



Shwachman-Diamond syndrome

Shwachman-Diamond syndrome is an inherited condition that affects many parts of the body, particularly the bone marrow, pancreas, and skeletal system.

The major function of bone marrow is to produce new blood cells. These include red blood cells, which carry oxygen to the body's tissues; white blood cells, which fight infection; and platelets, which are blood cell fragments that are necessary for normal blood clotting. In Shwachman-Diamond syndrome, the bone marrow malfunctions and does not make some or all types of white blood cells. A shortage of neutrophils, the most common type of white blood cell, causes a condition called neutropenia. Most people with Shwachman-Diamond syndrome have at least occasional episodes of neutropenia, which makes them more vulnerable to infections such as pneumonia, recurrent ear infections (otitis media), and skin infections. Less commonly, bone marrow abnormalities lead to a shortage of red blood cells (anemia), which causes fatigue and weakness, or a reduction in the amount of platelets (thrombocytopenia), which can result in easy bruising and abnormal bleeding.

People with Shwachman-Diamond syndrome have an increased risk of several serious complications related to their malfunctioning bone marrow. Specifically, they have a higher-than-average chance of developing myelodysplastic syndrome (MDS) and aplastic anemia, which are disorders that affect blood cell production, and a cancer of blood-forming tissue known as acute myeloid leukemia (AML).

Shwachman-Diamond syndrome also affects the pancreas, which is an organ that plays an essential role in digestion. One of this organ's main functions is to produce enzymes that help break down and use the nutrients from food. In most infants with Shwachman-Diamond syndrome, the pancreas does not produce enough of these enzymes. This condition is known as pancreatic insufficiency. Infants with pancreatic insufficiency have trouble digesting food and absorbing nutrients that are needed for growth. As a result, they often have fatty, foul-smelling stools (steatorrhea); are slow to grow and gain weight (failure to thrive); and experience malnutrition. Pancreatic insufficiency often improves with age in people with Shwachman-Diamond syndrome.

Skeletal abnormalities are another common feature of Shwachman-Diamond syndrome. Many affected individuals have problems with bone formation and growth, most often affecting the hips and knees. Low bone density is also frequently associated with this condition. Some infants are born with a narrow rib cage and short ribs, which can cause life-threatening problems with breathing. The combination of skeletal abnormalities and slow growth results in short stature in most people with this disorder.

The complications of this condition can affect several other parts of the body, including the liver, heart, endocrine system (which produces hormones), eyes, teeth, and skin.

Additionally, studies suggest that Shwachman-Diamond syndrome may be associated with delayed speech and the delayed development of motor skills such as sitting, standing, and walking.

Frequency

Researchers are not sure how common Shwachman-Diamond syndrome is. Several hundred cases have been reported in scientific studies.

Genetic Changes

Mutations in the *SBDS* gene have been identified in about 90 percent of people with the characteristic features of Shwachman-Diamond syndrome. This gene provides instructions for making a protein whose function is unknown, although it is active in cells throughout the body. Researchers suspect that the *SBDS* protein may play a role in processing RNA (a molecule that is a chemical cousin of DNA). This protein may also be involved in building ribosomes, which are cellular structures that process the cell's genetic instructions to create proteins. It is unclear how *SBDS* mutations lead to the major signs and symptoms of Shwachman-Diamond syndrome.

In cases where no *SBDS* mutation is found, the cause of this disorder is unknown.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Congenital Lipomatosis of Pancreas
- Metaphyseal chondrodysplasia, Shwachman type
- SDS
- Shwachman-Bodian-Diamond syndrome
- Shwachman-Bodian syndrome
- Shwachman-Diamond-Oski Syndrome
- Shwachman syndrome

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Shwachman syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0272170/>

Other Diagnosis and Management Resources

- GeneReview: Shwachman-Diamond Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1756>
- MedlinePlus Encyclopedia: Malabsorption
<https://medlineplus.gov/ency/article/000299.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Malabsorption
<https://medlineplus.gov/ency/article/000299.htm>
- Health Topic: Bone Marrow Diseases
<https://medlineplus.gov/bonemarrowdiseases.html>
- Health Topic: Pancreatic Diseases
<https://medlineplus.gov/pancreaticdiseases.html>

Genetic and Rare Diseases Information Center

- Shwachman-Diamond syndrome
<https://rarediseases.info.nih.gov/diseases/4863/shwachman-diamond-syndrome>

Additional NIH Resources

- National Cancer Institute: General Information about Myelodysplastic Syndromes
<https://www.cancer.gov/types/myeloproliferative/patient/myelodysplastic-treatment-pdq>
- National Cancer Institute: Inherited Bone Marrow Failure Syndromes
<https://dceg.cancer.gov/research/what-we-study/bone-marrow-failure-syndromes>
- National Cancer Institute: Leukemia
<https://www.cancer.gov/types/leukemia>

- National Cancer Institute: Myeloproliferative Disorders
<https://www.cancer.gov/types/myeloproliferative>
- National Institute of Diabetes and Digestive and Kidney Diseases: Aplastic Anemia and Myelodysplastic Syndromes
<https://www.niddk.nih.gov/health-information/blood-diseases/aplastic-anemia-myelodysplastic-syndromes>

Educational Resources

- Boston Children's Hospital
<http://www.childrenshospital.org/conditions-and-treatments/conditions/s/shwachman-diamond-syndrome>
- Disease InfoSearch: Shwachman syndrome
<http://www.diseaseinfosearch.org/Shwachman+syndrome/9297>
- MalaCards: shwachman-diamond syndrome
http://www.malacards.org/card/shwachman_diamond_syndrome
- Orphanet: Shwachman-Diamond syndrome
https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=811

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/shwachman-diamond-syndrome/>
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/shwachma.html>
- Shwachman-Diamond Syndrome Canada
<http://sdscanada.ca/>
- Shwachman-Diamond Syndrome Foundation
<http://www.shwachman-diamond.org/>

GeneReviews

- Shwachman-Diamond Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1756>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22Shwachman-Diamond+syndrome%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28shwachman-diamond+syndrome%5BTIAB%5D%29+OR+%28shwachman+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- SHWACHMAN-DIAMOND SYNDROME 1
<http://omim.org/entry/260400>

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