



## :: Osteogenesis imperfecta (OI)



- This document is a translation of the French recommendations drafted by Drs. Tourniaire, Topouchian, Baujat, Rostchild and Le Merrer, reviewed and published by Orphanet in 2008.
- Some of the procedures mentioned, particularly drug treatments, may not be validated in the country where you practice.

### Synonyms:

Lobstein's syndrome, Porak and Durante disease, brittle bone disease, osteopsathyrosis

### Definition:

Osteogenesis imperfecta is a rare condition caused by an abnormality of the extra-cellular matrix. It causes **bone fragility leading to fractures** that may be frequent, and a **variable articular hyperlaxity**. The severity of the conditions is very variable from one person to another. The severe forms may be accompanied by bony deformation and very impaired mobility. The skeletal abnormalities can be associated with poor dentition, blue sclera and deafness.

### Further information:

[See the Orphanet abstract](#)

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# Pre-hospital emergency care recommendations

## Call for a patient suffering from osteogenesis imperfecta

### Synonyms

- ▶ Lobstein's syndrome, Porak and Durante disease, brittle bone disease, osteopsathyrosis

### Aetiology

- ▶ abnormal collagen structure (mutations include those of the *COL1A1* and *COL1A2* genes)

### Special risks in an emergency

- ▶ bone fractures (long bones, ribs, vertebra) after trauma (even very minor trauma)
- ▶ sprains due to ligamentary hyperlaxity
- ▶ spontaneous haematoma (cutaneous and vascular fragility)
- ▶ pain syndrome

### Frequently used long term treatments

- ▶ level 1 analgesics (Paracetamol)
- ▶ vitamin D at preventive dose
- ▶ careful physiotherapy
- ▶ bisphosphonates (intravenous course, hospitalisation)

### Complications

- missing a fracture
- highly fragile cervical spine in some forms
- be wary of causing further trauma (bumping, traction on a limb, light knocks) as even a minor trauma can cause fractures or micro-fractures.

### Specific medical care prior to hospitalisation

- ▶ move with care under all circumstances
- ▶ organise transport to the hospital and advise the care team of safeguards
- ▶ warn the hospital reception team that the patient is en-route and of his/her pathology
- ▶ effective pain management: immobilisation

# Recommendations for hospital emergency departments

## Emergency issues and recommendations

### 1. Diagnostic situation

The child may present at casualty with **symptoms suggestive of a fracture** involving long bones, ribs or vertebra **following minor trauma**.

In a young child, **unusual crying and/or the absence of spontaneous movement of a limb** are signs suggesting a fracture.

Two situations may present:

#### ▶ **The diagnosis is known:**

- **ask if the patient has an emergency card.** The key elements of appropriate management are written on it especially pain management (pain scales) as are the names of the patient's doctors.
- **evaluate the severity**
  - what **type of fracture**; what **type of OI**.
  - Severity criteria and suspicion of complications:
  - proximal long bone fractures (femur and humerus)
  - fracture of 2 bones in arm or leg
  - fracture with displacement
  - vertebral fracture
  - large haematoma
  - reiterated fractures
- Emergency investigations and management: see point b.

#### ▶ **The diagnosis is not yet known:**

- Bone fragility should be considered when a fracture is caused by minor trauma or when there are repeated fractures in a child who may have blue sclera. When there are fracture(s) and bruises in an infant **the cause is not necessarily abuse and signs that could be associated with OI should be looked for**: blue sclera, poor dentition, fine and transparent skin, capillary fragility, wormian bones, family history of frequent fractures.

### 2. Immediate therapeutic measures for fractures

#### ▶ **At home and in the patient's living environment**

Emergency first aid must always be given on the fracture site immediately: **immobilisation and provision of analgesics**

#### ■ **Analgesics**

- if pain is initially **very intense**  
*Morphine used correctly does not produce any tolerance or dependence and is not dangerous. Families must have suitable analgesics stored in a safe place (e.g. stored with immobilisation equipment) in order to be able to administer them prior to the infant being transferred to hospital.*

Give **morphine orally: as soon as possible: 0.5 mg/kg** (maximum 20 mg)

Morphine syrup 5 mg/ml	Graduated pipette from 2.5 to 10 mg; graduation = 0.5 mg
Morphine breakable tablets	Available as 5 mg and 10 mg
Morphine capsules (can be opened)	Available as 5 mg, 10 mg and 20 mg

In principle, pain diminishes after immobilisation (see later).

Ibuprofen + paracetamol are then used; if insufficient, codein is used in combination with these two medicaments.

**If pain remains intense after immobilisation**, continue with morphine 0.2 mg/kg every 4 hours (consult if pain persists despite everything). When the pain has diminished, change to paracetamol + codein.

- if the **pain** from the fracture is **moderate**

Paracetamol + ibuprofen regularly  
+/- codein (authorised from 1 year old)

Weight	Medicine
Up to 10 kg	codein syrup: 0.5 mg per kg every 4 hours + paracetamol syrup: 1 dose per kg every 6 hours
From 10 kg	Paracetamol 500mg + codein 30mg (effervescent tablets): ½ tablet per 10 kg; 1 tablet per 20 kg Or Paracetamol 500mg + codein 30mg (capsules): 1 capsule per 20 kg Or Paracetamol 400mg + codein 20mg (tablets): 1 tablet per 10 kg

If pain is controlled, continue administering as long as necessary.

If pain is not controlled, give morphine (can be given immediately after codein is ineffective).

