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Shwachman-Diamond syndrome - etiology, symptoms, diagnosis, treatment, prognosis

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Abstract

Shwachman–Diamond syndrome (SDS) is a rare inherited disorder with exocrine pancreatic insufficiency, bone marrow dysfunction, and skeletal anomalies. Most cases result from biallelic SBDS mutations, though variants in EFL1, DNAJC21, and SRP54 are reported. Patients present with diarrhea, neutropenia, infections, growth delay, and skeletal abnormalities. Diagnosis combines clinical assessment, lab tests, pancreatic function, bone marrow analysis, and genetic confirmation. Management is supportive, including pancreatic enzymes, nutrition, infection prevention, and hematologic monitoring. Hematopoietic stem cell transplantation is reserved for severe marrow failure or malignancy. Prognosis depends on hematologic involvement, but early recognition and multidisciplinary care improve outcomes.

Introduction

SDS is a rare ribosomopathy presenting in infancy with pancreatic insufficiency, neutropenia, and skeletal anomalies. SBDS and related gene mutations disrupt ribosome biogenesis, increasing the risk of marrow failure or malignancy.

Purpose

To summarize SDS genetics, clinical features, diagnosis, and management, highlighting current and emerging therapies.

Material and Methods

A systematic review of PubMed, MEDLINE, Scopus, and Web of Science (1964–2024) used keywords “Shwachman–Diamond syndrome,” “SBDS,” “ribosomopathy,” and “bone marrow failure.” Studies on genetics, clinical features, diagnosis, or management were included.

Discussion

SDS is genetically and clinically heterogeneous. Diagnosis integrates clinical, lab, and genetic data. Management is supportive, with HSCT for severe marrow failure or malignancy. Early diagnosis and multidisciplinary care improve outcomes, while emerging molecular therapies may provide future options.

Keywords
Shwachman–Diamond syndrome, SBDS, ribosomopathy, pancreatic insufficiency, neutropenia, bone marrow failure, EFL1

Introduction:

Shwachman-Diamond syndrome (SDS) is a rare, inherited multisystem disorder characterized primarily by exocrine pancreatic insufficiency, hematologic abnormalities, and skeletal manifestations. First described in 1964, SDS is now recognized as a ribosomopathy a disease arising from defects in ribosome biogenesis most commonly caused by pathogenic variants in the SBDS gene. In recent years, additional genes, including EFL1, DNAJC21, and SRP54, have broadened the genetic and clinical spectrum of the disorder, highlighting its heterogeneity and complexity. [1,2,3]

Clinically, SDS presents most often in infancy or early childhood, with symptoms ranging from chronic diarrhea, malabsorption, and poor growth to neutropenia, recurrent infections, and varying degrees of bone marrow dysfunction. Over time, some patients develop progressive cytopenias, bone marrow failure, or myelodysplastic syndrome, underscoring the need for careful long-term surveillance. Skeletal abnormalities, liver dysfunction, and neurodevelopmental differences may further contribute to the disease burden.

Despite advances in genetic testing and molecular diagnostics, SDS remains challenging to diagnose due to its variable presentation and overlap with other inherited bone marrow failure syndromes. Early recognition is essential, as timely supportive care, nutritional management, and hematologic monitoring can significantly improve patient outcomes. [1,4]

This review provides an updated overview of the current knowledge on SDS, including its etiology, molecular mechanisms, clinical features, diagnostic strategies, and management approaches, aiming to support clinicians and researchers in better understanding and addressing this complex disorder. [5, 6]

Purpose:

The purpose of this review is to provide a comprehensive and up-to-date understanding of Shwachman-Diamond syndrome (SDS), with a particular focus on its clinical spectrum, epidemiology, genetic basis, and underlying pathophysiology. By integrating current research findings and international clinical guidelines, this work aims to enhance awareness of SDS, support earlier and more accurate diagnosis, and inform evidence-based approaches to clinical management. Additionally, the review seeks to address practical challenges in the care of individuals with SDS including variability in disease severity, progression to bone marrow failure, and the risk of malignant transformation and to offer recommendations that may help improve long-term outcomes and quality of life for affected patients.

