Shwachman-Diamond syndrome

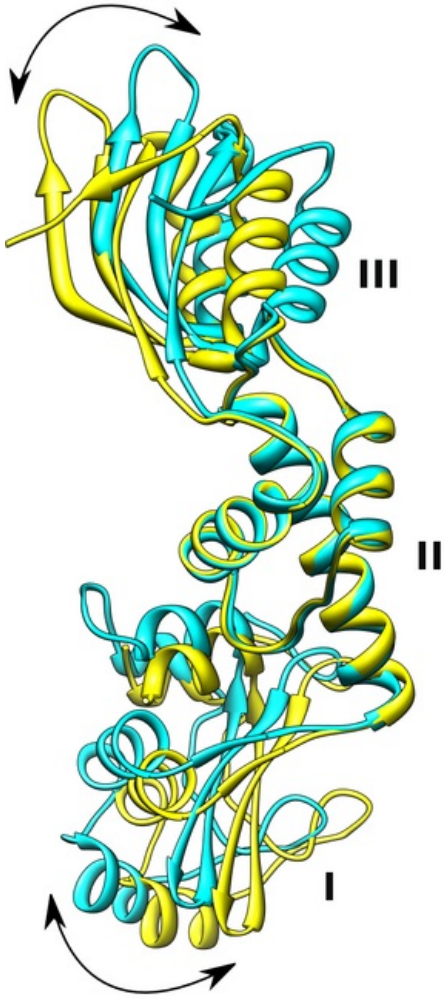
Shwachman-Diamond Syndrome (Shwachman-Bodian-Diamond Syndrome, SDS, Congenital Pancreatic Lipomatosis (https://omim.org/entry/260400); OMIM: 260400 (https://omim.org/entry/260400)) is a complex syndrome caused by a mutation in the *SBDS* gene in the 7q11 region and manifestations including:

- 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 charcteristics 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 demonstrably increased in the bone marrow.



SBDS gene, mutation in region 7q11

References

Related Articles

- Primary immunodeficiency
- Phagocytosis

Source

- ŠÍPEK, Antonín. *Genetic disorders of the immune system* [online]. Last revision June 9, 2006, [cited. December 24, 2009]. < http://www.genetika-biologie.cz/primarni-imunodeficience >.

References

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