#### ■ Immobilisation (to be done **after administering analgesics**)

- Abnormal movement at the fracture site is the main cause of pain. **Immobilisation is the best analgesic.** It makes movement and the possible transfer to an emergency and/or radiology service easier.
- **Immobilisation equipment must always be ready and available.** This equipment will have been prepared with a qualified person from the hospital care team (physiotherapist, rehabilitation doctor, orthopaedist...).
- **for the arm**, a gutter from the shoulder to the hand that keeps the elbow flexed at a right angle. The immobilisation is completed by bandaging the elbow against the body.
- **for the leg**, the pre-cut splint (in cardboard) is placed laterally alongside the thorax and down to the foot.
- **Apply light traction**, which must be even and continued along the axis of the leg on either side of the fracture site to avoid deformation, **before fitting the immobilisation equipment.**
- The sensitivity, colour and temperature of the extremity must be checked to **ensure there is no vascular compression or neurological problems.**
- For numerous small fractures without displacement (which the patient and parents learn to recognise quickly and which do not need complicated treatment), these home splints are often sufficient and are used until pain disappears (3 - 4 weeks).

#### ▶ Hospital management

##### ■ Orthopaedic evaluation

- type of fracture
- type of OI

## ■ Analgesics

### - In a specialised environment, it is possible to:

- use **morphine treatment**: possibly by IV: **0.1 mg/kg (by the titration method) then 0.025 mg/kg** every 5 minutes until adequate analgesia is obtained, or orally as described above (immediate release morphine at 0.5 mg/kg then followed by subsequent doses of 0.2 mg/kg).
- use **nerve blocks** (e.g.: the crural nerve for femur fractures) provided that they are performed by a competent person.

### - Before taking blood

- Local anaesthetics cream

### - Before any manipulation (radiograph, plastering)

- oxygen-nitrous oxide mix systematically
- **Gentle mobilisation**, especially during radiological examinations

## ■ Consolidating the immobilisation

- The **orthopaedic or surgical management** uses different bone repair techniques. If the deformations are significant with frequent fractures, it is possible to use telescopic wire splints or pins. The aim for these interventions is to reduce the fragility, prevent skeletal deformity, allow upright posture and give the infant more independence and avoid the need for using external apparatus.

## Orientation

- ▶ **Where**: a casualty service with an orthopaedic unit that is suitable for the patient's age and whenever possible, in the hospital managing the patient, otherwise in the nearest hospital with maximum collaboration between the treating medical team and the nearest competent centre.
- ▶ **When**: as soon as possible
- ▶ **How**: (see above) **immobilisation** of the whole limb by splint before moving. **Gentle mobilisation**, especially during radiological examinations. Pain management with **effective analgesics (morphine type drugs)**.

## Drug interactions

- ▶ There is no formal drug contra-indication in this condition but **it is wise to avoid any drug that has a risk of haemorrhage**, given the vascular fragility associated with osteogenesis imperfecta.

## Anaesthesia

- ▶ **Take care during intubation** due to the potential association of the condition with fragility of the cervical bones and the dental arch.
- ▶ **Pre-surgical analysis**: double determination of blood group, platelets haemogram, prothrombin time (PT) and kaolin-activated partial thromboplastin time test, electrolyte biochemistry (heightened risk of water loss and starvation ketosis), evaluation of platelet function by PFA if history of haemorrhage
- ▶ **During the intervention**: Gentle manipulation especially when inserting or during intubation. Risk of humerus lesions from the manometer armband.
- ▶ Initially insert **several IV catheters** because of the **haemorrhage risk**.
- ▶ Monitor the haemoglobin level daily for 4 days after osteotomy.

## Preventive measures

- ▶ Use **effective analgesics** from the start.
- ▶ **Always check the cervical spine** in severe forms.

## Additional therapeutic measures and hospitalisation

### ▶ For every patient

- It is important **to be very careful during every manipulation** especially if it is necessary to undress the patient.
- Obviously, it is important to **carefully explain every therapeutic decision to them** and their consequences for everyday life:
  - At home: transfer immobilised patient, use a hospital bed if necessary
  - If hospitalised: therapeutic stages, approximate durations

### ▶ Specificities in infants

- **Never move a child roughly if he/she is crying or protesting.** Every author refers to the high anxiety in infants, very high with every new fracture (they can be counted by dozens or even hundreds).
- It is essential to **reassure the infant** and to explain the actions taken to him/her.
- **Provide good support for the infant's spine:** hold under the head and buttocks and keep the back straight.
- Try to install a **safe environment** (beds with bars covered in foam for infants to avoid any shock, flexible toys...).
- **The parents often know the best way of moving the child and can indicate the action to be avoided: their involvement is essential in the management even in casualty.**

## Documentary resources

- ▶ Forin V. Ostéogenèse imparfaite. Encyclopédie Orphanet and La Presse Médicale. June 2007  
[www.orpha.net/data/patho/Pro/fr/OsteogeneseImparfaite-FRfrPro654v01.pdf](http://www.orpha.net/data/patho/Pro/fr/OsteogeneseImparfaite-FRfrPro654v01.pdf)

*These guidelines have been prepared in collaboration with Barbara Tourniaire (Hôpital Trousseau), Vicken Topouchian, Geneviève Baujat, Chantal Rostchild and Martine Le Merrer (Hôpital Necker-Enfants Malades) and Nicole Champavier (Association de l'Ostéogenèse Imparfaite).*



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