

The Courage to Live with Shwachman–Diamond Syndrome: Understanding and Caring for a Child with Shwachman–Diamond Syndrome

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ABSTRACT

Shwachman–Diamond syndrome (SDS) is a rare inherited multisystem disorder classically involving exocrine pancreatic dysfunction, bone-marrow dysfunction and skeletal abnormalities. Persistent or intermittent neutropenia is common and can predispose children to recurrent infections, while pancreatic insufficiency may cause malabsorption, steatorrhea, nutritional deficiencies and poor growth. The disorder also carries a long-term risk of marrow failure, myelodysplastic syndrome and acute myeloid leukemia. Most cases are associated with biallelic pathogenic variants in SBDS, with other genes such as DNAJC21, EFL1 and SRP54 accounting for a smaller proportion of genetically confirmed cases. Diagnosis combines clinical findings, hematologic and pancreatic investigations, skeletal assessment and molecular genetic testing. Management is individualized and multidisciplinary, including pancreatic enzyme replacement, nutritional support, infection prevention, hematologic surveillance, developmental care and genetic counselling. Nurses have an important role in monitoring growth, infection, feeding, medication adherence, family education and psychosocial support.^{1,3}

Keywords: Shwachman–Diamond syndrome, SBDS, neutropenia, pancreatic insufficiency, malnutrition, bone marrow failure, pediatric nursing

Case scenario

A 6-year-old child is brought to a tertiary pediatric service with poor weight gain, recurrent respiratory infections, easy fatigue, and frequent loose, bulky stools. The child has a history of feeding difficulty from infancy and delayed motor development.

Examination shows short stature, low body weight, and mild skeletal abnormalities. Laboratory evaluation demonstrates intermittent neutropenia and macrocytosis; fecal elastase is low. Genetic testing identifies biallelic pathogenic variants associated with Shwachman–Diamond syndrome.

INTRODUCTION

Shwachman–Diamond Syndrome (SDS) is a rare inherited disorder that primarily affects the pancreas, bone marrow, and skeletal system, leading to problems with digestion, blood cell production, and bone development. It is usually diagnosed in infancy or early childhood and requires lifelong medical care,

though many patients can live normal lives with proper management. SDS is most commonly associated with mutations in the SBDS gene, which plays a crucial role in ribosome function and cellular growth. The condition manifests with a triad of exocrine pancreatic insufficiency, bone marrow dysfunction, and skeletal abnormalities, though the severity can vary widely among patients. Complications may include recurrent infections, anemia, and an increased risk of developing myelodysplastic syndrome or acute myeloid leukemia. Early diagnosis and multidisciplinary management are essential to improve quality of life and long-term outcomes for affected individuals.^{6,7}

2. Review of literature

Shwachman–Diamond syndrome (SDS) is a rare inherited multisystem disorder characterized primarily by exocrine pancreatic insufficiency, hematologic abnormalities, and skeletal defects, with neutropenia being an early and persistent finding that predisposes affected individuals to recurrent infections and

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growth failure. The condition is most often caused by biallelic pathogenic variants in SBDS, DNAJC21, or EFL1 (autosomal recessive inheritance), or heterozygous variants in SRP54 (autosomal dominant inheritance), leading to impaired ribosome biogenesis and cellular stress. Diagnosis relies on the presence of pancreatic dysfunction and bone marrow failure, supported by molecular genetic testing. Management requires a multidisciplinary approach, including pancreatic enzyme replacement and vitamin supplementation, transfusions for cytopenias, cautious use of granulocyte-colony stimulating factor (G-CSF), and hematopoietic stem cell transplantation (HSCT) for severe marrow failure, myelodysplastic syndrome (MDS), or acute myeloid leukemia (AML). Surveillance involves regular nutritional assessments, hematologic monitoring, bone marrow examinations, orthopedic imaging, neuropsychological evaluations, and dental care. Genetic counseling is essential, as carrier testing, prenatal diagnosis, and preimplantation genetic testing are possible once the familial pathogenic variant is identified. Despite improved supportive care and transplantation strategies, SDS remains associated with significant morbidity and mortality, particularly due to its predisposition to leukemia, making early diagnosis and lifelong multidisciplinary management critical.^{1,4}