Material and methods:

This work presents a systematic review of the available literature concerning the etiology, clinical presentation, diagnosis, and management of Shwachman-Diamond syndrome (SDS). A comprehensive search of major scientific databases, including PubMed, MEDLINE, Scopus, and Web of Science, was performed to identify peer-reviewed articles, clinical case reports, cohort studies, and reviews published between 1964 (the year SDS was first described) and 2024. Search terms included “Shwachman-Diamond syndrome,” “SBDS gene,” “ribosomopathies,” “exocrine pancreatic insufficiency,” “bone marrow failure,” and “EFL1 mutations.”

Studies were eligible for inclusion if they addressed one or more key aspects of SDS: genetic etiology, molecular mechanisms, diagnostic criteria, hematologic or pancreatic manifestations, or clinical management strategies. Articles not written in English, publications without original clinical or molecular data, and studies with insufficient methodological detail were excluded.

Data extraction focused on genetic findings (including SBDS and non-SBDS variants), pathophysiological mechanisms, diagnostic approaches such as genetic testing and functional assays, therapeutic interventions, and patient outcomes. Particular emphasis was placed on studies exploring the molecular basis of ribosome biogenesis defects and their clinical consequences. The collected data were synthesized to highlight current knowledge, identify trends in diagnosis and management, and support an updated understanding of the clinical and genetic spectrum of Shwachman-Diamond syndrome.

Etiology:

Shwachman-Diamond syndrome (SDS) most commonly arises from mutations in the SBDS gene located on chromosome 7. This gene encodes the ubiquitously expressed SBDS protein, essential for normal cellular function across multiple organ systems. Because the SBDS gene lies in close proximity to its pseudogene SBDSP, gene-conversion events between these two loci can introduce pathogenic variants that underlie the disorder. [1,2]

SDS, however, is genetically heterogeneous. A particularly severe and ultimately fatal case has been linked to biallelic variants in the EFL1 gene, specifically c.89A>G (p.His30Arg) and c.2599A>G (p.Asn867Asp). Functional studies performed on the patient's B-lymphoblastoid cells and SV40-transformed fibroblasts demonstrated that these compound heterozygous EFL1 variants disrupt the maturation of ribosomes, impairing overall protein synthesis. This defect in ribosome assembly contributes to a profound and aggressive form of Shwachman-Diamond syndrome. [1,2]

Symptoms:

Shwachman-Diamond syndrome (SDS) presents with a wide range of symptoms due to its multisystem involvement. The clinical picture can vary significantly between patients, but several hallmark features are commonly observed. [1,2,3]

The most frequent early manifestation is exocrine pancreatic insufficiency, leading to chronic diarrhea, steatorrhea, abdominal distension, and poor weight gain. These gastrointestinal symptoms typically appear in infancy and contribute to failure to thrive and nutritional deficiencies, particularly involving fat-soluble vitamins. [1,2,5]

Hematologic abnormalities are another major feature of SDS. Chronic neutropenia is the most prevalent finding, predisposing patients to recurrent bacterial infections such as pneumonia, otitis media, and skin infections. Additional blood abnormalities may include anemia, thrombocytopenia, or episodic pancytopenia, reflecting underlying bone marrow dysfunction. As patients age, some may progress to bone marrow failure, myelodysplastic syndrome, or, in rare cases, acute myeloid leukemia. [1,2,4]

Skeletal involvement is also characteristic of SDS and may present as metaphyseal dysostosis, short stature, delayed bone age, or rib cage abnormalities. These features can lead to reduced mobility, chronic pain, or orthopedic complications. [1,6,8]

