orphanaesthesia

Anaesthesia recommendations for

Osteogenesis imperfecta

Disease name: Osteogenesis imperfecta

ICD 10: Q78.0

Synonyms: Brittle bone disease, Lobstein syndrome, Porak-Durante-disease, Vrolik-syndrome

Disease summary: Osteogenesis imperfecta (OI) is a hereditary disease characterized by bone fragility and short stature. The predominant molecular reasons are mutations in COL1A1 or COL1A2. Inheritance follows an autosomal dominant pattern, sporadic mosaics and recessive forms are described. The incidence is described as 1:10.000 to 1:20.000 live births. The clinical spectrum represents a continuum ranging from perinatal lethality (type II) to nearly asymptomatic individuals (type I) with occasional fractures and normal stature. Besides the pathological fractures, due to minor trauma, the clinical presentation may also include bone deformity, scoliosis, growth retardation, early hearing loss, blue sclera, abnormal dentin structure, reduced muscle tone, mitral valve prolapse and platelet dysfunction. OI type III is the most severe form in children surviving the neonatal period and leads to extreme short stature. Patients with mild to moderate bone deformities and variable short stature are classified as OI type IV.

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

Typical surgery

Typical surgery includes all kind of osteosynthesis and orthopaedic interventions with focus on the paediatric patients. Abnormal dentine structures occur as extra-skeletal manifestations of OI or as a distinct entity (dentogenesis imperfecta) and frequently need surgical dental care.

Type of anaesthesia

Regional anaesthesia:

With respect to the frequently presented abnormal anatomy of extremities of patients with OI, peripheral nerve blocks might be difficult to obtain. In particular, nerve stimulation can theoretically lead to contraction induced fractures, ultrasound guidance has therefore to be preferred when administering peripheral nerve blocks.

Case series report of successful anaesthetic treatment of patients with OI with neuroaxial anaesthetic procedures (spinal anaesthesia, epidural anaesthesia and caudal nerve block). One has to keep in mind that the full implications of coagulopathy have not been delineated, and, due to growth retardation, the epidural dosage should be reduced and adapted accordingly.

General anaesthesia:

There are several reports of patients with OI who experienced a perioperative hypermetabolic state with fever. This hyperthermia seems not to be of the malignant type. Either increased cellular energy metabolism or central nervous temperature dysregulation are discussed as possible causes. An endocrinological coherence is also suggested; at least 50% of the patients with OI have increased serum thyroxine levels.

One case series describes normal caffeine halothan contracture test (CHCT) results in patients with OI and reported malignant hyperthermia (MH). However, there is one convincing report of a patient with OI and MH who underwent general anaesthesia. In summary, evidence of an association between OI and MH is weak. Occurring hyperthermia in patients with OI can usually be controlled with standard cooling measures.

Difficulties in airway management, vein access or the application of regional anaesthesia procedures are much more frequent in the severe forms of type III.

Suxamethonium should be avoided for the following reasons:

- Lethal hyperkalaemic responses to suxamethonium injections after immobilisation are reported frequently. The up-regulation of two new isoforms of acetylcholine receptors in immobilized or denervated body parts seems to be responsible for the excessive release of potassium ions. Patients with OI are frequently bounded on wheelchair or are otherwise immobilized.

- Literature shows reports of contraction induced-fractures after administration of suxamethonium.

One author reports of a patient with OI who presented with lactic acidosis in context of propofol infusion.

Atropin can result in excessive increased body temperature and should be avoided if possible.

Necessary additional pre-operative testing (beside standard care)

Coagulopathy: Evidence shows a decrease of platelet retention, reduced collagen induced platelet aggregation, reduced factor VIII activity and an increased capillary fragility. Platelet counts and standard coagulation tests should be complemented with platelet function test and factor VIII activity, especially when past medical history presents episodes of haemorrhagic diathesis. Significant intraoperative blood loss is notably higher in the more severe OI types.

Preoperative spirometric tests can show restrictive pulmonary disorders especially when patients present with thoracic dysmorphologies.

Preoperative blood gas analyses show the baseline of gas exchange for later comparison with intra- and postoperative blood gases.

In severely affected patients, there is a risk of a basilar invagination and an atlanto-occipital dislocation. Therefore, an x-ray of the cervical spine might be useful especially when the operation requires a complex positioning of the patient.

When case history shows symptoms of congenital heart defect or malformation of the thoracic vessels, a preoperative echocardiography should be performed.

Particular preparation for airway management

Difficult airway must always be assumed and anticipated in patients with OI. Visualization of the larynx can be hindered by secondary distortion due to thoracic kyphoscoliosis and decreased neck mobility. Overextension of the cervical spine can lead to atlanto-axial dislocation or even fractures and has to be avoided. Patients with dentinogenesis imperfecta are at increased risk of tooth damage or loss during intubation, a preoperative documentation of dental abnormalities can prevent legal trouble. The use of videolaryngoscopy reduces the need for movements of the head for endotracheal intubation.

