

What is the Shwachman-Diamond Syndrome Registry (SDSR)?

The SDSR is a research organization dedicated to developing better treatments for and improving the health of patients with Shwachman-Diamond Syndrome (SDS) and Shwachman-Diamond-like (SDS-like) syndromes. The Registry works to accelerate SDS research by collecting and studying clinical information and samples from patients and their family members, facilitating high-impact research collaborations, and providing education. The SDSR's ultimate goal is to cure SDS!

Who Can Join the SDS Registry?

Patients who have been **clinically or genetically diagnosed** with SDS or who are **suspected to have** SDS or an SDS-like syndrome and/or their family members are eligible to join.

Why Should I Join the SDS Registry?

SDS is a rare disease, and medical information is scarce. By participating in the Registry, you will be contributing invaluable medical information about SDS and SDS-like syndromes.

What Do I Need to Do to Participate in the SDS Registry?

1. Contact the SDSR at SDSRegistry-dl@childrens.harvard.edu /617-919-4227 for Boston Children's Hospital or sarah.loveless@cchmc.org or 513-803-7656 for Cincinnati Children's Hospital to learn more about the Registry and review the consent form with a member of the study team.
2. Return the original, signed consent form to the Registry. Your participation does not begin until the SDSR receives the original, signed consent form.
3. Sign a medical release form to allow the SDSR to obtain clinical reports and laboratory test results from your providers. The Registry will use this medical information to gain insights into SDS and SDS-like syndromes.
4. Because SDS is poorly understood, the SDSR collects blood and bone marrow samples for research. The Registry collects research samples at the same time as clinical testing, so there are **no extra needle pokes or procedures**.

What if I Am Already a Part of the SDS Registry?

The Registry encourages current participants to:

- **Reach out a month before any bone marrow procedure** to arrange for a specimen collection kit.
- **Check in periodically** to ensure their medical and contact information are up-to-date.

Current participants can contact the SDSR at SDSRegistry-dl@childrens.harvard.edu or 617-919-4227 if you are followed by Boston or sara.loveless@cchmc.org or 513-803-7656 if you are followed at Cincinnati.

What Has the SDS Registry Accomplished, and What Is It Working On?

- Studies made possible by the SDSR have **led to better ways to identify and monitor patients with SDS and SDS-like syndromes.**
- The Registry is extensively engaged in ongoing SDS research, including research into:
 - Low blood counts
 - Developing better treatments for low blood counts
 - Myelodysplastic syndrome (MDS) and leukemia in SDS
 - Understanding why leukemia develops in SDS
 - Developing better tests to monitor for impending leukemia
 - Developing effective treatments for MDS and leukemia
 - Bone marrow transplant in SDS
 - Developing less toxic transplant regimens
 - Medical complications in SDS, including gastrointestinal, immunologic, endocrine, and skeletal complications
 - SDS in adults

The SDSR is hosting an SDS Family Day on Sunday, September 15, 2019 at Boston Children's Hospital. SDS families are invited to hear updates from the Registry, learn about exciting research, discover cutting-edge clinical trials, and connect with other families.

Together, we can beat SDS!