

Shwachman-Diamond's syndrome

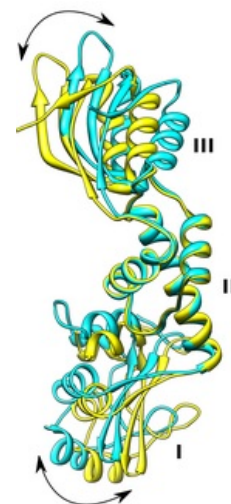
Shwachman-Diamond's Syndrome (Shwachman-Bodian-Diamond Syndrome, SDS, Congenital Pancreatic Lipomatosis (<https://omim.org/entry/260400>); OMIM: 260400 (<https://omim.org/entry/260400>)) is a complex syndrome caused by a mutation in the **SBDS** gene in the 7q11 region and manifestations including:

- **Disorder of the exocrine part of the pancreas** (the exocrine part of the pancreas is replaced by fat, but the endocrine part - Langerhans cells - is normal)
- Skeletal abnormalities
- Abnormalities of a hematological nature

An **increased risk** of cancer, especially leukemia, is being described. Heredity of this mutation has an autosomal recessive character.

From an immunological point of view, these characteristics are present:

- **Neutropenia**
- **Disorders of chemotaxis** in polymorphonuclear leukocytes (phagocytosis dysfunction)
- Absolute lymphocyte count is **normal**, but the percentage of B-lymphocytes may be low or show different defects
- **Thrombocytopenia** is also relatively common
- Apoptosis is increased in the bone marrow



SBDS protein

References

Related Articles

- Primary immunodeficiency
- Phagocytosis

Sources

- ŠÍPEK, Antonín. *Genetic disorders of the immune system* [online]. [cit. 24. 12. 2009]. <<http://www.genetika-biologie.cz/primarni-imunodeficiency>>.
- BARTŮŇKOVÁ, Jiřina. *Imunodeficiency*. 1. edition. Praha : Grada, 2002. pp. 228. ISBN 80-247-0244-4.

Portal: Genetics

Cause	mutation of gene SBDS in region 7q11
Classification and references	
MeSH ID	C53733 (https://www.ncbi.nlm.nih.gov/medgen/537330)
OMIM	260400 (https://omim.org/entry/260400)
orphanet	ORPHA8 (https://www.orphanet.org/contributors/sease_Search.php?g=EN&ca_id=55)
Medscape	958476 (https://emedicine.medscape.com/article/958476-overview)