I have come to appreciate how complex an illness it is. It is with great anticipation that I move forward to getting to know you in these new forums. I am honored to be able to make an ongoing commitment to the SDS Foundation" (Nancy Cincotta).

Celebrating Our Own A Mother's Intuition

by Brittany Bryant



I always knew that something wasn't quite right with my unborn child. . . . I couldn't put my finger on it, but I had this pit in my stomach from the moment he was conceived. . .

Jayden was born 3 weeks early . . . in January 2020. He spent 10 days in the NICU due to

low blood sugar and feeding difficulties. He was born weighing 4 lbs 15 ozs. The assumption was once he was born he would catch back up without issue. Except he wasn't...in fact he wasn't growing. He would "spit up" his formula. . . . At 10 weeks he was hospitalized with failure to thrive.

While he was in the hospital the doctors ran all sorts of labs and tests and thought he had cystic fibrosis. However, those tests were all negative. . . .

It seemed every month or two he would get sick. Starting with colds, then RSV, followed by croup and then ear and sinus infections. In October 2020, during his bout of croup I noticed he had a rash of purple dots scattered on his chest. . . . This time his pediatrician decided to send him for labs. His CBC came back and everything was low. . . .

In February 2022, we had our first appointment with the hematology department. The doctor thought it was just his antibodies attacking due to being sick. However, those tests were negative. Next appointment he ran more labs, again all negative. It was finally decided that in April 2022, Jayden should have a bone marrow biopsy to determine what was causing the low blood levels. The bone marrow biopsy showed he had bone marrow failure but not why. The doctor recommended Jayden get a genetic test that would tell us what was causing the bone marrow failure. . . .

August 26, 2022, is a day the world stood still for me. The doctor explained to me that Jayden had not only one rare genetic bone marrow failure disease but two. He was diagnosed with Shwachman-Diamond Syndrome and RUNX1. We were told these diseases predispose him to leukemia . . . My husband and I were so scared and spent the next days crying and in fear. However, the pit of worry in my stomach finally disappeared. I had always known something was wrong and once we had the diagnosis that worry was gone. Now the worry became a different kind of worry.

Luckily, I came across The Shwachman-Diamond Syndrome Foundation website that had resources and information on the registry and which doctors to see. Without hesitation or delay I booked a consult with Dr. Myers at Cincinnati Children's. . . .

It was October 25, 2022, and for the first time he said the words to us we didn't want to hear, Stem Cell Transplant. . . . On June 28, 2023, Jayden received his bone marrow transplant. I am happy to say that he is now free of SDS and RUNX1 in his cells. . . .

For a 3 year old having to go to as many doctor's appointments and be hospitalized for such a long period of time, he has handled it all so well. We look forward to returning home and knowing he has new healthy marrow.

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

SDSF Live



Our past SDFS Videos are available on line. Click the button below for our library.

[Click for SDFS Live Videos](#)

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

Shwachman-Diamond Syndrome Foundation
Support...Research...Cure

All Email Blasts (emails)

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.



Help Us Find a Cure



Click the button below to access the SDSF Swag Shop. We have a wide variety of SDSF merchandise to choose from.

[Visit the SDSF Swag Shop](#)

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, please set Shwachman-Diamond Syndrome Foundation as your charitable organization. Every little bit helps us fund research to help find a cure for SDS!

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.

All Email Blasts (emails)
Welcome New SDS Families

Jiangning, Nanjing, China
 Yamunanagar, India
 Dublin, Ohio
 Villeurbanne, France
 Forest Hills, New York
 Kyneton, Victoria, Australia
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Thank You For Your Donations

Donations received between April 30, 2023 - August 15, 2023

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Lorraine Lagesse	Give Lively Foundation
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In Memory of Michael Billings - Linda Azzarello, Pamela Pauline, Brooke Goldin, Gena Wade, Terry Billings

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In Memory of Robert Jenkins - Susan Butsch, Carol Walker, Lois Dickinson

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In Honor of Jordyn Jones - Mrinmoyee Das

In Honor of Logan Stone - Susan Utz, Matthew Stone

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In Honor of Kaitlyn Sampson - Marzena Fusilier

In Honor of Michelle Pugliares - Lorene Pugliares

In Memory of Jacob Reitenauer - Vickie Reitenauer

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The Many Supporters Through Various Other Fundraisers

Thank you for your monthly donation

All Email Blasts (7 emails)

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