



Shwachman-Diamond Syndrome

1600 7th Ave S • Birmingham, AL 35233 • 205.638.9285 • Hematology/Oncology

What is Shwachman-Diamond Syndrome?

- Shwachman-Diamond syndrome (SDS) is a rare condition that affects the bone marrow.
- Decreased number of white blood cells occurs often with this condition
- Low numbers of red blood cells and platelets can occur.
- Poor growth from to diarrhea and difficulty absorbing foods due to abnormal pancreas enzymes
- In some cases, children may be short and have skeletal abnormalities.

What causes Shwachman-Diamond Syndrome?

- SDS is caused by mutations in a gene called SBDS. Every person has two copies of genes. In SDS, both copies of the SBDS gene are abnormal. In most cases, the parents each only of have one abnormal gene so they show no signs of the syndrome.
- Genetic tests can identify the exact mutation in about 90 percent of children with SDS.

What are the symptoms of Shwachman-Diamond Syndrome?

- SDS can vary greatly from child to child. It can impact several parts of the body including the bone marrow, pancreas and skeleton. Some of the most common symptoms of SDS are:
 - Chronic, greasy, and foul-smelling diarrhea
 - Frequent infections
 - Poor growth
 - Pale skin
 - Tiredness and lack of energy
 - Easy bruising or bleeding
 - Skeletal abnormalities, including growth plate changes, rib cage deformities, scoliosis (curvature of the spine), delayed tooth development, dental abscesses, cavities and gum problems
- Children with SDS have a higher than normal risk of developing blood disorders like myelodysplastic syndrome (MDS) and leukemia.

How is Shwachman-Diamond Syndrome diagnosed?

- Delay in the diagnosis of SDS is common because it is a rare condition and often "specialist" doctors are the first to consider this as a possible diagnosis.
- If the main symptoms are diarrhea or weight loss, the gastrointestinal (GI) specialists may order specific blood and stool tests, or genetic testing, that can confirm this disease.
- If the main symptoms are infection, fever, bruising, or loss of energy, these may be due to low blood counts, and you may be referred to a hematologist specialist who may also order specific blood tests that confirm this disease. If the blood counts are low, a bone marrow aspiration and biopsy may be needed to help your hematologist doctor learn if your bone marrow is working properly. In SDS, the bone marrow tests may show that not enough red blood cells, white blood cells, and platelets are being made in the bone marrow. This test is performed at Children's of Alabama under sedation so the child will not feel any pain while it is being done.
- Since SDS impacts other parts of your body, additional tests may include:
 - Kidney, liver and pancreatic function tests
 - Pancreatic testing
 - Stool collection
 - Xrays of the skeleton

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What are the treatments for Shwachman-Diamond Syndrome?

- **Transfusion** - When anemia gets worse or platelets are low, patients may need to get a transfusion of red blood cells or platelets. These transfusions may need to be done on a regular basis.
- **Pancreatic enzyme replacement** - This may be needed to help break down food so that the patient can better absorb nutrients. Many times patients are able to outgrow their need for enzyme replacement as they get older.
- **Antibiotics** - When the white blood cells are too low to fight infection some patients may need antibiotics to treat or prevent infection.
- **Granulocyte colony stimulating factor (GCSF)** - This medicine is given as an injection under the skin and can help increase the number of white blood cells.
- **Bone marrow transplantation** - This is used when the blood counts are very low and other treatments are not working, or if a patient develops myelodysplastic syndrome or leukemia. There are experts at Children's of Alabama that have specialized in bone marrow transplant for SDS and there are specific protocols used which have been published and show excellent results.

What is the long-term outlook for children with Shwachman-Diamond Syndrome?

- With modern therapy, most children with SDS lead normal lives, but may need to take daily medications and have regular visits to the doctors.
- Children with SDS have a small but significant chance of developing blood disorders such as myelodysplastic syndrome (MDS) or leukemia. Nearly 5 percent of children with SDS will develop leukemia, with the risk rising to 25 percent by adulthood.
- Recurrent infections, including pneumonia, ear and skin infections, are common. Doctors at Children's of Alabama have done research into why infections are increased in SDS. One reason is that the white blood cell count tends to be low. The white blood cells that are present do not move around normally and do not function the way they are supposed to.
- Many children with SDS also have growth problems. Sometimes it can be due to loss of vitamins due to problems with absorption in the intestines. Vitamin replacement may be needed.