orphanaesthesia

Anaesthesia recommendations for patients suffering from

Shwachman–Diamond syndrome

Disease name: Shwachman–Diamond syndrome

ICD 10: Q45.3

Synonyms: Shwachman Syndrome, Shwachman-Bodian-Diamond Syndrome, Shwachman-Diamond-Oski syndrome, Shwachman-Bodian syndrome, Pancreatic Insufficiency and congenital lipomatosis of pancreas, Metaphyseal chondrodysplasia (Shwachman-Diamond type), SDS

Disease summary: Shwachman-Diamond syndrome (SDS) is a rare autosomal recessive disorder. Approximately 90% of patients with SDS have biallelic mutations in the Shwachman-Bodian-Diamond-Syndrome gene, which encodes a protein involved in the ribosome maturation. SDS is characterized by exocrine pancreatic insufficiency, bone marrow dysfunction and skeletal deformities. Immunologic, hepatic and cardiac disorders may also be present. Pancreatic lipomatosis may be seen on imaging. Pancreatic insufficiency improves with age in many patients. Neutropenia is evident in most individuals with SDS, and approximately 50% have anaemia or thrombocytopenia. Bone marrow examination usually shows hypocellularity. Patients with SDS may develop aplastic anaemia, myelodysplastic syndrome or acute myelogenous leukemia. Skeletal manifestations of SDS include short stature, rib cage dysplasia, metaphyseal dysostosis and osteoporosis. Medications used to treat SDS include p.o. pancreatic enzymes, fat-soluble vitamins (A, D, E and K) and granulocyte-colony stimulating factor (G-CSF). Even with adequate pancreatic enzyme replacement, most patients with SDS remain at or below the 3rd percentile for height.

Medicine in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

Find more information on the disease, its centres of reference and patient organisations on Orphanet: www.orpha.net

Typical surgery

Orthopaedic surgery for correction of skeletal deformities (extremities, thoracic cavity).
Cardiac surgery for correction of congenital heart defects (typically in early infancy).
Caesarean section for hypoplastic birth canal.

Type of anaesthesia

Both general and regional anaesthesia have been described for patients suffering from SDS. There are no known contraindications to specific anaesthetic agents.

Skeletal deformities may render landmark-based approaches to regional anaesthesia infeasible. Sonographic guidance is advisable. Neuroaxial blockage may be hindered by scoliosis.

Coagulopathy and thrombopenia have to be ruled out before performing neuroaxial blockage or regional anaesthesia techniques with high bleeding risks.

Epinephrine admixtures to local anaesthetics should be avoided in cardiac risk patients, since it may cause tachycardia and arrhythmias.

Necessary additional diagnostic procedures (preoperative)

Full blood cell count and coagulation profile should be obtained to evaluate coagulation disorders (vitamin K resorption deficit and elevated INR, bone marrow hypoplasia and thrombopenia, anaemia or neutropenia). Beyond that, a careful patient history helps to identify patients with bleeding disorders.

Lung volumes, esp. vital capacity might be impaired in SDS patients with thoracic deformities.

Preoperative echocardiography is helpful to evaluate a cardiac dysfunction.

Particular preparation for airway management

Dental misalignment as well as dysplastic facial bones and cervical vertebrae may lead to intubation difficulties. A thorough clinical evaluation will help to discover relevant airway pathology. Fibreoptic intubation in awake/ mildly sedated patients might be preferable in these SDS patients.

Recurrent airway infections due to neutropenia might result in bronchial hyperreagibility.

Particular preparation for transfusion or administration of blood products

Pancytopenia or single cytopenias (neutropenia, anaemia or thrombopenia) are frequent and should be evaluated before every surgical intervention. Transfusion thresholds should be adjusted individually as chronically anaemic patients may be adapted to low Hb-levels. The immunological impact of transfusion must be considered in this immune-compromised patient collective.

Vitamin K deficiency and hepatic disease (besides thrombopenia) may lead to bleeding disorders. POC testing such as thromboelastometry and pro-coagulants should be available.

Particular preparation for anticoagulation

The need for perioperative anticoagulation should be weighed against bleeding risks and is best addressed in a multidisciplinary discussion.

Particular precautions for positioning, transport or mobilisation

Skeletal deformities are frequent and vitamin D deficiency may result in decreased osseous stability. Patient positioning should be performed carefully to avoid pressure ulcers and fractures.

Probable interaction between anaesthetic agents and patient's long term medication

CNS dopamine transporters may be affected in SDS. However, the clinical consequences for the use of drugs interacting with the dopaminergic system (e.g. haloperidol, droperidol) are uncertain.

Anaesthesiologic procedure

General as well as regional anaesthesia have been performed successfully in SDS.

Depending on the type of surgery, the anaesthetist has to evaluate the difficulty of airway management against the feasibility and bleeding risk of regional techniques.

Immunodeficiency (neutropenia) requires maximum anti-septical precautions for all invasive procedures.

Subglottic stenosis in patients with SDS is reported in literature. Smaller endotracheal tubes than estimated should be available. In patients with inspiratory stridor and/ or dyspnoea, this pathology should be ruled out by ENT laryngoscopy. Alternatively perform awake bronchoscopic intubation.

Particular or additional monitoring

Close monitoring of neuromuscular blockage is recommended especially in patients with airway and thoracic deformities or clinical muscle weakness.

Possible complications

Difficult airway, failed neuroaxial blockades, bone fractures, bleeding. Cardiac decompensation, especially in children with uncorrected heart defects. Ecchymosis may be seen with high pressure tourniquets.

Postoperative care

Airway and thoracic deformities may result in increased work of breathing. Adequate and prolonged post-operative monitoring at PACU, ICU or IMC level should be provided.

Differential diagnostics

Shwachman Diamond Syndrome shares clinical symptoms with cystic fibrosis (CF, e.g. exocrine pancreatic insufficiency, recurring respiratory infections). Genetic testing will render the correct diagnosis.

Ambulatory anaesthesia

Not reported. The possibility to perform ambulatory anaesthesia should be discussed individually for each patient. Adequate safety precautions must be provided.

Obstetrical anaesthesia

Caesarean section in cases of hypoplastic or deformed birth canal. Careful evaluation of spinal or epidural anaesthesia (deformity, bleeding disorder) against general anaesthesia (possible difficult airway).

Literature and internet links

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