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Osteopathia Striata (with cranial sclerosis)

orphananesthesia

a project of the German Society of Anaesthesiology and Intensive Care Medicine

SUPPLEMENT NR. 9 | 2025

OrphanAnesthesia –
ein krankheitsübergreifendes Projekt des Wissenschaftlichen Arbeitskreises Kinderanästhesie der Deutschen Gesellschaft für Anästhesiologie und Intensivmedizin e.V.

Ziel des Projektes ist die Veröffentlichung von Handlungsempfehlungen zur anästhesiologischen Betreuung von Patientinnen und Patienten mit seltenen Erkrankungen. Damit will OrphanAnesthesia einen wichtigen Beitrag zur Erhöhung der Patientensicherheit leisten.

Patientinnen und Patienten mit seltenen Erkrankungen benötigen für verschiedene diagnostische oder therapeutische Prozeduren eine anästhesiologische Betreuung, die mit einem erhöhten Risiko für anästhesieassoziierte Komplikationen einhergehen. Weil diese Erkrankungen selten auftreten, können Anästhesistinnen und Anästhesisten damit keine Erfahrungen gesammelt haben, sodass für die Planung der Narkose die Einholung weiterer Information unerlässlich ist. Durch vorhandene spezifische Informationen kann die Inzidenz von mit der Narkose assoziierten Komplikationen gesenkt werden. Zur Verfügung stehendes Wissen schafft Sicherheit im Prozess der Patientenversorgung.

Die Handlungsempfehlungen von OrphanAnesthesia sind standardisiert und durchlaufen nach ihrer Erstellung einen Peer-Review-Prozess, an dem eine Anästhesistin bzw. ein Anästhesist sowie eine weitere Krankheitsexpertin bzw. ein weiterer Krankheitsexperte (z. B. Pädiaterin bzw. Pädiater oder Neurologin bzw. Neurologe) beteiligt sind. Das Projekt ist international ausgerichtet, sodass die Handlungsempfehlungen grundsätzlich in englischer Sprache veröffentlicht werden.

Ab Heft 5/2014 werden im monatlichen Rhythmus je zwei Handlungsempfehlungen als Supplement der A&I unter www.ai-online.info veröffentlicht. Als Bestandteil der A&I sind die Handlungsempfehlungen damit auch zitierfähig. Sonderdrucke können gegen Entgelt bestellt werden.

Bisher in A&I publizierte Handlungsempfehlungen finden Sie unter:

www.ai-online.info/Orphsuppl
www.orphananesthesia.eu

OrphanAnesthesia –
a project of the Scientific Working Group of Paediatric Anaesthesia of the German Society of Anaesthesiology and Intensive Care Medicine

The target of OrphanAnesthesia is the publication of anaesthesia recommendations for patients suffering from rare diseases in order to improve patients' safety. When it comes to the management of patients with rare diseases, there are only sparse evidence-based facts and even far less knowledge in the anaesthetic outcome. OrphanAnesthesia would like to merge this knowledge based on scientific publications and proven experience of specialists making it available for physicians worldwide free of charge.

All OrphanAnesthesia recommendations are standardized and need to pass a peer review process. They are being reviewed by at least one anaesthesiologist and another disease expert (e.g. paediatrician or neurologist) involved in the treatment of this group of patients.

The project OrphanAnesthesia is internationally oriented. Thus all recommendations will be published in English.

Starting with issue 5/2014, we'll publish the OrphanAnesthesia recommenations as a monthly supplement of A&I (Anästhesiologie & Intensivmedizin). Thus they can be accessed and downloaded via www.ai-online.info. As being part of the journal, the recommendations will be quotable. Reprints can be ordered for payment.

Find a survey of the recommendations published until now on:

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orphananesthesia

Anaesthesia recommendations for
Osteopathia Striata (with cranial sclerosis)

Disease name: Osteopathia Striata with cranial sclerosis [OS-CS]

ICD 10: Q78.8

Synonyms: Osteopathia striata - Cranial sclerosis syndrome, Hyperostosis generalisata with striations, Robinow-Unger syndrome, Horan-Beighton syndrome [1,3].

Disease summary:
Osteopathia Striata is a disease of sclerosing bone dysplasia, with longitudinal striations in metaphyses of the long bones and pelvis. The combination of this with hyperostosis of the craniofacial bones gives the condition Osteopathia striata with cranial sclerosis: OS-CS. OS-CS is an X-linked dominant congenital disorder with a prevalence of <1/1, 000,000.The causative mutation is located in the Wilms tumour gene on the X-chromosome (WTX). Males and females can both be affected, with a variable clinical manifestation even occurring within the same family [1]. The affection of male phenotypes can be mild or severe. Males mildly affected present with clinical features similar to those of affected females. Males with a severe phenotype suffer from a multi-malformation syndrome which is usually lethal in utero or during the early neonatal period [2].
OS-CS is predominantly diagnosed on the basis of clinical and radiological findings, followed by genetic testing [1,10]. Often suspected shortly after birth particularly in children with more clinically significant disease, however, some patients with milder clinical variants may remain undiagnosed until adulthood.

Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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

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Emergency information

A	**AIRWAY / ANAESTHETIC TECHNIQUE**	All types of anaesthesia can be considered although neuraxial might be more difficult than usual. Difficult airway is common due to frequent presence of Pierre Robin sequence, micrognathia, cleft palate and other craniofacial abnormalities. Post-operatively, the risk of apnoeas and airway obstruction requires closer monitoring.
B	**BLOOD PRODUCTS (COAGULATION)**	No special considerations regarding blood transfusion. No association with coagulation disorders have been reported.
C	**CIRCULATION**	These patients might have a coexisting ventricular septal defect, aortic stenosis, hypoplasia of the right or left heart, a patent ductus arteriosus. Chronic airway anomalies may result in pulmonary hypertension. No conduction defects have been reported.
D	**DRUGS**	There are no contraindications to common anaesthetic agents. Beware of opiate sensitivity and using premedication in patients with chronic airway anomalies.
E	**EQUIPMENT**	Difficult airway equipment should be available. Special attention should be given to positioning due to craniofacial defects and thick bones.

Disease summary

Features include:

- Craniofacial abnormalities: macrocephaly, coronal craniosynostosis, hypertelorism, high forehead with frontal bossing, broad nasal bridge, coarse facial features, prominent occipital bony protrusion, micrognathia, epicanthal folds, facial hyperostosis (overgrowth of the facial bones, square-shaped face), flat occiput, dental arch discrepancies, microdontia [1,2,3,10,16]
- Other skeletal deformities: hardening of the distal parts of the bones, vertebral anomalies (scoliosis, spondylolisthesis), sclerotic ribs, pectus excavatum, anomalies of extremities, pelvic sclerosis, talipes, short stature [1,2,3]
- Neurological: hydrocephalus, abnormal corpus callosum, developmental delay (usually mild), large fontanelles, wide sutures, hearing loss (conductive and sensorineural), cranial nerve palsies, spina bifida occulta (rare), hypotonia [1,2,3,5]
- Airway abnormalities: Pierre-Robin sequence (triad consisting of micrognathia, glossoptosis and cleft palate), laryngotracheomalacia, prominent jaw, dense maxillary and mandibular bone, retrognathia (rare), breathing difficulties, obstructive sleep apnea, cleft lip/palate, bifid uvula [1,2,3,12,15,16]
- Cardiac anomalies (in ~25% of females): ventricular septal defect, aortic stenosis, hypoplastic right or left heart, patent ductus arteriosus [1,2]

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- Gastrointestinal: Hirschsprung disease, anal malformations, omphalocele, intestinal malrotation, feeding difficulties, reflux, gastro-oesophageal dysmotility, pyloric stenosis [2,3,5,15]
- Genitourinary: non-specific kidney abnormalities
- Malignancies: Wilms tumour (~5%, although no direct association with OSCS has been proven), hepatoblastoma, adult onset colorectal/ovarian cancer (case reports) [4,15,16].

Patients presenting with Hirschsprung disease or Pierre Robin Sequence are usually affected with severe phenotype.

Typical surgery

Children with OS-CS may present for a large variety of surgeries due to potential multi-system involvement. These include, but are not limited to:

- Diagnostic procedures in the early childhood i.e. MRI, microlaryngoscopy and bronchoscopy (MLB), intestinal biopsy
- Abdominal surgery i.e. intestine malrotation, pyloric stenosis, PEG for poor feeding/aspiration
- Scoliosis surgery, spinal fusion, orthopaedic surgeries such as club feet repair
- Cleft palate repair, dental procedures (dental anomalies present in around 30% of patients)
- Neurosurgery for nerve decompression, VP shunt
- Surgery for genitourinary anomalies
- Oncological procedures [lines, biopsies, etc.] and oncological surgery for malignancies
- ENT procedures such as myringotomy tubes, implants
- Craniofacial / maxillofacial surgery such as mandibular lengthening, orthognathic bimaxillary surgery, surgically assisted maxillary expansion [2,5,10,14,15]

Type of anaesthesia

General anaesthesia with intubation is recommended for most of the procedures due to the nature of surgery and young age. No reports available suggest excluding either inhalational or intravenous anaesthesia.
There are no reports on the use of regional or neuraxial anaesthesia in patients with OS-CS, but no reason has been found why they should not be considered in appropriate patients. Challenges to neuraxial anaesthesia include sclerotic vertebral bones, lumbar stenosis, scoliosis or previous spinal surgery, and abnormalities such as hydrocephalus (with or without VP shunt) or spina bifida may be contraindications.

Necessary additional pre-operative testing (beside standard care)

Pre-operative testing and investigations will be guided by severity of disease and suspected systems involved. Children with an established OS-CS diagnosis will often be under regular surveillance by multiple specialties (such as 3 monthly abdominal scans to exclude abdominal tumours, cardiology follow up, etc.). For newly diagnosed patients, in addition to a standard pre-operative assessment, pre-operative testing and investigations may include:

- Cardiology assessment, including echocardiography
- Neurological examination

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- Suspected difficult airway or airway abnormalities (such as cleft palate) may require an ENT/craniofacial evaluation

Particular preparation for airway management

Patients with OS-CS often have challenges associated with cranial and maxillofacial defects (as above) in addition to potentially limited mouth opening due to possible sclerosis of mastoid process and ankylosis of TMJ - difficult airways in these patients are not uncommon [15,16].