Other reported symptoms include mild hepatic dysfunction, hepatomegaly, and intermittent elevations of liver enzymes. Neurodevelopmental manifestations such as learning difficulties, developmental delay, or attention deficits have been described in a subset of patients. [5,7,9]

Because of this broad clinical spectrum, the symptoms of SDS may overlap with those of other genetic or metabolic conditions, making early recognition and appropriate diagnostic evaluation essential for optimal care. [1,2,3]

Diagnosis:

The diagnosis of Shwachman-Diamond syndrome (SDS) is based on the combination of characteristic clinical features, laboratory abnormalities, and confirmation through molecular genetic testing. Recent literature emphasizes that although SDS has a recognizable clinical profile, its presentation can vary substantially, making genetic analysis essential for an accurate diagnosis. [11,12]

A clinical suspicion of SDS typically arises in infants or young children who present with symptoms of exocrine pancreatic dysfunction such as chronic diarrhea, steatorrhea, malabsorption, or poor weight gain together with persistent or intermittent neutropenia. Additional clues include recurrent bacterial infections, failure to thrive, growth delay, or skeletal abnormalities such as metaphyseal dysostosis. Laboratory findings often reveal low fecal elastase levels indicating pancreatic insufficiency, deficiencies of fat-soluble vitamins, and fluctuating cytopenias on complete blood counts. [13,14]

Evaluation of bone marrow may be necessary when cytopenias are recurrent or severe. Bone marrow aspirate and biopsy can show hypocellularity, mild dysplasia, or other features consistent with an inherited bone marrow failure syndrome. Ongoing hematologic surveillance is recommended due to the increased risk of bone marrow failure, myelodysplastic syndrome, and acute myeloid leukemia. [14,15]

Genetic testing is the cornerstone of diagnosis. Most individuals with classical SDS have biallelic pathogenic variants in the SBDS gene. Because SBDS has a highly homologous pseudogene located nearby, specialized sequencing methods or confirmatory Sanger testing may be required to accurately identify disease-causing variants. In recent years, next-generation sequencing panels and whole-exome sequencing have revealed that mutations in additional genes—particularly EFL1, DNAJC21, and SRP54 can produce SDS-like phenotypes. As a result, broader genetic testing is increasingly recommended, especially in patients with atypical presentations or in those lacking identifiable SBDS variants. [2,11]

Family testing and genetic counseling play an important role, as SDS is inherited in an autosomal recessive pattern. When pathogenic variants are detected, testing parents and siblings helps clarify carrier status and informs future reproductive planning. [3,15]

Overall, the current diagnostic approach integrates clinical assessment, pancreatic and hematologic evaluation, and comprehensive genetic testing, allowing early identification of affected individuals and guiding appropriate surveillance and management. [3,16]

Treatment:

Management of Shwachman–Diamond syndrome (SDS) is primarily supportive and requires a multidisciplinary approach, although recent research is beginning to expand potential therapeutic strategies. [4,6,10]

For exocrine pancreatic insufficiency, patients are treated with oral pancreatic enzyme replacement and supplementation of fat-soluble vitamins (A, D, E, and K). Regular monitoring of pancreatic function is important, as some patients may experience improvement over time, potentially allowing adjustments to enzyme therapy. Nutritional support is also critical, with dietitian-guided interventions to address poor growth and failure to thrive, and regular assessment of height, weight, and overall nutritional status. [11,15,18]

Hematologic management focuses on controlling cytopenias and preventing infections. Anemia or thrombocytopenia may require red blood cell or platelet transfusions. Patients with neutropenia and recurrent infections may receive granulocyte-colony stimulating factor (G-CSF), particularly before invasive procedures or in the setting of frequent infections. G-CSF therapy is intended to reduce infection risk rather than normalize blood counts, and long-term use requires careful monitoring due to potential risks. [1,2,3]

