

Anesthesia recommendations for **Saethre-Chatzen syndrome**

Disease name: Saethre-Chatzen syndrome

ICD 10: Q87.0

ORPHAcode: 794

Synonyms: Acro-cephalo-syndactyly (ACS) syndrome, ACS III

Disease summary: Saethre-Chatzen Syndrome (SCS) is named after Haakon Saethre, a Norwegian psychiatrist (1931) and F. Chatzen, a German psychiatrist (1932) who independently described a collection of clinical features in two different families. It is among the five most common craniosynostosis syndromes [1,2].

SCS is a subtype of craniosynostosis syndromes with variable presentation that may include craniosynostosis, brachydactyly, syndactyly, ptosis, facial asymmetry, low frontal hairline, strabismus, small ears with a prominent crus, and other limb defects [3,4]. Intellectual development is usually unaffected. Patients may have obstructive sleep apnea (OSA), cleft palate, maxillary hypoplasia, tracheal cartilaginous sleeve (TCS), vertebral anomalies (fusion of C1 and C2) and congenital heart malformations. Raised intracranial pressure (ICP) can be significant in severe cases and may lead to seizures and death.

SCS is an autosomal dominant condition associated with a mutation in the TWIST1 gene on chromosome 7. It has a prevalence of 1:25,000 to 1:50,000 births [5]. Robinow-Sorauf syndrome is currently considered as a phenotypic variant of SCS and mutations of the FGFR2 (10q26.3), TCF12 (15q21) or FGFR2 (10q26) genes can produce a similar phenotype. Diagnosis is usually clinical but can be confirmed with genetic testing. It is characterized by premature fusion of unilateral or bilateral coronal sutures. If monitored and treated from an early age, the prognosis is good.

Most patients will not have a problem with general anesthesia and airway management as long as the relevant co-existing conditions are managed effectively.

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.



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

Emergency information

A	AIRWAY / ANESTHETIC TECHNIQUE	SCS can be associated with cleft palate and patients may have tracheal cartilaginous sleeves. Consider CT scan to assess for tracheal abnormalities.
B	BLOOD PRODUCTS (COAGULATION)	No specific transfusion practices are necessary.
C	CIRCULATION	SCS can be associated with congenital heart malformations. Echocardiogram can be useful to exclude pulmonary hypertension secondary to OSA.
D	DRUGS	There are no specific drugs that should be avoided.
E	EQUIPMENT	Standard monitoring should be administered throughout the complete anesthetic period.

Typical surgery and procedures

Patients with SCS commonly undergo craniofacial surgery, including cranial vault expansion to prevent elevated intracranial pressure. Midfacial surgery may be performed to address OSA, while procedures to correct syndactyly, ptosis, and strabismus are also frequently indicated.

Additional interventions can include adenotonsillectomy, placement of grommets/tympanostomy tubes, and cleft palate repair. In cases involving airway abnormalities, procedures such as microlaryngobronchoscopy (MLB) and reconstructive surgery or tracheostomy for tracheal cartilaginous sleeves may be required [6,7].

Type of anesthesia

Both total intravenous anesthesia (TIVA) and inhalational techniques can be safely used in patients with SCS. Peripheral intravenous access may be challenging due to limb abnormalities and prior cannulation attempts.

Regional anesthesia is feasible and provides the advantage of reducing opioid requirements, though limb deformities can make peripheral nerve blocks technically difficult without ultrasound guidance. Altered anatomy should also be considered when planning ophthalmic regional techniques, such as peribulbar anesthesia. There is a report of inadvertent dural

puncture during caudal anesthesia [8], highlighting the need for careful technique and anatomical awareness.

Necessary additional preoperative testing (beside standard care)

In patients with a history suggestive of OSA, sleep studies and echocardiography can be performed to assess severity. Individuals with congenital heart disease should have a recent echocardiogram prior to intervention. An ENT evaluation and/or imaging may be considered to better characterize the airway anatomy when indicated.

