

## Facts about Osteogenesis Imperfecta for Medical Professionals

### Care for the Adult Patient with OI

Osteogenesis imperfecta (OI) is a lifelong systemic connective tissue disorder. Although the hallmark is fragile bones, this connective tissue abnormality is expressed in many organs besides the skeleton. The combination of OI's variable series of issues and normal aging requires that medical care be **approached in an individual yet comprehensive manner**. A care plan for an adult who has OI will need to address issues and concerns of clinical importance in OI plus the health issues occurring in individuals of the same age who do not have OI. This document addresses needs of patients with all types of OI from mild to severe.

**Pulmonary issues are the major cause of death in adults who have OI. Lung tissue is affected in every type of OI including Type I (mild) leading to an increased incidence of respiratory infections and other breathing problems. Monitoring of respiratory function and aggressive treatment for respiratory infections are mandatory. Evaluation for hypoxemia, for sleep-disordered breathing and the need for supplemental oxygen should be considered. Issues may be more serious in people with more severe forms of OI.**

Most adults with OI experience a decrease in the rate of fractures after puberty, but may see an increase in tendon, muscle and joint problems. Fracture rate may begin to increase again around age 40 when bone mineral density decreases with hormonal and age-related changes in bone turnover. Issues that may become more serious after childhood include progressive hearing loss, development of heart disease including valve disorders, hypertension, kidney stones, chronic pain, gastrointestinal complaints including constipation, ulcers, dental issues, obesity, spine curves, and craniocervical deformities such as basilar impression.

#### Use of Bone Building Drugs

Adults who have OI may be treated with drugs such as bisphosphonates, denosumab (Prolia®) and teriparatide (Forteo®) that were developed to treat age-related bone loss. These drugs increase bone mass, but do not improve bone quality or address the non-skeletal features of OI. In studies, the effect on fracture frequency is not consistently significant. Those with mild Type I may benefit from Forteo® if they are sustaining fractures or losing bone mass. Treatment for all types of OI should be individualized based on bone turnover markers, declining bone mass, fracture frequency and treatment for other conditions such as cancer.

#### General Advice

- Know your patient's OI history including OI type, surgical placement of rods and other hardware, and history of previous or current use of medications.
- **Assess the pulmonary function of adult patients with OI at each visit and prior to any medical procedures. Underlying pulmonary problems may increase the risk of many common medical procedures and anesthesia.**
- At the annual physical:
  - As customary, examine the patient in a gown and on the exam table.
  - Include a review of systems as for others; the OI adult's basic health needs are the same as other adults.
- Review all medicines and supplements as some may contribute to bone loss.
  - Dosage may need to be adjusted for short statured adults based on weight and height.
  - Monitor use of NSAIDS as they can delay bone healing after fracture.
  - Limit use of fluoroquinolone antibiotics due to risk of tendon rupture.
- Healthy behaviors to encourage: healthy diet including lipid control, weight control, physical activity, and smoking prevention and/or cessation.
- Immunizations including flu and pneumonia vaccines are highly recommended.
- Establish a plan so the patient and ER personnel can reach you in case of an emergency.

This fact sheet was prepared with assistance from Dr. Jay Shapiro, Bethesda, MD, Dr. Robert Sandhaus, Denver, CO and Dr. Cathleen Raggio, New York, NY, April 2016.

### Specific Considerations

- Conditions that may be made worse due to OI: asthma, osteoporosis, COPD, sleep apnea, dental issues, hearing.
- Conditions that may be masked by OI: arthritis, vocal cord problems.
- Conditions that appear to occur often in adult OI: anxiety disorders, reversible and irreversible airway obstruction and other breathing problems, non-union of fractures, glaucoma, hearing loss, heart valve and aortic disease, hypertension, pain syndromes, basilar impression, tendon and ligament injuries.
- Conditions that appear to occur more often in adults with more severe OI: shortness of breath and sleep apnea.
- Areas where bone, blood vessel and tissue fragility related to OI may complicate treatment or testing: cancer surgery, pregnancy, heart valve surgery, urinary tract infections, colonoscopy, ulcer diagnosis, other endoscopic procedures, and intubation for surgery.
- Areas where the effect of OI is not well understood: gastrointestinal function (includes GERD, constipation, celiac disease), heart problems and kidney stones.

### Annual Physical

1. Gastrointestinal – assess for GERD and chronic constipation.
2. Hearing – assess if referral to audiologist is needed starting around age 20.
3. History of kidney stone.
4. Orthopedic – assess scoliosis and other chest or back deformity; limb length discrepancy; osteoarthritis in joints.
5. Pain assessment– changes in general pain level or new pain may not be related to OI.
6. Pulmonary Function – spirometry and oximetry or arterial blood gas. Fatigue may be a symptom.
7. Dermatology – signs of skin cancer.
8. Cardiovascular Health – EKG; auscultation for valvar and aortic disease, carotid artery stenosis.
9. Neurologic Exam – for symptoms related to basilar impression, limb length differences, changes in muscle strength; unequal gait, balance, risk for falling and evaluation of back pain.
10. Note women’s menstrual status.

### Additional Testing when Indicated

1. Bone Density Testing (DXA) every 3 years (may vary with treatment).
2. Echocardiogram as a baseline to assess aortic or mitral valve disease and pulmonary artery pressure; annually if necessary.
3. Complete metabolic panel with fasting blood sugar, complete blood count, lipid panel, and HbA1c.
4. Testing for Hepatitis C for those who had surgery before 1992.
5. Assess calcium and vitamin D status as needed.

### Additional Medical Consultations as Indicated

1. Ophthalmology – general vision testing; glaucoma; retinal detachment
2. Audiology – track progress of hearing loss; hearing aid assessment; surgical treatments
3. Gastrointestinal – chronic constipation, GERD, celiac disease
4. Urology – UTIs, renal stones/ nephrocalcinosis
5. Neurology – back pain, evaluation for basilar impression, peripheral neuropathy due to spinal or nerve involvement secondary to fracture or limb deformities
6. Physical Therapy – gait and limb assessment; function assessment; strength and ADL assessment
7. Occupational Therapy – ADL assessment; mobility aide adjustment
8. Dermatology – skin cancer; rosacea
9. Mental Health – substance abuse; depression
10. Dental evaluation

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11. Pulmonologist and/or Sleep Specialist – Complete pulmonary function tests (PFT) pre- and post-bronchodilator, formal or in-home sleep study, evaluation for vocal cord dysfunction (VCD)

For more information on osteogenesis imperfecta visit the OI Foundation website section for Medical Professionals [www.oif.org/MedEd](http://www.oif.org/MedEd)