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 in the Wilms tumour gene on the X chromosome (WTX). Males and females can be affected, with variable clinical manifestation even within the same family [1]. Male phenotype can be mild or severe. Mildly affected males present with clinical features similar to affected females. Males with severe phenotype suffer from a multi-malformation syndrome which is usually lethal in utero or the early neonatal period [2].

OS-CS is predominantly diagnosed based on 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.

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Medicine is in progress

Perhaps new knowledge

Every patient is unique

Perhaps the diagnosis is wrong

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

## Emergency information

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| A | **AIRWAY / ANAESTHETIC TECHNIQUE** | All types of anaesthesia can be considered although neuraxial might be more difficult than usual.<br>Difficult airway is common due to frequent presence of Pierre Robin sequence, micrognathia, cleft palate and other craniofacial abnormalities. Postoperatively, risk of apneas and airway obstruction requires closer monitoring. |
| **B** | **BLOOD PRODUCTS (COAGULATION)** | No special considerations regarding blood transfusion. No association with coagulation disorders reported. |
| **C** | **CIRCULATION** | These patients might have coexisting ventricular septal defect, aortic stenosis, hypoplastic right or left heart, patent ductus arteriosus.<br>Chronic airway anomalies may result in pulmonary hypertension. No conduction defects 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 to positioning is advised 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 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]

- Gastrointestinal: Hirschsprung disease, anal malformations, omphalocele, intestinal malrotation, feeding difficulties, reflux, gastroesophageal dysmotility, pyloric stenosis [2,3,5,15]
- Genitourinary: non-specific kidney abnormalities
- Malignancies: Wilms tumour (~5% however direct association with OSCS has not 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 use of regional or neuraxial anaesthesia in patients with OS-CS, but no reason found why they may 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 specialities (such as 3 monthly abdominal scans to exclude abdominal tumours, cardiology follow up etc.). For newly diagnosed patients, in addition to standard pre-operative assessment, pre-operative testing and investigations may include:

- Cardiology assessment, including echocardiography
- Neurological examination

- Suspected difficult airway or airway abnormalities (such as cleft palate) may need 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 potential limited mouth opening due to possible sclerosis of mastoid process and ankylosis of TMJ - difficult airways in these patients is not uncommon [15,16].

In the presence of 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 can improve as the patients grow [6]. Patients with tracheomalacia can be difficult to ventilate following muscle relaxant administration therefore inhalational induction with preservation of spontaneous breathing along with CPAP may be preferable. Should oxygenation and rescue ventilating techniques fail, ventilating bronchoscope might maintain oxygenation in these cases [9].

With 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 video bronchoscope. 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. Cervical spine sclerosis can 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 upper and lower extremities can pose challenges in both positioning and intravenous access.
Increased bony density due to sclerosis usually results in hard bones. Less frequently, some bone may be thinned and easily fractured during surgery (fractures of long bones following mild trauma have been reported) [15].

## Interactions of chronic disease and anaesthesia medications

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

## Anaesthetic procedure

Patients with OS-CS may present with stridor or severe obstructive sleep apnea 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 potential 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 needs to be taken.

## Particular or additional monitoring

Intraoperative monitoring as per APAGBI / national guidelines should be used. For major surgeries, patients with cardiac defects or high risk of bleeding, invasive arterial blood pressure should be considered. Relevant monitoring and precautions should be applied as per associated conditions.

## Possible complications

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

## Post-operative care

Postoperatively, 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. Apnea monitoring overnight is warranted in these patients. In OS-CS patients undergoing major airway surgery, steroids for airway oedema and delaying extubation may be considered. [8]

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

Patients with OS-CS with difficult airways are at high risk of 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 be avoided. Most other disease related problems are chronic but still require specific attention.

## Ambulatory anaesthesia

No good evidence available for specific recommendations.

## Obstetrical anaesthesia

Many patients with OS-CS survive until adulthood. There are reported cases of successful births however type of delivery and anaesthetic involvement is 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 [2].
Even though skeletal deformities in OS-CS are not absolute contraindication for neuraxial anaesthesia, the procedure might be challenging or impossible. Benefits may have to be weighed against the risks of difficult airway. Some adult patients might still have macrocephaly and cleft palate at presentation.