The presence of dyspnea at rest and/or biphasic stridor should prompt laryngotracheal explorations for a cartilaginous tracheal sleeve. Additionally, orbital imaging (MRI, CT, or ultrasound) can be useful to identify anatomical variations when planning ophthalmic regional techniques, such as peribulbar anesthesia.

Particular preparation for airway management

Although a difficult airway is not commonly reported in patients with SCS, a careful upper airway evaluation should be considered, as for all patients with syndromic craniosynostosis. Certain features may make a difficult airway more likely including cleft palate (~5%), high-arched or narrow palate (~25%), cervical spine fusion, or midfacial hypoplasia [9,10]. Appropriate airway adjuncts should be readily available, ranging from simple devices (oropharyngeal or nasopharyngeal airways) to advanced equipment (videolaryngoscopy with standard or angled blade or fiberoptic bronchoscope), particularly in patients with a history of prior difficult airway management [11].

Identification of a tracheal sleeve can be challenging and should be evaluated by an experienced pediatric otolaryngologist. Although tracheal sleeve is characterized by the absence of some tracheal rings and of a flat posterior tracheal wall, it results in a continuous cartilaginous tube that is less distensible than a normal trachea because fibrous tissue between cartilaginous rings as well as the pars membranacea are lacking. This rigidity of the trachea alters the mechanical properties of the airway and airflow dynamics, reducing the efficacy of natural airway protective mechanisms and airway clearing. Moreover, the progressive intraluminal accumulation of granulation tissue results in progressive luminal stenosis [12].

Particular preparation for transfusion or administration of blood products

There is no evidence for specific transfusion practices. Significant blood loss requiring transfusion may occur during craniofacial surgery. General blood-conservation strategies, including the use of tranexamic acid and cell salvage techniques, should be considered as part of perioperative planning.

Particular preparation for anticoagulation

Not reported.

Particular precautions for positioning, transportation and mobilization

Patients with cervical spine fusion and limb abnormalities require particular care during positioning and transport. Those with fixed flexion deformities or joint contractures should be supported and maintained in their natural, pre-existing positions throughout anesthesia to avoid injury and discomfort.

Interactions of chronic disease and anesthesia medications

There are no known anesthetic medications that need to be specifically avoided.

Anesthetic procedure

General anesthesia is considered safe in patients with SCS, and both inhalational and TIVA techniques may be used [13,14,15]. There is no evidence to support the preferential use of any specific induction or maintenance agents. Neuromuscular blocking agents can be administered safely and reversed using standard reversal agents [16].

Opioid administration should be judicious, particularly in patients with coexisting OSA [17,18]. Vasoactive medications may be required for hemodynamic management in individuals with congenital heart disease. The use of local anesthetics is not contraindicated and may be beneficial in reducing perioperative opioid requirements.

Particular or additional monitoring

Intraoperative monitoring needs to be tailored to the procedure. High-risk surgery (for example craniofacial corrective surgery) may require invasive monitoring including arterial and/or central venous cannulation.

Of note, the trigemino-cardiac reflex may result in the absence of tachycardia associated with hypotension. This should be borne in mind during the facial phase of craniosynostosis repair [19].

Possible complications

Airway compromise and repeated attempts at securing the airway may lead to airway edema. Respiratory depression due to sedatives may be problematic in patients with OSA and raised ICP.

Postoperative care

No specific requirement for intensive care or high dependency care. Depending on comorbidities, patients may need a higher level of care in a suitable environment. The use of a nasopharyngeal airway may be useful in case of severe OSA.

Disease-related acute problems and effect on anesthesia and recovery

No specific issues.

Ambulatory anesthesia

Minor procedures especially in patients without comorbidities can be carried out as day case procedures. Patients with severe OSA may need overnight monitoring especially after major surgery.

Obstetrical anesthesia

No problems reported.

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