Shwachman-Diamond syndrome (SDS) is an autosomal recessive disease which results in inherited bone marrow failure (IBMF) and is characterized by exocrine pancreatic dysfunction and diverse clinical phenotypes. In the present study, we reviewed the internationally published reports on SDS patients, in order to summarize the clinical features, epidemiology, and treatment of SDS. The result shows the clinical features of 156 patients with SDS were

summarized. The three major clinical features of SDS were found to be peripheral blood cytopenia (96.8%), exocrine pancreatic dysfunction (83.3%), and failure to thrive (83.3%). The detection rate of SDS mutations was 94.6% (125/132). Mutations in SBDS, DNAJC21, SRP54, ELF6, and ELF1 have been reported. The male-to-female ratio was approximately 1.3/1. The median age of onset was 0.16 years, but the diagnostic age lagged by a median age of 1.3 years. The conclusion of Pancreatic exocrine insufficiency and growth failure were common initial symptoms. SDS onset occurred early in childhood, and individual differences were obvious. Comprehensive collection and analysis of case-related data can help clinicians understand the clinical characteristics of SDS, which may improve early diagnosis and promote effective clinical intervention.^{5,7}

3. Etiology and genetics

- **Major gene – SBDS:** Most genetically confirmed cases are related to biallelic pathogenic variants in the SBDS gene on chromosome 7.
- **Cellular role:** SBDS protein participates in ribosome maturation and normal cellular stress responses; impaired function contributes to the multisystem phenotype.
- **Other genes:** Rare SDS-like cases can be associated with DNAJC21, EFL1 and SRP54 variants.
- **Inheritance:** The classic form is usually autosomal recessive; parents of an affected child are commonly asymptomatic carriers.
- **Clinical variability:** The frequency and severity of pancreatic, hematologic and skeletal findings vary among affected children.^{2,5}