## Links and references


1. http://www.orpha.net/ – Portal of rare diseases and orphan drugs.
2. Gear R, Savarirayan R. Osteopathia Striata with Cranial Sclerosis. 2021 Apr 15 [Updated 2023 Mar 30]. In: Adam MP, Mirzaa GM, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2023.
3. http://www.rarediseases.org
4. Quarello, P, Perotti, D, Carli, D, Giorgio, E, Sirchia, F, Brusco, A, Ferrero, GB, Mussa, A, Spadea, M, Ciceri, S, Spreafico, F & Fagioli, F 2021, 'Wilms tumour occurring in a patient with osteopathia striata with cranial sclerosis: A still unsolved biological question', Pediatric blood & cancer, vol. 68, no. 9, p. e29132, viewed 5 June 2023, <https://search.ebscohost.com/login.aspx?direct=true&db=mdl&AN=34028980&site=ehost-live>.
5. Heikoop D; Brick L; Chitayat D; Colaiacovo S; Dupuis L; Faghfoury H; Goobie S; Mendoza R; Napier M; Nowaczyk M; Oh R; Silver J; Prasad C; Saleh M. 'The phenotypic spectrum of AMER1-related osteopathia striata with cranial sclerosis'. The first Canadian cohort. American journal of medical genetics. Part A [Am J Med Genet A], ISSN: 1552-4833, 2021 Dec; Vol. 185 (12), pp. 3793-3803; Publisher: Wiley-Blackwell.
6. Hegde N, Singh A. Anesthetic Consideration in Pierre Robin Sequence. [Updated 2023 Jan 26]. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2023 Jan-Available from: https://www.ncbi.nlm.nih.gov/books/NBK576442/
7. Friedhoff R.J. Anaesthesia for paediatric craniofacial surgery. Finnanest Vol33 Nro4 2000.
8. An Approach to the Airway Management in Children with Craniofacial Anomalies. Sep 2020. DOI: 10.5772/intechopen.93426
9. Vimmi Oshan, FRCA , Robert WM Walker, DCH FRCA, Anaesthesia for complex airway surgery in children, *Continuing Education in Anaesthesia Critical Care & Pain*, Volume 13, Issue 2, April 2013, Pages 47–51, https://doi.org/10.1093/bjaceaccp/mks058
10. Afia Sultana, Mats Sjöström. Orthognathic surgery in a patient with osteopathia striata combined with cranial sclerosis: A case report. Oral and Maxillofacial Surgery Cases, Volume 6, Issue 4,2020. https://doi.org/10.1016/j.omsc.2020.100194
11. Koudstaal MJ, Wolvius EB, Ongkosuwito EM, van der Wal KGH. Surgically Assisted Rapid Maxillary Expansion in Two Cases of Osteopathia Striata with Cranial Sclerosis. The Cleft Palate Craniofacial Journal. 2008;45(3):337-342. doi: https://doi.org/10.1597/07-016
12. Griffiths AL, Heggie A, Holman S, Robertson SP, White SM. Obstructive sleep apnea successfully treated by mandibular distraction osteogenesis in a rare skeletal dysplasia. The Journal of craniofacial surgery. 2013;24(2):508-510. doi:10.1097/SCS.0b013e3182800f04
13. Wu J, Li X, Chen S. Special manifestations and treatment of rare cases of snoring with special facial features and hearing loss in children. The Journal of international medical research. 2022;50(7):3000605221108085. doi:10.1177/03000605221108085
14. Koudstaal, M.J. et al. (2008) 'Surgically assisted rapid maxillary expansion in two cases of osteopathia striata with cranial sclerosis', The Cleft palate-craniofacial journal : official publication of the American Cleft Palate-Craniofacial Association, 45(3), pp. 337–342. doi:10.1597/07-016.
15. Katsevman, G.A. et al. (2016) 'Osteopathia striata with cranial sclerosis (OSCS): review of the literature and case report demonstrating challenges of spinal fusion after trauma', Acta neurochirurgica, 158(6), pp. 1115–1120. doi:10.1007/s00701-016-2794-4.
16. R. Jagtap, M.B. Garrido, M. Hansen. Osteopathia striata in the mandible with cranial sclerosis: a case report and review of the literature. J Korean Assoc Oral Maxillofac Surg 2021; 47(2): 141~144.

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