For patients who develop severe bone marrow failure, myelodysplastic syndrome (MDS), or acute myeloid leukemia (AML), allogeneic hematopoietic stem cell transplantation (HSCT) is currently the only curative option. Outcomes are generally better when HSCT is performed for bone marrow failure rather than overt leukemia, but risks of relapse and non-relapse mortality remain significant. [1,2,3]

Emerging research highlights potential molecular and inflammatory targets in SDS. Studies indicate a systemic inflammatory state in some patients, suggesting that anti-inflammatory strategies could be beneficial in the future. At the molecular level, experimental therapies aimed at correcting aberrant splicing in SBDS mutations, including RNA-based or gene-editing

approaches, have shown promise in preclinical models, indicating a potential future avenue for disease-modifying treatment. [1,2]

Overall, optimal care for SDS requires a multidisciplinary team, often including a hematologist, gastroenterologist, dietitian, endocrinologist, orthopedist, and, when necessary, transplant specialists, immunologists, and psychologists. This coordinated approach aims to manage the multisystem manifestations of the disorder, prevent complications, and improve long-term outcomes. [2,3]

Prognosis:

The prognosis of Shwachman–Diamond syndrome (SDS) varies widely due to its clinical and genetic heterogeneity. Many patients experience a relatively stable course with supportive care, while others develop severe complications that significantly impact life expectancy. [17,18]

Hematologic outcomes are a major determinant of prognosis. Chronic neutropenia and intermittent cytopenias may persist throughout life, but the risk of progression to bone marrow failure, myelodysplastic syndrome (MDS), or acute myeloid leukemia (AML) is substantial. Studies indicate that up to 10–30% of patients may develop MDS or AML during childhood or adolescence, which significantly worsens long-term survival and often necessitates hematopoietic stem cell transplantation (HSCT). [19,20]

Exocrine pancreatic insufficiency typically responds well to enzyme replacement therapy and nutritional support, although persistent malabsorption can contribute to growth delay and vitamin deficiencies if not adequately managed. Growth and skeletal abnormalities, such as metaphyseal dysostosis or short stature, may require orthopedic or endocrinologic interventions but generally do not affect survival. [18,23]

The variability in disease severity is partly influenced by genotype. Patients with classical SBDS biallelic mutations generally have a more predictable disease course, while those with variants in EFL1, DNAJC21, or SRP54 may present with more severe or atypical phenotypes. Functional studies suggest that impaired ribosome biogenesis in these cases can lead to more profound hematologic and systemic complications. [16,22]

With modern supportive care, including pancreatic enzyme replacement, vigilant infection management, and timely HSCT when indicated, many patients can survive into adulthood. Lifelong monitoring is essential to detect hematologic progression, monitor growth and nutritional status, and manage systemic complications. Overall, early diagnosis and

multidisciplinary management remain critical for improving both quality of life and survival in SDS patients. [17,21]

Conclusion:

hwachman–Diamond syndrome (SDS) is a rare, multisystem, inherited disorder characterized by exocrine pancreatic insufficiency, hematologic abnormalities, and skeletal manifestations. Advances in molecular genetics have established the central role of SBDS mutations and have expanded the spectrum of causative genes, including EFL1, DNAJC21, and SRP54, highlighting the genetic and clinical heterogeneity of the disease.

Diagnosis relies on careful clinical assessment, laboratory evaluation, and molecular confirmation, with genetic testing now serving as the diagnostic cornerstone. Management remains largely supportive, focusing on pancreatic enzyme replacement, nutritional support, infection prevention, and vigilant hematologic monitoring. Hematopoietic stem cell transplantation offers a potential cure for patients with severe bone marrow failure or malignant transformation.

Prognosis varies according to disease severity and genotype, with hematologic complications being the primary determinant of long-term outcomes. Early recognition, multidisciplinary care, and ongoing research into molecular mechanisms and potential targeted therapies are essential for improving quality of life and survival. Continued advances in genetic diagnostics and experimental therapies hold promise for more personalized and effective management strategies for SDS in the future.

DISCLOSURE

Author's contribution

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