# SHWACHMAN-DIAMOND SYNDROME REGISTRY 2025 SUMMER NEWSLETTER

## Progress and News

**Living with SDS Let you voice be heard!**

**New email communication option**

SDSR

Shwachman-Diamond Syndrome Registry

**Advances and Progress presented on at the 2025 SDS Scientific Congress**

# LIVING WITH SDS

## MAKE YOUR VOICES HEARD

The SDS Registry is inviting input from individuals with SDS as well as parents caring for children with SDS in order to better understand the patient and family experience of SDS. By hearing directly from patients and families, rather than an interpretation of a patient's condition by a medical provider, the SDS Registry aims to better understand the patient and family experience of SDS and its associated treatments. The goal is to improve medical care and to inform research priorities to align with areas of importance to patients and families. Knowledge gained from this study will improve partnerships between patients and medical providers for shared medical decisions that better align with patient and caregiver values and preferences.

This study is open to participants in the SDS Registry and their caregivers. The average time to complete the survey is 30 minutes.

An email from SDS Registry research assistant Elizabeth Korn with a link to the online survey for this study is being sent to all participants on the SDS Registry. The email will be sent via a secure link with the subject title "SDS Updates from Boston Children's Hospital". If you want to confirm that the email is a legitimate link from the SDS Registry, please call or email the SDS Registry at the email or phone number below.

Please reach out to the SDS Registry team if you have already joined the registry and have not received the link for the study. If you are not already in the SDS Registry and would like to participate, please contact the SDS Registry.

**SDS Registry Email**: SDSRegistry-dL@childrens.harvard.edu
**SDS Registry Phone**: 617-919-1574

SDSR
*Shwachman-Diamond Syndrome Registry*

## Want to receive non-secure emails?

**The SDS Registry now has a new option to opt-in to non-secure emails for easier access. If you would like to change your communication preferences, please reach out to our team or be on the lookout for any calls or secure emails asking about this updated preference!**

# Advances and Progress from the SDSR

## Hot off the press!

SDS Registry Publication in the Journal of Pediatrics: Dr. Jane Koo led an SDS Registry study characterizing growth patterns in Shwachman-Diamond Syndrome. We studied growth after bone marrow transplant as well as the effects of growth hormone for patients with SDS. These results were shared at the 2025 SDS Scientific Congress. A short summary is presented below.

To read a full version, follow this link: https://www.jpeds.com/article/S0022-3476(25)00321-X/fulltext.

# Presented at 2025 SDS Scientific Congress

**Research from the SDS Registry contributed to 11 presentations on research progress advancing diagnosis and medical care for SDS at the 11th International SDS Scientific Congress this past June. Dr. Kasiani Myers chaired the meeting and opened the meeting with a patient family interview.**

## Surveillance and Timing of Transplant in SDS -presented by Dr. Akiko Shimamura

**Dr. Akiko Shimamura discussed data from the SDS Registry and others to guide evidence-based recommendations for leukemia surveillance and risk stratification. Dr. Shimamura reviewed the recommended tests for annual surveillance to identify patients at high risk of progression to leukemia, and outlined indications for referral to a hematopoietic stem cell transplant center experienced with SDS.**

Funding support from the National Institutes of Health

## Transplant for SDS and Inherited BMF - presented by Dr. Kasiani Myers

**Bone marrow transplant currently remains the only curative treatment for severe hematologic disease in patients with SDS but is a challenging therapy. The decision to undergo BMT should be made with a team experienced in SDS as SDS specific transplant approaches are important to achieve good outcomes. Recent advances in transplant approaches for SDS have improved outcomes for those transplanted for bone marrow failure, but there is still work to be done to improve outcomes for those transplanted for MDS or leukemia. At the Congress the registry shared promising early data on improved transplant outcomes of patients transplanted for high risk disease (when they are showing early signs in their bone marrow testing of higher risk of developing MDS or AML).**

Funding support from the National Institutes of Health

## Creating specific growth charts for children and Young Adults with SDS - presented by Dr. Jane Koo

**We created growth charts using data from 127 patients with genetically confirmed biallelic mutations in SBDS, which showed decreased height and weight compared to the general population however had conserved body mass index compared to the general population. In a small subset of patients on growth hormone, there was no overall change in height velocity following initiation. Patients who received bone marrow transplant also had slower growth overall after bone marrow transplant.**

Funding support from the National Institutes of Health

## Shwachman Diamond Syndrome: Tackling a Rare Disease - presented by Dr. Akiko Shimamura

Dr. Shimamura was invited to present an overview summarizing past and current advances in our understanding of SDS, and discussed future areas of high priority for scientific and clinical research. She highlighted the critical role of patients/families participating in the SDS Registries internationally which make this progress possible.

## Gastrointestinal and hepatic manifestations in individuals with SDS - presented by Dr. Jane Koo

We found liver injury as defined by elevations in liver enzymes was very common in a large cohort of patients with genetically confirmed biallelic mutations in SBDS.

Funding support from the National Institutes of Health

## Non-Invasive surveillance strategies in SDS -presented by Dr. Felicia Anderson

We analyzed paired blood and bone marrow samples from SDS patients to assess whether sequencing results are comparable. All TP53 mutations at clinical detection levels were detected in paired blood and marrow samples. Ongoing analyses aim to determine if blood is a reliable source for somatic genetic testing and if it can be integrated into the surveillance strategies in SDS.

