

# orphananesthesia

## Anaesthesia recommendations for patients suffering from **Osteogenesis imperfecta**

**Disease name:** Osteogenesis imperfecta

**ICD 10:** Q78.0

**Synonyms:** brittle bone disease, Lobstein syndrome

Osteogenesis imperfecta (OI) is a hereditary disease characterized by bone fragility and short stature. The 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 1:20,000 life 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, 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.

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Medicine in progress



Perhaps new knowledge

Every patient is unique

Perhaps the diagnostic is wrong

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### Typical surgery

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Typical surgery includes all kind of osteosynthesis and orthopaedic interventions with focus on the paediatric patients.

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### Type of anaesthesia

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#### **Regional anaesthesia:**

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

Single cases and small 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.

Suxamethonium should be avoided for the following reasons:

- Lethal hyperkalemic 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 diagnostic procedures (preoperative)**

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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 esp. when past medical history presents episodes of haemorrhagic diathesis.

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 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 preoperatively echocardiography should be performed.

### **Particular preparation for airway management**

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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 - a subtype of OI which leads to maldevelopment of teeth - are at increased risk of tooth damage or loss during intubation, a preoperative documentation of dental abnormalities can prevent legal trouble.

The fiberoptic intubation provided by an experienced staff seems to be the safest method to secure the airway of patients with OI. Laryngeal masks and other supraglottic airway devices have also been used successfully, and are indispensable in emergency situations.

### **Particular preparation for transfusion or administration of blood products**

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

It has to be kept in mind, that most of the OI-patients 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**

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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.

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#### **Particular precautions for positioning, transport or mobilisation**

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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.

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

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

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#### **Anaesthesiologic procedure**

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Venous access could be difficult, consider central venous line.

Avoid mechanical non-invasive blood pressure, consider arterial line for invasive blood pressure.

Avoid suxamethonium.

There are no reports about the effect of non-depolarizing neuromuscular blocking agents.

Anticipate difficult airway.

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

In order to prevent atopy and allergic reactions, latex contact should be avoided.

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#### **Particular or additional monitoring**

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Beside the standard monitoring for general and regional anaesthesia, a continuous monitoring of body temperature is mandatory for patients with OI undergoing general anaesthesia. Blood gas analysis are recommended every hour.

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.

Mechanical inflation of blood pressure cuffs for non-invasive blood pressure measurement may result in fractures and should be avoided. The positioning of patient's hands and arms

for the placement of an arterial line has to be arranged carefully with respect to the brittle bones of patients with OI.

Neuromuscular monitoring is recommended.

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#### Possible complications

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

The usage of non-invasive blood pressure cuffs with mechanical inflation has to be seen critically. Compression induced fractures are conceivable.

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.

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#### Postoperative care

In general, the postoperative care does not differ from those 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.

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

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#### Information about emergency-like situations / Differential diagnostics

*caused by the illness to give a tool to distinguish between a side effect of the anaesthetic procedure and a manifestation of the diseases, e.g.:*

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

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#### Ambulatory anaesthesia

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

### **Obstetrical anaesthesia**

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Pregnant patients with OI are extremely rare. However, there are case reports of successful caesarean delivery with spinal anaesthesia. Due to 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 in specialized hospitals with neonatale icu.

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