

Facts about Osteogenesis Imperfecta

Pulmonary Issues

Respiratory complications are a leading cause of death for children and adults who have OI, especially those with severe or Type III. Although respiratory problems are usually more severe in those with severe OI, all types of OI, including Type I (Mild) are affected to some degree. The primary collagen defect affects lung tissue in all people with OI, including those with a mild phenotype. Altered lung tissue predisposes the patient to respiratory infections. Rates of asthma and pneumonia are higher in children and adults who have OI than in the unaffected population. Chest wall deformities, rib fractures, scoliosis, kyphoscoliosis, chest wall muscle weakness, chronic bronchitis or asthma, limited mobility and the effects of gastrointestinal problems such as constipation and reflux all contribute to poor pulmonary function. Fatigue, breathlessness and wheezing are frequent symptoms. Manifestations can include asthma, recurrent pneumonia, exercise intolerance, pulmonary hypertension, and sleep apnea.

Many infants with OI are at risk for aspiration with swallowing. Gastroesophageal reflux can also lead to aspiration and/or trigger asthma in infants and children.

Chest wall architecture is a major contributing factor to pulmonary function problems in children. Adults may develop new or worsening problems over time due to over-weight, loss of mobility and progressive scoliosis. Evaluating lung disease by radiographs, pulmonary function tests or sleep studies may be helpful in watching progression of respiratory issues in OI. Testing can be difficult due to anatomical differences associated with OI.

Treatments may include bronchodilators, airway clearance devices as appropriate for OI, supplemental oxygen, bi-level positive airway pressure, and pulmonary rehabilitation.

Recommendations to patients who have OI should include:

- Do not smoke and avoid second hand smoke.
- Promptly seek care for all respiratory infections.
- Maintain good hydration.
- Keep upper body strong.
- Avoid obesity.
- Get flu and pneumonia immunizations.

References

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This fact sheet was prepared with assistance from Dr. Hollis Chaney, Children's National Medical Center and Dr. Robert Sandhaus, Colorado School of Medicine, February 2015.