



APRIL 2022

HELLO FROM THE SDSR!

Shwachman Diamond Syndrome Registry

Recent Updates from the Registry:

DR. AKIKO SHIMAMURA & DR. KASIANI MYERS

1) New tests to monitor for leukemia risk

Somatic mutation testing is recommended as part of bone marrow surveillance for people with SDS, with special attention to TP53. TP53 mutations by themselves do not necessarily mean that a transplant is recommended, but this provides important information to assess leukemia risk. Research from the SDS Registry has discovered that people with only mildly low blood counts remain at risk for leukemia, so regular marrow evaluations are recommended. However, a current research focus of the SDSR is investigating whether somatic mutation testing of the blood can reduce the required frequency of marrow examinations.

2) SDS Registry investigators, as well as other research groups, are developing potential therapies targeting EIF6 to treat SDS.

Work from the SDSR identified frequent acquisition of EIF6 mutations in the bone marrows of patients with SDS and demonstrated that these EIF6 mutations rescued the ribosomal abnormalities in SDS and improved blood production. Subsequent work from another research group led by Patrick Revy, Alan Warren, and Jean Donadieu and colleagues in Europe found similar results. These results clearly show that loss of EIF6 is a mechanism driving selection of del20q clones studied in depth by Roberto Valli, Emanuela Maserati and colleagues in Italy. Several research groups, including SDS Registry investigators, are working on developing potential therapies targeting EIF6 to treat SDS.

3) The SDSR continues to support collaborative studies led by other researchers around the world.

Updates are posted on the SDSR website (sdsregistry.org).

How you can help:

JOIN THE SDS REGISTRY

You can join from anywhere. Contact the SDS Registry to learn how to join.

SEND IN SAMPLES OF BLOOD AND MARROW

Only when they are being obtained as part of clinical care. SDS Registry coordinators are happy to work with you to facilitate this.

SHARE YOUR MEDICAL RECORDS

Including updates every year. If you have records available, you can send them directly to the SDS Registry. If you don't know how to obtain your records, SDS Registry coordinators are available to help.

SDS is rare, so every patient counts!

TO GET IN TOUCH, CONTACT: SDSREGISTRY-DL@CHILDRENS.HARVARD.EDU
OR CHECK OUT OUR [WEBSITE: SDSREGISTRY.ORG](https://sdsregistry.org)

SDSR
Shwachman-Diamond
Syndrome Registry