orphanaesthesia

# Anesthesia recommendations for

# Shwachman-Diamond syndrome

**Disease name:** Shwachman-Diamond syndrome

**ICD 10:** Q45.3

**ORPHAcode:** 811

**OMIM:** 260400

**Synonyms:** Shwachman syndrome; Shwachman-Bodian-Diamond syndrome; Shwachman-Diamond-Oski syndrome; Shwachman-Bodian syndrome; SDS

**Brief disease summary:** Shwachman-Diamond syndrome (SDS) is a rare inherited multisystem disorder characterized by exocrine pancreatic dysfunction, bone marrow dysfunction and skeletal abnormalities. Immunological, hepatic and cardiac disorders may also occur. Persistent or intermittent neutropenia is common; anemia and thrombocytopenia may also be present. Patients are at increased risk of bone marrow failure, myelodysplastic syndrome (MDS), and acute myeloid leukemia (AML). Skeletal manifestations include short stature, rib cage dysplasia, metaphyseal dysostosis and osteoporosis [1-3]. Contemporary diagnostic evaluation recognizes pathogenic variants in SBDS and, in a smaller proportion of patients with an SDS phenotype, DNAJC21, EFL1 or SRP54 [4].

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Diagnosis may be incorrect; if uncertainty exists, the diagnosis should be re-evaluated.

Every patient is unique; individual circumstances must always guide clinical care.

Medicine is in progress; new clinical knowledge may not be yet reflected in this recommendation.

Recommendations are not rules or laws; they provide a framework to support clinical decision-making. Although this recommendation has passed a structured review process, it does not meet the formal criteria of a guideline.

Translations may not always reflect the most recent updates of the English version.

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**Find more information on the disease, its centers of reference and patient organizations on Orphanet: www.orpha.net**

## Emergency information

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| A | **AIRWAY / ANESTHETIC TECHNIQUE** | Craniofacial, dental and cervical skeletal abnormalities may make tracheal intubation difficult. Subglottic stenosis has been reported. Prepare a difficult-airway strategy; videolaryngoscopy and flexible bronchoscopy should be available as appropriate. Regional/neuraxial techniques require assessment of anatomy and bleeding risk. |
| **B** | **BLOOD PRODUCTS (COAGULATION)** | Cytopenias are common. Review a recent complete blood count and assess clinically relevant coagulopathy / bleeding history before invasive or neuraxial procedures. Ensure that appropriate blood products are available based on current blood counts and anticipated blood loss. |
| **C** | **CIRCULATION** | Congenital heart disease and myocardial dysfunction have been reported. Cardiac assessment should be guided by history, examination, symptoms and the planned procedure. |
| **D** | **DRUGS** | No SDS-specific contraindication to commonly used anesthetic agents is established. Consider chronic pancreatic enzyme/vitamin therapy, hematological treatment and transplant-related medication when relevant. |
| **E** | **EQUIPMENT** | Careful positioning is required because skeletal dysplasia/osteopenia may increase fracture risk. Patients with significant thoracic deformity, respiratory impairment, severe cytopenia or recent hematopoietic stem cell transplantation (HSCT) may require enhanced PACU/IMC/ICU care. |

## Additional disease information

Pancreatic insufficiency may improve with age, but malabsorption and deficiencies of fat-soluble vitamins can persist. Bone marrow is often hypocellular on examination. Recurrent infections may reflect both neutropenia and broader immune dysfunction [2]. A history of progressive marrow failure, clonal hematological disease, myelodysplastic syndrome/acute myeloid leukemia, or HSCT is particularly relevant to perioperative planning [5,6].

## Typical surgery and procedures

Orthopedic surgery for correction of skeletal deformities (extremities or thoracic cavity), cardiac surgery for congenital heart defects, and cesarean section in patients with a hypoplastic or deformed birth canal have been reported [1,3,7]. Procedures related to hematological complications or HSCT may also require anesthesia or sedation [5,6].

## Type of anesthesia

Both general and regional anesthesia have been described in patients with SDS. There are no known contraindications to specific anesthetic agents. Skeletal deformity or scoliosis may make landmark-based regional or neuraxial techniques difficult; ultrasound guidance may be useful. Thrombocytopenia and clinically relevant coagulopathy must be assessed before neuraxial or other deep regional techniques. The choice of technique should be individualized based on airway anatomy, thoracic and skeletal deformity, current hematological status, bleeding risk, infection risk and the planned procedure.

## Necessary additional preoperative testing (beside standard care)

A complete blood count should be obtained to identify neutropenia, anemia and thrombocytopenia. Coagulation testing should be considered when clinically indicated, particularly with bleeding history, liver dysfunction, malabsorption or possible vitamin K deficiency. Thoracic deformity may reduce lung volumes. Echocardiography may be useful when cardiac dysfunction or congenital heart disease is suspected [3]. Review the current hematological course, including recent blood counts, transfusion requirements, marrow failure, MDS/AML and any history of HSCT. In complex cases, perioperative planning should be coordinated with the treating hematology/transplant team [5,6].