Funding support from the National Institutes of Health, the SDS Foundation, the Olivo Fund, and Ray of Hope

## Addressing Neuropsychological Needs in SDS - presented by Dr. Thea Quinton

Dr. Thea Quinton provided an overview and update of neurocognitive and behavioral challenges and resources for patients and families with SDS. For more information, see prior webinar on the SDS Registry website: sdsregistry.org

Funding support from the National Institutes of Health

## Adult phenotypes of SDS - presented by Dr. Chris Reilly

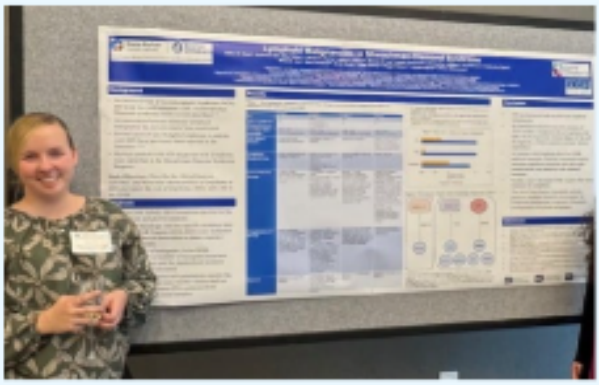
Patients with SDS have a high risk of developing blood cancer, and this risk increases with age. Patients do better with transplant when performed prior to developing MDS or acute leukemia. Using blood mutation sequencing is a powerful tool to help determine if and when patients should be considered for a stem cell transplant.

Funding support from the National Institutes of Health

## Genetic Re-evaluation of SDS-like and Neutropenia Conditions - presented by Dr. Helen Reed

**A subset of individuals with a clinical diagnosis of Shwachman-Diamond syndrome do not have an identified genetic mutation. In a cohort of >100 individuals with SDS-like conditions or severe neutropenia, 33 had a candidate gene identified through whole exome or whole genome sequencing, some in genes not associated with SDS or severe congenital neutropenia.**

Funding support from the National Institutes of Health



## Leukemia Risk in SDS: Results of an International Collaborative Study - presented by Dr. Jean Dondieu

**Dr. Jean Donadieu presented an interim analysis on behalf of the international collaborative project organized by the SDS Registry to investigate the lifetime risk of MDS and AML in SDS. Subsequent to the talk, this analysis is now nearly complete and will be submitted for publication in the near future.**

Funding support from the National Institutes of Health

## Infection Profile in Children with Shwachman-Diamond Syndrome and Low Absolute B-Cell Count - presented by Dr. Nicholas Gloude

**We conducted a retrospective study to characterize immunologic function in patients with biallelic mutations in SBDS enrolled on the SDS Registry. Overall, the results indicate limited immune deficits, largely quantitative and qualitative changes in the B cell compartment, in a small subset of patients with SDS. Patients with SDS with low B cells may be at increased risk for sinopulmonary or skin and soft tissue infections particularly if they also have low IgG levels. Initial immunological evaluation including a complete blood count, lymphocyte subpopulations, and immunoglobulin levels may be beneficial to check in patients with SDS. However, it is reasonable to reserve a more detailed assessment for patients with recurrent or serious infections.**

Funding support from the National Institutes of Health

## Gene Therapy for SDS - Presented by Dr. Dan Bauer

**In collaboration with the SDS Registry, Dr. Bauer shared exciting advances from his lab which has developed a specific and efficient gene editing method to correct the most common disease-causing mutation in the SBDS gene. He outlined steps to open a clinical trial with the goal of improving blood cell production and reducing the risk of leukemia.**

Funding support from the National Institutes of Health

## Clonal Evolution and TP53 In SDS -Presented by Dr. Alyssa Kennedy

**Clonal hematopoiesis is common and persistent in patients with SDS. The clonal expansion can be maladaptive, such as TP53 mutations, or adaptive as is the case in EIF6 mutations. TP53 mutations are screened for in yearly bone marrow biopsies and if these mutations are found, your doctor may monitor these mutations and blood counts more frequently.**

Funding support from the National Institutes of Health

## Modeling IBMF on a Bone Marrow Chip - Presented by Dr. David Chou

**The human bone marrow chip recreates the maturation of multiple human blood cell types in a microfluidic chip. Our studies have shown that it may help identify whether drugs in early stages of development are effective. Furthermore, bone marrow chips made from SDS patient cells are able to model the hypocellularity and altered blood cell development observed in patients, so they may be useful models for testing new candidate drugs for SDS.**

Funding support from the Natinal Institutes of Health, FDA, and BARDA

# Other Registry Presentations

**Dr. Kasiani Myers and Dr. Akiko Shimamura gave invited oral presentations at the American Society of Pediatric Hematology/Oncology meeting in Louisville KY and at the MDS 2025 meeting in Rotterdam, Netherlands to share data from the SDS Registry informing specific tests for surveillance in SDS, patterns that identify patients at high risk for progression to leukemia, and outcomes of transplant for patients with high-risk features.**

## *Special thanks!*

**Thanks to all our generous donors who have supported these studies with donations to the SDS Registry. If you would like to support SDS Registry studies, please note that only funds submitted through this specific link are received by the SDS Registry: www.sdsregistry.org**
**The yellow donation button is in the upper right corner.**