Shwachman syndrome

GENERAL INFORMATION

Description:

Shwachman syndrome is characterized by exocrine pancreatic insufficiency, hematologic abnormalities, including increased risk of malignant transformation, and skeletal abnormalities. Males and females are affected with equal frequency.

Alternative names:

- SDS
- Pancreatic insufficiency and bone marrow dysfunction
- Shwachman Bodian Diamond syndrome
- Shwachman Diamond syndrome
- Lipomatosis of pancreas, congenital

Classification:

- Defects of phagocyte function

Inheritance:

Autosomal recessive

OMIM:

- #260400 Shwachman-Diamond syndrome; SDS
- *607444 SBDS gene; SBDS

Cross references:

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Infants often become ill with symptoms by the age of four or six months old. Early problems include failure to thrive, loose, foul-smelling, greasy stools, feeding problems and recurrent infections. The bone marrow in which blood cells are produced is also affected. White blood cells may be low in number and/or not function properly. Anemia and problems with blood clotting may be present. Growth soon slows and remains below normal; only a few children show growth beyond the third percentile. Infections are present early in at least 85% of children. These occasionally lead to death. Diagnosis is generally made in the first few years of life, although occasionally diagnosis is delayed. A typical hyposignal in T2 is showed on magnetic resonance imaging (MRI). Other clinical signs include bone defects with metaphyseal dysostosis, pectus carinatum, cutaneous deffects like ichthyosis, and psychomotor retardation. Hematological disturbances like neutropenia with lowered chemotaxis, thrombopenia and anaemia worsens and evolves toward aplasia.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Shwachman syndrome, ORPHANET

Genetic:

- Academic Medical Centre, Amsterdam, EDDNAL

Therapeutic options:

- Pancreatic enzyme supplementation, as long as neutropenia is moderate and asymptomatic. Prevention or treatment of invasive infections. Correction of hematologic abnormalities, the hematopoietic growth factor is sometimes beneficial. Prevention of orthopedic deformities. In cases when the disease evolves towards medular aplasia or malignant transformation, bone marrow transplant is requiered.
- Shwachman-Diamond Syndrome, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies
- Study of Unrelated Allogeneic Bone Marrow Transplantation in Patients With Benign Congenital Bone Marrow Failure Disorders, ClinicalTrials.gov
- Cancer in Inherited Bone Marrow Failure Syndromes, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: SBDS
Alias(es): CGI-97, FLJ10917, SDS, SWDS, Shwachman-Bodian-Diamond syndrome, Shwachman-Bodian-Diamond syndrome protein (CGI-97)

Localization:

Reference sequences:

DNA: SBDS_DNA (IDRefSeq) , **cDNA:** AY169963 (EMBL) , **Protein:** Q9Y3A5 (SWISSPROT) Other Sequences

Chromosomal Location:

7q11

Maps:

SBDS (Map View)

Variations / Mutations:

- SBDSbase; Mutation registry for Shwachman syndrome

Other gene-based resources:

Ensembl: ENSG00000126524, GENATLAS: SBDS, GeneCard: SBDS, UniGene: 110445, Entrez Gene: 51119, euGenes: 51119, GDB: 110445, HomoloGene: 6438

PROTEIN INFORMATION

Description:

Protein function:

Might possibly play a role in RNA metabolism.

Tissue specificity:

Widely expressed.

Similarity:

Belongs to the upf0023 family.

Other features:

Other related resourses:

InterPro: IPR002140; UPF0023, Pfam: PF01172; UPF0023, ProDom: PD009796; UPF0023, PROSITE: PS01267; UPF0023

Expression pattern for human:

Tissue	Exp. (%)	Clones
whole peripheral blood	98.06	2:8
small intestine	0.21	3:5498
human embryonic stem cells differentiated to an early endodermal cell type	0.20	2:3937
hippocampus	0.18	1:2148
adrenal gland	0.15	1:2677
cerebellum	0.12	1:3339
pericardium	0.10	1:3970
peripheral_nervous_system	0.07	4:21972
thymus	0.07	1:5542
stomach	0.06	12:75571

Animal models:

Mouse:

MGD: ; 4

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- NIH/National Institute of Allergy and Infectious Diseases
- European Society for Immunodeficiencies

Disease specific:

- Shwachman-Diamond Syndrome International
- Shwachman-Diamond Syndrome Canada Inc.
- Shwachman-Diamond Syndrome Support
- Associazione Italiana Sindrome di Shwachman
- Stichting Shwachman Syndroom Support
- Shwachman-Diamond Syndrome Dtl. (Germany)
- Shwachman-Diamond Syndrome America

Other information sources:

- Shwachman syndrome