orphanaesthesia

Anaesthesia recommendations for patients suffering from

Saethre-Chotzen syndrome

Disease name: Saethre-Chotzen syndrome

ICD 10: Q87.0

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

Saethre-Chotzen Syndrome (SCS) is named after Haakon Saethre, a Norwegian psychiatrist (1931) and F. Chotzen, 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.

SCS is a subtype of craniosynostosis syndromes with variable presentation that may include craniosynostosis, brachydactyly, syndactyly (especially fingers 2 and 3), ptosis, facial asymmetry, low frontal hairline, strabismus, small ears with a prominent crus, and other limb defects. Patients usually have normal intelligence. Although less common, patients may also have obstructive sleep apnoea (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 and has a prevalence of 1:25,000 to 1:50,000 births. Patients with deletion in chromosome 7 rather than a mutation have a higher risk of learning disabilities. Diagnosis is usually clinical but can be confirmed with genetic testing. It is characterised by premature fusion of unilateral or bilateral coronal sutures. If monitored and treated from an early age, preferably on a multidisciplinary craniofacial team, the prognosis is good.

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

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

Patients typically come for craniofacial surgery including cranial vault expansion to prevent increase in ICP, mid facial surgery for OSA and correction of syndactyly, ptosis and strabismus. They may also present for adenotonsillectomy, grommets/ tympanostomy tubes/ ear tubes and cleft palate repair. Other procedures include microlaryngobronchoscopy (MLB) and reconstructive surgery for tracheal cartilaginous sleeves.

Type of anaesthesia

There is no evidence in literature recommending inhalational or total intravenous anaesthesia (TIVA) over the other. Peripheral intravenous access can occasionally be difficult due to limb defects and repeated cannulations. The only documented complication with regional anaesthesia is a case of inadvertent dural puncture while performing caudal anaesthesia.

Regional anaesthesia can be used especially as an opioid-sparing technique, although limb deformities can potentially make peripheral nerve blocks challenging. Altered anatomy needs to be considered when planning ophthalmic regional techniques like peribulbar anaesthesia.

Sedation in a patient with history of OSA or raised ICP is preferably avoided.

Necessary additional diagnostic procedures (preoperative)

In patients with history suggestive of OSA, sleep studies and echocardiogram can be performed to quantify severity. Patients with congenital heart disease should also have a recent echocardiogram done. Imaging of cervical spine should be should be considered and is deemed mandatory in some centres. Otolaryngology review and/or imaging should be arranged to rule out problems with airway anatomy where deemed appropriate. Imaging of the eye and orbit (MRI/CT/ Ultrasound) may be considered to rule out altered anatomy if planning an ophthalmic regional technique like peribulbar anaesthesia.

Particular preparation for airway management

There are no recorded cases of difficult airway in these patients.

Patients who may have a difficult airway include those with cleft palate (~5%), high arched or narrow palate (~25%), cervical spine fusion and midfacial hypoplasia. Adjuncts for difficult airway should be available at hand, ranging from simple (oropharyngeal/ nasopharyngeal airways) to the advanced (Glidescope/ fibreoptic bronchoscope) especially for those with a history of previous difficult airway.

Particular preparation for transfusion or administration of blood products

There is no evidence for specific transfusion practices in such cases. Excessive bleeding and requirement for transfusion can be expected in craniofacial surgery and general measures like tranexamic acid and cell salvage should be considered.

Particular preparation for anticoagulation

No specific evidence.

Particular precautions for positioning, transport or mobilisation

Patients with cervical spine fusion and those with limb defects will need special attention to positioning and transport. Patients with fixed flexion deformities or contractures will need to be maintained in their natural positions under anaesthesia.

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

Some patients may be on anticonvulsants which may affect metabolism of anaesthetic drugs.

Anaesthesiologic procedure

General anaesthesia is safe, including inhalational and TIVA technique. There is no evidence favouring any particular induction or maintenance agent. Muscle relaxants can be safely used and reversed with standard reversal agents. Opioid use needs to be cautious in patients with OSA. Vasoactive drugs may be needed to manage coexisting congenital heart diseases. Local anaesthetic use is not contraindicated and may even help by reducing opioid use.

Particular or additional monitoring

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

Patients with severe OSA may need postoperative pulse oximetry and apnoea monitoring.

Possible complications

Airway compromise and repeated attempts at securing the airway may lead to airway oedema. 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 (for example in case of OSA, congenital heart disease).

Information about emergency-like situations / Differential diagnostics

No particular complications related to the syndrome itself.

Ambulatory anaesthesia

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 anaesthesia

No problems reported.

Literature and internet links

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This guideline has been prepared by:

Authors
Prakash Krishnan, Anaesthetics Registrar, Great Ormond Street Hospital, London, UK
drpk.nhs@gmail.com

Jonathan Smith, Consultant Anaesthetist, Great Ormond Street Hospital, London, UK
Jonathan.Smith@gosh.nhs.uk

Peer revision 1
Emily Gallagher, Craniofacial Center, Department of Paediatrics, Seattle Children's Hospital, USA
emily.gallagher@seattlechildrens.org

Peer revision 2
Thiago Gonçalves dos Santos Martins, Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo (USP) - Serviço de Oftalmologia, São Paulo (SP), Brazil
thiagogsmartins@yahoo.com.br