4. Major genetic features

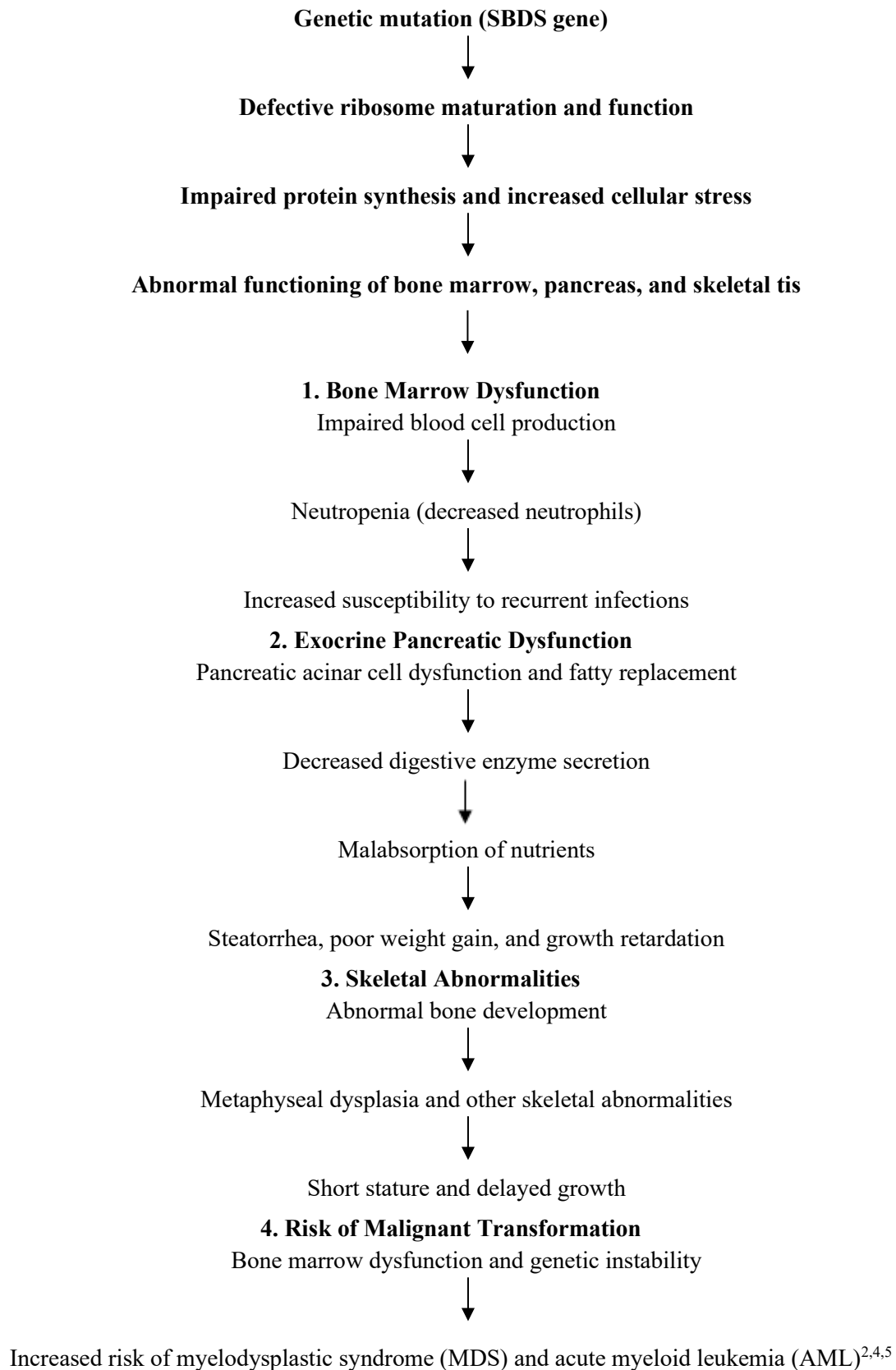
Gene	Main association	Inheritance	Clinical relevance
<i>SBDS</i>	Classical SDS	Autosomal recessive	Ribosome maturation; most common genetic cause
<i>DNAJC21</i>	SDS-like phenotype	Autosomal recessive	Bone-marrow dysfunction and pancreatic features may occur
<i>EFL1</i>	SDS-like phenotype	Autosomal recessive	Ribosome biogenesis; rare cause
<i>SRP54</i>	SDS-like phenotype	Often autosomal dominant	Congenital neutropenia / pancreatic phenotype in some patients

5. Epidemiology

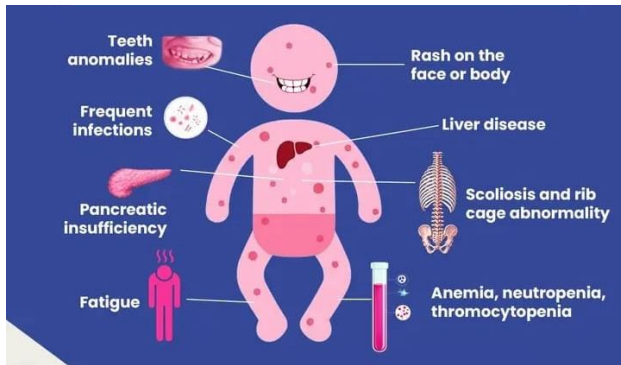
- SDS is rare and is thought to be underdiagnosed because manifestations can be variable.
- Reported frequency is approximately 1 in 80,000 newborns in MedlinePlus Genetics summaries; estimates vary by population and ascertainment.

- There is no simple sex predilection for the classic autosomal-recessive form.

