



Facts about OI for Medical Professionals

OI and Common Medical Tests and Procedures

Introduction

This list summarizes issues of particular concern in treatment settings for people with osteogenesis imperfecta (OI). Medical care should be approached as for any other adult of similar age. No omissions should arise because of concern for OI. Patients with OI vary in height and mobility. This should be considered in exams, procedures and treatments.

In general decreased respiratory reserve, undiagnosed cardiac problems may need to be discussed with the adult person with OI before consenting. Do not hesitate to ask the patient for information about their OI, advice about how to position them for an exam or x-ray or for treatment information based on their experience.

Additional information can be found on the OI Foundation website, www.oif.org.

The OI Foundation is grateful to the medical professionals who contributed information to this chart. Dr. Cathleen Raggio, Hospital for Special Surgery, New York, NY; Dr. Robert Sandhaus, National Jewish Health, Denver, CO; Dr. Jay Shapiro, Bethesda, MD; Dr. David Vernick, Boston, MA; Dr. Leelach DeKoven, Shriners Hospitals for Children-Chicago

Emergency Room

Emergency Room staff members are requested to contact the patient’s PCP or a physician who is familiar with the osteogenesis imperfecta as soon as possible. Adult patients with OI are often an excellent source of information about safe handling and whether a bone is likely to be broken.

Patients with OI come to the emergency room most often for one of 4 reasons:

- Fractures
- Pulmonary Problem
- Cardiovascular Problem
- Accidental Trauma – Falls and car accidents may cause more extensive injury to the person with OI than the same amount of force would inflict on others. It may be necessary to look beyond the simple complaint.

Anesthesia consultation should be considered before any procedure using conscious or complete sedation.

Topic	General Concerns	OI Concerns	Notes
BLOOD WORK & IV PLACEMENT		Thin subcutaneous tissue; Thin walled veins; Contractures and Distorted anatomy	Experienced technician
CARDIOVASCULAR CONCERNS		In addition to myocardial infarction, Heart valve disorders, right-sided heart failure, central & peripheral aneurysms are seen in OI	
CPR		Broken ribs and sternum	No contraindication. Must be performed being aware of chest wall fracture and necessity of obtaining adequate airway.
INTUBATION		Spine and neck deformity and fragile tissues complicate the procedure	

PULMONARY CONCERNS		Restrictive pulmonary disease and infections are major risk factors for people with OI	
RADIOLOGIC PROCEDURES		RODS may interfere with X-rays and MRIs	Determine placement of rods in arms, legs & spine; metal from stapes surgery
X-Rays		Be aware of joint contractures and deformities that make positioning difficult	Do not try to straighten a curved limb
MRI		Rods and stapes metal may interfere (heat up)	Proceed with caution
CAT Scan	Radiation exposure	Concern for adequate positioning	Cumulative radiation dose is a consequence of treating OI and is not a contraindication for any treatment.

COMMON TESTS & PROCEDURES

This list of common tests and procedures is in alphabetical order.

Anesthesia consultation should be considered before any procedure using conscious or complete sedation.

Topic	General Concerns	OI Concerns	Notes
Blood Pressure		Arm curves and fracture interfere with measurement	Measure in a prudent manner; manual cuff preferred; pediatric size cuff may be needed for some adults; Do not avoid testing
BLOOD WORK & IV PLACEMENT		Thin subcutaneous tissue; Thin walled veins; Contractures and Distorted anatomy	Experienced technician

Bone Density/BMD		Radiation risk is minimal	Short stature & contractures may complicate positioning	Assist patient on and off table. Best to compare patient to self; no national norms for OI.
CANCER SCREENINGS				
Mammogram			Short stature; chest deformity	Accommodations for short stature and/or inability to stand
Mohs Surgery			Oozing may be greater than expected.	
Mole Biopsy			Oozing may be greater than average	
Pap/Pelvic Exam			Deformity of pelvis and spine curves may complicate positioning	Pediatric equipment may be needed
ENDOSCOPY			Chest deformity seen in those with more severe OI	Procedure should be done under full staff supervision in an appropriate acute care setting
Hepatitis C			Higher risk if surgery before 1992 because of exposure to untested blood products	Screening is indicated. Treatment is possible and successful.
LITHOTRIPSY			Distorted pelvic anatomy	Appropriate when indicated
Neurological			Headaches secondary to Basilar Impression; Peripheral nerve compression; Neuropathy	Evaluation for Basilar Impression
RADIOLOGIC PROCEDURES			RODS may interfere with X-rays and MRIs	Determine placement of rods in arms, legs & spine; metal from stapes surgery

X-Rays			Be aware of joint contractures and deformities that make positioning difficult	Do not try to straighten a curved limb
MRI			Rods and stapes metal may interfere (heat up)	Proceed with caution
CAT Scan		Radiation exposure	Concern for adequate positioning	Cumulative radiation dose is a consequence of treating OI and is not a contraindication for any treatment.
VACCINATIONS INCLUDING Flu & PNEUMONIA		Follow CDC recommendations.	All are highly recommended. OI is considered a high risk population.	Shot is recommended; the live virus is not an option

ADDITIONAL TESTS AND PROCEDURES

ANESTHESIA

- **Anesthesia consultation should be considered before any procedure using conscious or complete sedation.**
- Dose by the person's height and weight; not just age
- Before surgery get a PFT, neck flexion study, pain medication list and history of reaction to anesthesia
- Pulmonary function tests to include: FVC (forced vital capacity), MVV (Maximum voluntary ventilation) and measure of oxygenation and ventilation such as arterial blood gas
- Take precautions regarding degree of restrictive lung disease

Topic	General Concerns	OI Concerns	Notes
Intubation	Standard Concerns	Head and neck abnormalities complicate airway management and positioning. Abnormalities include megaloccephaly, macroglossia, short neck, mid-face & mandibular deformities, limited range of motion of the cervical spine & dentinogenesis imperfecta. Teeth may be brittle and may break or chip.	Usually, careful positioning and use of a video assist device is sufficient for patient of concern.

