

What is Shwachman-Diamond Syndrome (SDS)?

Shwachman-Diamond syndrome (SDS) is an inherited rare disease that affects many parts of the body, particularly the **bone marrow**, **pancreas**, and **skeletal system**. As a bone marrow failure disorder, it puts patients at high risk of life-threatening complications such as serious infections (sepsis), aplastic anemia, myelodysplastic syndrome (MDS), and acute myeloid leukemia (AML).

We estimate that about 2,000-3,000 people have SDS in the United States, and a similar number in Europe, many of them un- or misdiagnosed. The estimated incidence of SDS is 1:70,000 live births.

There is no cure for SDS thus far, and we need better treatment options, now!

In over 90% of patients, SDS is caused by mutations of the *SBDS* gene. This gene provides instructions for making the SBDS protein, which is found in almost all cells in the body. It is involved in several cellular processes and plays an important role in ribosome maturation. Ribosomes are cellular structures that "read" the cell's genetic instructions (DNA) to create proteins. As of now, it is unclear how *SBDS* mutations lead to the symptoms of SDS.

There are a few other genes associated with SDS, but they account for only a handful of patients. The genetic cause for the remaining <10% of patients is still unknown.

People with SDS may experience any combination of these symptoms and more, ranging from one to many, and from mild to severe:



About **1 in 3** SDS patients develop **MDS / AML** by age 30.

SDS increases the chance of **AML** by several **thousand** fold.

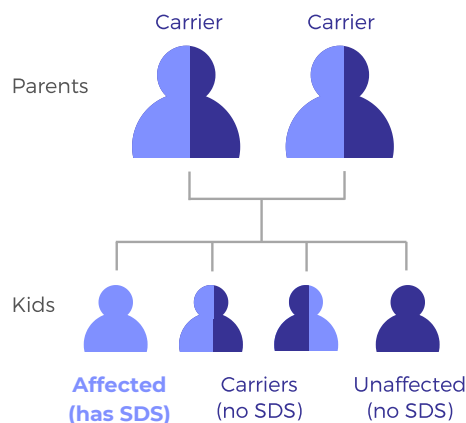
The prognosis for AML in SDS patients is **extremely poor**.

- Frequent or serious infections, including sepsis
- Bone marrow problems and low blood counts (e.g. neutopenia, anemia, and more)
- Skeletal Problems (most often affecting hips, knees, and ribs)
- Endocrine system problems (growth hormones, diabetes, and more)
- Failure to thrive
- Pancreatic exocrine insufficiency (PEI) and malabsorption
- Narrow rib cage and short ribs, which can cause life-threatening problems with breathing
- Developmental delays, executive function disorders, and learning challenges
- Small Stature
- Enlarged liver and liver inflammation
- Dental issues



As a **genetic (inherited) disorder**, SDS cannot be caught from someone. It is also not something a patient can outgrow or get over like a cold.

SDS caused by *SBDS* gene mutations is inherited in an autosomal **recessive** pattern, which means both copies of the gene in each cell need to have a mutation (loss of function) in order to cause SDS. Typically, the parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene (they are carriers), but they do not show signs and symptoms of the condition.



Parents who are carriers have a **25% (1 in 4) chance of having a child with SDS**, a 50% chance of their children being carriers, and a 25% chance of their children being free of *SBDS* mutations.

Are you an SDS patient?

How can YOU help?



Donate to the Shwachman-Diamond Syndrome Alliance. Your donation helps find and support more patients worldwide and build infrastructure to enable and fund research for a cure.
www.SDSAlliance.org/donate



Raise awareness and learn more about SDS at www.SDSAlliance.org. Awareness helps patients get diagnosed and access better treatments and support sooner. Prompt diagnosis can save their life!



Join the Be The Match Registry to become a potential **blood stem cell donor**, or **host a drive**. 1 in 3 SDS patients will need a bone marrow transplant by age 30.
<https://join.bethematch.org/SwabForSDS>

YOU ARE NOT ALONE!

Reach out to us or your local SDS support group. We can help you get connected:
www.SDSAlliance.org/local-support-organizations

PARTICIPATE IN RESEARCH:

Join an SDS registry that covers your region. It is the basis on which all research and therapy development relies. Find your registry at www.SDSAlliance.org/sds-registries

SEEK QUALIFIED MEDICAL CARE!

SDS is a rare disease and it is important to find the right specialists. We and your local SDS support group can help. Reach out today.