6. Pathophysiology



7. Clinical manifestations



A. Gastrointestinal / nutritional

- Pancreatic exocrine insufficiency
- Steatorrhea or bulky, foul-smelling stools
- Poor weight gain and growth failure
- Deficiency of fat-soluble vitamins (A, D, E and K)
- Feeding difficulties

B. Hematologic / immune

- Persistent or intermittent neutropenia
- Recurrent bacterial infections
- Anemia and/or thrombocytopenia
- Easy bruising or bleeding when platelet counts are low
- Risk of marrow failure, MDS and AML

C. Skeletal

- Short stature
- Metaphyseal dysplasia / delayed ossification

- Hip or knee abnormalities
- Low bone density
- Thoracic abnormalities in some infants

D. Other features

- Delayed motor or speech development
- Liver abnormalities in some children
- Dental, skin, endocrine, cardiac or eye findings may occur
- Psychosocial impact on child and family.^{5,6,7,8}

8. Diagnosis and investigations

Diagnosis is based on the characteristic combination of exocrine pancreatic dysfunction, hematologic abnormalities and/or compatible skeletal findings, supported by molecular genetic testing. No single clinical feature is sufficient in every child.

1. Clinical assessment

- Detailed growth and nutritional history: weight, height, BMI, stool characteristics and feeding pattern.
- History of recurrent infections, fever, oral ulcers, bruising or bleeding.
- Developmental assessment and review of motor and speech milestones.
- Physical examination for short stature, skeletal abnormalities and signs of malnutrition.

2. Laboratory and imaging investigations

Area	Investigation	Purpose
Hematology	CBC with differential; platelet count; serial ANC	Detect neutropenia, anemia, thrombocytopenia and trends over time
Bone marrow	Bone marrow examination when indicated; cytogenetics	Assess cellularity, dysplasia and marrow-failure / clonal changes
Pancreas	Fecal elastase; age-appropriate pancreatic enzymes; pancreatic imaging when indicated	Assess exocrine pancreatic function
Nutrition	Vitamins A, D, E, K; PT; nutritional assessment	Identify malabsorption and vitamin deficiencies
Genetics	Molecular testing for <i>SBDS</i> and other relevant genes	Confirm genetic diagnosis and support counselling
Skeleton	Skeletal survey / targeted radiographs	Identify metaphyseal and other skeletal abnormalities

3. DIFFERENTIAL DIAGNOSIS

- Fanconi anemia and other inherited bone-marrow-failure syndromes
- Congenital neutropenia / cyclic neutropenia
- Cystic fibrosis and other causes of exocrine pancreatic insufficiency
- Dyskeratosis congenita and other telomere disorders
- Other syndromic causes of growth failure and recurrent infection^{2,6}

4. Medical management

- **Pancreatic insufficiency:** pancreatic enzyme replacement therapy with meals/snacks as prescribed; monitor stool pattern, growth and nutritional response.
- **Nutrition:** individualized high-energy nutrition when required, monitoring of vitamins A, D, E and K, and supplementation according to laboratory findings.
- **Hematologic care:** serial CBC and specialist follow-up; transfusion support when clinically indicated.
- **Neutropenia / infection:** prompt assessment of fever and infection; selected children with severe recurrent neutropenia may be considered for G-CSF under specialist care.
- **Bone marrow failure / MDS / AML:** specialist surveillance; hematopoietic stem-cell transplantation may be considered for severe marrow failure, MDS or AML.
- **Development and rehabilitation:** physiotherapy, occupational therapy, speech therapy and individualized educational support as indicated.
- **Genetic counselling:** explain inheritance, recurrence considerations and implications for family members.^{5,8,9}

9. Nursing management

Growth and nutrition

Assessment: Monitor weight, height, BMI, stool pattern, appetite, feeding tolerance and signs of vitamin deficiency.

Interventions: Give pancreatic enzymes exactly as prescribed; provide small, nutrient-dense meals if tolerated; monitor intake/output; coordinate with dietitian; administer prescribed vitamin supplements.

Goal: Maintain adequate nutrition and promote steady growth.