A case series of 205 anaesthetic procedures in patients with OI report of an overall incidence of airway difficulties of 1.5% of which 66% occurred in patients with OI Type III. In all cases the difficult airway situation could have been overcome by using videolaryngoscopic or fiberoptic endotracheal intubation.

Laryngeal masks and other supraglottic airway devices have also been used successfully, and are indispensible in emergency situations.

Particular preparation for transfusion or administration of blood products

Special transfusion associated implications of patients with OI are not known.

It has to be kept in mind that most of the patients with OI have a reduced body weight compared to their age. Therefore, all transfusions and drugs have to be calculated on body weight, also in adult patients.

Particular preparation for anticoagulation

The review of the literature gives no advice for particular preparation for anticoagulation in patients with OI. However, when administering anticoagulation in patients with OI, the potential of a hereditary coagulopathy always has to be considered.

One also has to keep in mind that patients with OI are at a higher risk of traumatic injuries than patients without OI. Therapeutic anticoagulation and traumatic injuries may result in devastating haemorrhages in these patients.

Particular precautions for positioning, transportation and mobilisation

Transport, positioning, and mobilisation of patients with OI have to be provided with special precaution and respect to the bone fragility of these patients. Vacuum mattresses and positioning aids may help to prevent further fractures in patients with OI. As far as possible, the positioning should be done in cooperation with the awake patient before anaesthesia induction is administered.

By observing these instructions, the reported rate of perioperative fractures is considerably lower than previously estimated. There occurred two perioperative fractures (2.6%) in 76 anaesthetic procedures of OI Type III, and not a single perioperative fracture was reported in 129 anaesthetic procedures of other Types of OI.

Interactions of chronic disease and anaesthesia medications

The most common chronic medication of patients with OI are bisphosphonates. Interactions between bisphosphonates and anaesthetic drugs leading to adverse events are not known.

Anaesthetic procedure

Vascular catheterisation could be difficult. The positioning of the patient's hands and arms has to be arranged carefully with respect to the brittle bones and can be severely restricted by fixed skeletal deformities.

The typically reported proneness to fractures due to blood pressure cuff use is questionable. In a case series of 205 anaesthetic procedures of patients suffering from OI, a single new humerus fracture on postoperative day 1 was reported; however, due to the delayed occurrence and the undocumented localization of the blood pressure cuff, this case could also not be attributed clearly to the use of blood pressure cuffs.

Take care of bone fragility when positioning the patient especially when overextending the head for intubation.

There are no reports about potentially harmful effects of non-depolarizing neuromuscular blocking agents.

In order to prevent atopy and allergic reactions, avoidance of latex contact should be considered.

Particular or additional monitoring

With respect to the extreme bone fragility, monitoring and documentation of the correct positioning of the patient with OI is even more important than in patients without OI.

The positioning of the patient's hands and arms for the placement of an arterial line has to be arranged carefully with respect to the brittle bones.

Neuromuscular monitoring is recommended and has been used safely.

Possible complications

Beside the above-described possible complications, there are certain procedures with special complications in patients with OI.

OI is generally considered a contraindication for intraosseous access. Literature reports of a case with three failed attempts using the EZ-IO drill to establish intraosseous access.

Nonetheless, it is a possibility in case of emergency to gain access. It has to be kept in mind that many OI patients underwent rodding of long bones which might interfere with an intraosseous access.

Post-operative care

In general, the postoperative care does not differ from the one of patients without OI. The postoperative transfer to ICU might be necessary when patients present severe intraoperative complications or persisting disorders like unbalanced haemorrhagic diathesis or restrictive pulmonary symptoms.

The use of non-invasive CPAP ventilation in OI patients has not been reported.

A special focus has to be set on careful mobilisation to prevent further trauma.

Disease-related acute problems and effect on anaesthesia and recovery

There are no typical disease triggered emergency-like situations, which can be confounded with anaesthesia side effects.

Ambulatory anaesthesia

There are no reported experiences in patients with OI in the ambulatory anaesthesia setting. The authors advise against performing ambulatory anaesthesia in patients with OI.

Obstetrical anaesthesia

Pregnant patients with OI are extremely rare. However, there are case reports of successful caesarean delivery with neuroaxial regional anaesthesia even in severe cases of OI Type III. Due to the fragility of the maternal skeleton and a craniopelvic mismatch, sectio ceasarea is the preferred treatment of pregnant patients with OI. The treatment of those patients should be exclusively obtained in specialized hospitals with neonatal ICU.

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