## Particular preparation for airway management

Dental misalignment and dysplastic facial bones or cervical vertebrae may lead to intubation difficulties. A focused airway evaluation and review of previous anesthetic records are recommended. Subglottic stenosis has been reported; unexplained stridor or dyspnea should prompt further airway assessment, and smaller tracheal tubes should be available when subglottic narrowing is suspected.

Airway management should be individualized. Videolaryngoscopy should be considered as part of the difficult-airway strategy, and flexible bronchoscopy should be available when significant anatomical abnormalities or subglottic stenosis is suspected. Successful tracheal intubation with a videolaryngoscope has been reported in an adult with SDS and congenital epidermolysis bullosa [8]. Awake or mildly sedated flexible bronchoscopic intubation remains an option when loss of spontaneous ventilation or conventional airway instrumentation is considered hazardous.

## Particular preparation for transfusion or administration of blood products

Pancytopenia or single cytopenias are frequent and should be evaluated before surgery. Transfusion thresholds should be individualized. Vitamin K deficiency and hepatic disease may contribute to bleeding in addition to thrombocytopenia. Point-of-care coagulation testing may be useful during procedures associated with significant bleeding when available and clinically appropriate. Blood-product availability should be matched to the patient's current counts, bleeding history and expected surgical blood loss.

## Particular preparation for anticoagulation

The indication for perioperative anticoagulation or thromboprophylaxis should be balanced against the individual bleeding risk. Complex cases are best discussed with the surgical and hematology teams.

## Particular precautions for positioning, transportation and mobilization

Skeletal deformities are frequent, and reduced bone mineral density may increase osseous fragility. Positioning, transfers and mobilization should be performed carefully to reduce the risk of pressure injury and fracture [1]. Particular care is warranted in patients with marked thoracic deformity or previous HSCT-related organ dysfunction.

## Interactions of chronic disease and anesthesia medications

No clinically established SDS-specific interaction with commonly used anesthetic drugs has been demonstrated. Chronic medications, including pancreatic enzyme replacement, vitamin supplementation, granulocyte colony-stimulating factor (G-CSF), immunosuppressive therapy or other transplant-related medication, should be reviewed individually.

## Anesthetic procedure

General and regional anesthesia have both been performed successfully in SDS. The anesthesiologist should balance airway difficulty against the feasibility and bleeding risk of regional techniques. Strict aseptic technique is important for invasive procedures in patients with neutropenia or immune dysfunction [2]. In patients with severe neutropenia, recurrent infection or recent HSCT, the timing of elective surgery and perioperative infection-prevention strategy should be discussed with the treating hematology/transplant team [5,6].

## Particular or additional monitoring

Neuromuscular blockade should be monitored when neuromuscular blocking drugs are used, especially in patients with airway/thoracic deformity, respiratory impairment or clinical muscle weakness.

The need for invasive hemodynamic or other advanced monitoring should be determined by cardiac status, surgical magnitude and current organ dysfunction.

## Possible complications

Possible complications include difficult airway management, failed or traumatic neuraxial blockade, fractures or positioning injuries, bleeding, infection, and postoperative respiratory compromise. Cardiac decompensation may occur in patients with clinically significant cardiac disease.

## Postoperative care

Airway and thoracic deformities may increase the work of breathing. Adequate postoperative observation in PACU and, when clinically indicated, IMC or ICU should be provided.

Postoperative disposition should additionally reflect current hematological status, infection risk, previous HSCT, surgical magnitude and the intraoperative course.

## Disease-related acute problems and effect on anesthesia and recovery

Important disease-related problems that may mimic or complicate anesthetic recovery include acute bleeding due to thrombocytopenia/coagulopathy, fever or infection in the setting of neutropenia/immune dysfunction, respiratory compromise related to thoracic or airway abnormalities, and cardiac decompensation in patients with cardiac involvement. These possibilities should be distinguished from anesthetic adverse effects.

## Ambulatory anesthesia

Evidence specific to ambulatory anesthesia in SDS is very limited. Suitability should be determined individually according to airway, respiratory, hematological and infection-related risk, the planned procedure and the availability of appropriate postoperative observation.

## Obstetrical anesthesia

Pregnancy has been reported in women with SDS [7]. For cesarean delivery, the feasibility and bleeding risk of neuraxial anesthesia should be weighed against the potential airway risk of general anesthesia. Skeletal or pelvic deformity may influence both obstetric management and neuraxial access. A recent platelet count and assessment for clinically relevant coagulopathy are important before neuraxial procedures.

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