Infection prevention

Assessment: Monitor temperature, ANC when available, respiratory symptoms, skin/oral lesions and other infection signs.

Interventions: Hand hygiene, aseptic technique, timely reporting of fever, vaccination according to specialist plan, avoid unnecessary exposure to infectious contacts.

Goal: Reduce preventable infections and detect infection early.

Bleeding / hematologic monitoring

Assessment: Observe for bruising, petechiae, pallor, fatigue and bleeding.

Interventions: Monitor CBC results; avoid unnecessary trauma when thrombocytopenic; administer blood products as ordered; educate family about warning signs.

Goal: Maintain safe hematologic status.

Development and psychosocial care

Assessment: Assess developmental milestones, school participation, communication and family concerns.

Interventions: Encourage age-appropriate play; coordinate therapy services; provide simple explanations; support caregivers and promote normal family routines.

Goal: Maximize developmental potential and family coping.

10. Nursing care plan

Assessment	Nursing Diagnosis	Goals / Expected Outcomes	Nursing Interventions	Evaluation
Poor weight gain, steatorrhea, pancreatic insufficiency	Imbalanced nutrition: less than body requirements related to malabsorption	Child will maintain adequate nutritional status and demonstrate improved weight gain	Administer pancreatic enzymes as prescribed Monitor weight and stool pattern Provide nutrient-dense diet as ordered Monitor fat-soluble vitamins	Weight and nutritional indicators improve
Intermittent neutropenia, recurrent infections	Risk for infection related to impaired immune defense / neutropenia	Child will remain free from preventable infection and receive early treatment when infection occurs	Monitor temperature and infection signs Strict hand hygiene Follow infection-control precautions Teach family fever warning signs	No new infection or infection detected early
Anemia / thrombocytopenia may occur	Risk for bleeding related to reduced platelet count, when present	Child will remain free from avoidable bleeding	Observe for petechiae/bruising Avoid unnecessary trauma Monitor platelet count Administer prescribed therapy	No active bleeding
Short stature, skeletal abnormalities	Impaired physical mobility related to musculoskeletal abnormalities, when present	Child will participate in safe age-appropriate activity	Assess mobility and pain Encourage safe activity Coordinate physiotherapy Use fall-prevention measures	Mobility maintained or improved
Chronic illness; family questions and worry	Anxiety (family) related to chronic condition and uncertainty	Family will verbalize understanding and demonstrate effective coping	Explain condition in simple language Teach home monitoring and medicines Encourage questions Refer for genetic counseling/support	Family demonstrates understanding and coping

11. Important multidisciplinary care

Pediatrician / pediatric hematologist
Gastroenterologist and dietitian
Geneticist / genetic counsellor
Physiotherapist and occupational therapist
Speech and developmental therapist when required
Orthopedic specialist for significant skeletal problems
Nursing team for continuous monitoring, education and coordination
Priority Nursing

12. Problems

Nutritional deficiency and poor growth
Risk of infection due to neutropenia--
Risk of bleeding when thrombocytopenia is present
Delayed development / impaired mobility
Caregiver knowledge deficit and anxiety^{6,7,8,9}

CONCLUSION

Shwachman–Diamond syndrome is a rare inherited multisystem disorder in which pancreatic dysfunction, bone-marrow abnormalities and skeletal involvement form the central clinical pattern. Children may present with recurrent infections, poor growth, steatorrhea, neutropenia or orthopedic abnormalities, and the severity can change over time. Diagnosis requires careful clinical assessment together with hematologic, pancreatic, skeletal and genetic evaluation. Management is supportive and multidisciplinary, with particular attention to pancreatic enzyme replacement, nutrition, infection prevention, hematologic surveillance and early recognition of marrow complications. Nurses are essential in day-to-day monitoring, medication and feeding support, family education, infection prevention and coordination of

long-term care. Early recognition and structured follow-up can help improve nutrition, development, safety and quality of life.^{2,7,9}

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