Hyperthermia	Patients generally exhibit hypothermia under general anesthesia because of impaired thermoregulation. Warming devices are routinely used to maintain normothermia.	Some reports and some review articles have looked at intraoperative hypermetabolism which is manifested as hyperpyrexia with or without hypercarbia. There is no substantiating evidence to support this finding. Raised temperature may occur, but it is NOT malignant hyperthermia.	It is advisable to monitor temperature as per ASA standards, and use warming and cooling devices as necessary to maintain normothermia. OI DOES NOT have an associated increased risk with malignant hyperthermia.
Access	IV's can be challenging to place.	Positioning and existing fractures and bony protrusions may make IV access more difficult. Risk of causing a fracture.	Tourniquets can be used for IV access but only with special care and attention during the positioning process.
Neuroaxial and/or Regional Anesthesia	Standard concerns	Bone and Spine deformities may make these anesthetic adjuncts more challenging.	Regional anesthesia and Neuroaxial anesthesia have been safely used for OI patients. Care should be used when positioning.

GASTROINTESTINAL

- Tissue may be more fragile
- Issues that may be evaluated include swallowing, stomach ulcers, constipation and various digestive complaints.

Topic	General Concerns	OI Concerns	Notes
COLONOSCOPY		Risk of perforation of colon or bow; Distorted anatomy	Consider virtual colonoscopy Consider stool sample kit
ENDOSCOPY		Chest deformity seen in those with more severe OI	Procedure should be done under full staff supervision in an appropriate acute care setting

HEARING

- Hearing Loss is seen in about 50% of adults with OI and may get worse over time.
- Discuss ototoxic drugs and ways to protect one's hearing.

Topic	General Concerns	OI Concerns	Notes
Audiology Exam	Hearing screening should start in childhood	General exam – normal appearance but drum may appear thin and translucent	May notice some increase in vascularity in middle ear
Cerumen (Wax) removal		No different than anyone else	Note slanting canal
HEARING AIDS	Work well May need earlier in life	Loss is conductive and sensorineural in most; Loss is progressive so yearly checks are important	Work just as well in OI Start hearing monitoring in childhood
Bone Anchored	Is bone strong enough to hold implant?	Bone likely not strong enough to hold the implant effectively (not recommended in OI)	Not recommended
Cochlear Implant	When hearing aids are not enough	Good results in OI Need to be adjusted to account for the decrease in bone density	Small studies indicate that the surgery can be successful
Stapedectomy	May be option if conductive loss is major part of hearing loss	Success rate not as high as with non-OI surgery. Surgery is more difficult. Increased risk of hearing loss with surgery.	Greater success if physician is experienced with procedure and with bone fragility

HEART

- Baseline echocardiogram is suggested for young adults especially if there is a family history
- Monitor for valve disease if there is a family history
- Rate for hypertension is believed to be similar to that seen in others but some small studies suggest that it may be slightly higher.

Topic	General Concerns	OI Concerns	Notes
ECHOCARDIOGRAM		Chest wall deformities may make positioning difficult	A trans-esophageal echocardiogram may be needed for some patients
HYPERTENSION			Medication may need to be adjusted to lower dose once readings stabilize

PULMONARY TESTING

- Begin with a chest x-ray to evaluate chest architecture
- Periodic pulmonary function tests are essential
- Lung connective tissue is altered in all types of OI
- Any endoscopic procedure must be done with advanced information about potential risks e.g. hypoventilation, bronchospasms.
- Severity of symptoms is affected by the individual's height, presence of spine curves.
- Sleep Apnea is a concern but the incidence is unknown.
- Pulmonary Function Test results must be compared to individuals' previous values rather than calculated norms.
- Predicted values are unreliable for those with short stature.

Topic	General Concerns	OI Concerns	Notes
Arterial Blood Gas (ABG)	Assure good collateral circulation before drawing arterial blood.	Limbs distorted by multiple fractures and surgery may make artery hard to find and stick.	Significance of results the same as those without OI.
Pulmonary Function Tests (PFT) and Spirometry	Can be difficult to perform in children and people with severe lung disease.	Added difficulties due to positioning issues, especially for body box lung volumes.	Compare patient to self because predicted values (based on height) are unreliable for patients with short stature.

Pulmonary Rehab	Insurance reimbursement is low or absent	Low strength; short stature; fracture risk.	Beneficial Adapt equipment and exercises as needed
Sleep Study	Sleep problems are often overlooked by medical professionals	Positioning may be difficult. Adequate study with good position is needed	Meet with center ahead of time to assess ability to work with a person of short stature with spine curves. Custom fitted mask for CPap or BiPap may be needed due to facial deformities. Bring pillows and bolsters from own bed to study site.
Oximetry	Measures percent of hemoglobin carrying oxygen; does not measure other values such as CO2 levels, pH, and CO levels. Need ABG for that information.	None	Excellent, quick and inexpensive way to measure oxygenation.
CT Scan of Chest	Significant radiation dose	Positioning for test can be difficult. Interpretation of results can be difficult because of distorted chest architecture.	

SURGERY

Elective surgery plan should include a pre-op anesthesia consultation.

In addition pulmonary and cardiology consultations as indicated by the patient's general health and the procedure.

Persons with severe OI may have limited reserves.

Topic	General Concerns	OI Concerns	Notes
Bleeding		Bleeding may be greater and average	Review history of bleeding during prior surgeries. Use techniques to minimize blood loss.