Shwachman-Diamond Syndrome (SDS)

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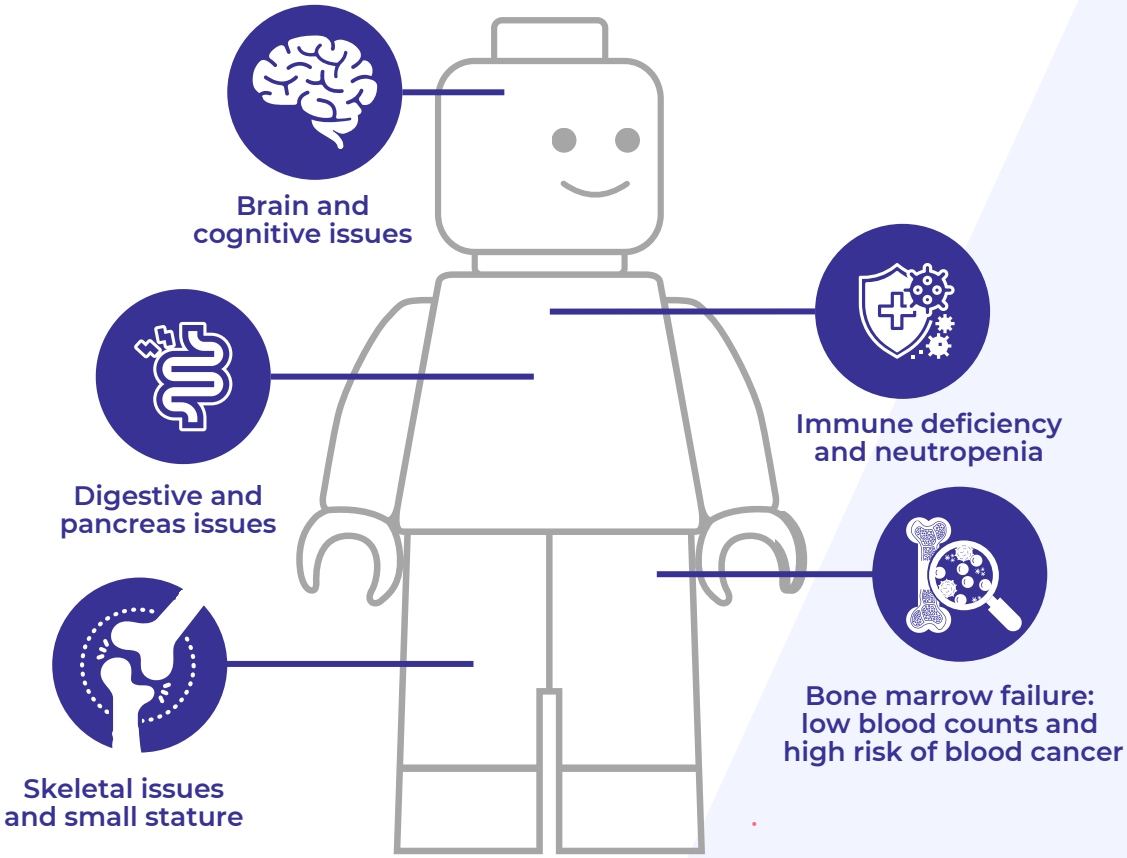
WHAT IS SDS?

US ICD-10 code
D61.02

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*.

SDS affects **each person differently**, with changes over time, but there are several frequently observed **symptoms**:



The bone marrow related issues are of particular concern and can be life-threatening. About **1 in 3* SDS patients will develop MDS or AML (leukemia)** by age 30 & more later.

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SHWACHMAN-DIAMOND SYNDROME ALLIANCE

*www.ncbi.nlm.nih.gov/books/NBK1756

www.SDSAlliance.org/about-sds

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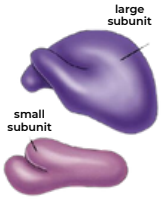
Shwachman-Diamond Syndrome, or SDS for short, is a rare genetic disorder in which the cells in the body cannot make enough ribosomes and protein. Ribosomes are large protein complexes that make all proteins in our cells and body. Proteins are one of the major building blocks of life.

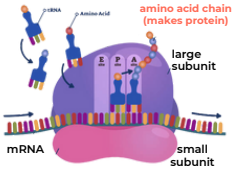
Ribosomes make proteins in our cells by assembling amino acid chains, like hands building towers of Lego blocks.

If we don't have enough ribosomes, our cells struggle to make all the proteins - including enzymes - we need. No wonder that so many organ systems are affected.



The two subunits of the ribosome **separated**: ribosome **doesn't work**

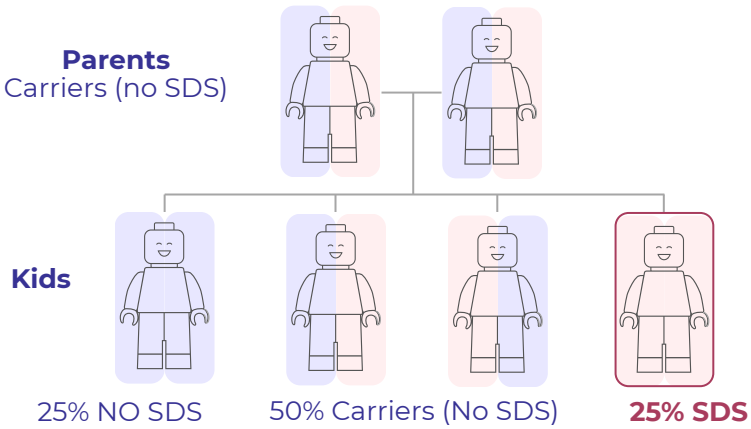
The two subunits of the ribosome **together**: ribosome **works**



Cross section of a **working ribosome**, moving along the mRNA and extruding newly formed **amino acid chain (protein)** on the top right.

The Ribosome strings together amino acids to make proteins, like little hands building Lego towers.

In over 90% of patients, SDS is caused by mutations of the ***SBDS*** gene. There are a few other genes associated with SDS, but they account for less than 1% of patients. The genetic cause for the remaining <10% of patients remains unknown. 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: both copies of the gene need to have a mutation (loose function) 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.

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