In the presence of a Pierre Robin sequence, OS-CS present with the same challenges - bag mask ventilation and direct laryngoscopy may be difficult, airway obstruction on induction can be relieved by lateral or prone position or nasopharyngeal airway, and airway difficulties might improve as patients grow older [6]. Patients with tracheomalacia may be difficult to ventilate following muscle relaxant administration for which reason inhalational induction along with preservation of spontaneous breathing along with CPAP may be preferable. Should oxygenation and rescue ventilating techniques fail, a ventilating bronchoscope might be able to maintain oxygenation in these cases [9].

In cases of potential difficult mask ventilation and intubation in OS-CS, difficult airway equipment should be immediately available with various sizes of masks and ETT tubes, stylets, oro- and nasopharyngeal airway adjuncts and ideally a videobronchoscope. An ENT surgeon should be present for emergency tracheostomy or bronchoscopy if the need arises [6]. Peripheral intravenous access should be secured before induction of anaesthesia [8].

Particular preparation for transfusion or administration of blood products

No considerations reported in the literature.

Particular preparation for anticoagulation

No specific considerations reported.

Particular precautions for positioning, transportation and mobilisation

Patients with OS-CS are often of small weight and short stature. Sclerosis of the cervical spine might result in neck stiffness. Associated macrocephaly can lead to difficulties in positioning and easy airway obstruction. In older age, stiffness in joints and deformities of the upper and lower extremities can pose challenges in both positioning and intravenous access.
An increased bone density due to sclerosis often results in hard bones. Less frequently, some bone tissue may be thinned and easily fractured during surgery (fractures of long bones following mild trauma have been reported) [15].

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Interactions of chronic disease and anaesthesia medications

OS-CS patients who have obstructive sleep apnoea or central hypotonia are at risk of opiate hypersensitivity. Lower doses and post-operative monitoring for respiratory distress should be applied.

Anaesthetic procedure

Patients with OS-CS may present with stridor or severe obstructive sleep apnoea requiring CPAP. Due to poor respiratory reserve (young age, sclerotic ribs, CPAP or oxygen therapy at home, cardiac defects) they are at risk of rapid desaturation on induction of anaesthesia.
Sedative premedication is not advised due to the potential occurrence of a difficult airway.
Multimodal analgesia with opioid-sparing techniques would be recommended where possible. There are no specific contraindications to regional anaesthesia or neuraxial blocks, however, they might be difficult to perform and extra care in positioning will then need to be taken.

Particular or additional monitoring

Intraoperative monitoring according to APAGBI / national guidelines should be used. In cases of major surgeries, patients with cardiac defects or a high risk of bleeding, invasive arterial blood pressure should be considered. Relevant monitoring and precautions should be applied depending on the given conditions.

Possible complications

A high bone density and alterations in bone architecture increase the risk of delayed union, nonunion or infection [15]. Procedures involving osteotomies can be more difficult for surgeons and this may prolong surgery [10].
CPAP ventilation in the post-operative period of cleft palate repair can increase the risk of complications like wound dehiscence and airway compromise.

Post-operative care

Post-operatively, patients with Pierre Robin sequence, obstructive sleep apnea, chronic hypoxia or central hypotonia are at increased risk of adverse airway events following anaesthesia and opiate use. Apnoea monitoring overnight is warranted in these patients. In OS-CS patients undergoing major airway surgery, steroids may be considered for airway oedema and delaying extubation. [8]

Disease-related acute problems and effect on anaesthesia and recovery

Patients with OS-CS with difficult airways are at high risk of suffering from airway complications.
Reflux and abnormal GI motility are common in these patients which puts them at risk of pulmonary aspiration and respiratory infection.
Intraoperative hypoxia, hypercapnia or acidosis can exacerbate existing pulmonary hypertension and should therefore be avoided. Most other disease-related problems are chronic but they require specific attention notwithstanding.

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Ambulatory anaesthesia

Convincing evidence leading to specific recommendations is not available yet.

Obstetrical anaesthesia

Many patients with OS-CS survive until adulthood. There are reported cases of successful births, however, the type of delivery and any anaesthetic involvement remains unknown.
Anaesthetic procedures for women in labour can vary from labour analgesia to anaesthesia for caesarean section or instrumental delivery. There are no guidelines on anaesthetic management of women with OS-CS. Therefore general considerations for pregnant individuals with skeletal dysplasia should be applied instead [2].
Even though skeletal deformities in OS-CS are not an absolute contraindication for neuraxial anaesthesia, the procedure might be challenging or impossible. Any benefits may have to be weighed against the risks of a difficult airway. Some adult patients might still have macrocephaly and cleft palate at presentation.

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