



Respiratory Issues in Osteogenesis Imperfecta

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The Osteogenesis Imperfecta Foundation, Inc. is the only voluntary national health organization dedicated to helping people cope with the problems associated with osteogenesis imperfecta. The Foundation's mission is to improve the quality of life for people affected by OI through research to find treatments and a cure, education, awareness, and mutual support.

Introduction

The respiratory system's job is to bring oxygen into the body and remove carbon dioxide, the waste product of breathing. Because oxygen is the fuel needed by all cells and all organs in the human body, getting enough of it is important for good health. Breathing problems can lead to shortness of breath, tiredness, insomnia, headaches, sleep disturbances and can make a person more susceptible to infections such as pneumonia. Respiratory illnesses can be very serious, even life threatening health problems for people with OI.

OI Affects the Lungs

The primary respiratory problem affecting people with OI is loss of lung capacity. Other problems include ineffective cough, poor secretion clearance, airway diseases such as asthma, sleep apnea, and low oxygen. These problems affect people of all ages and all types of OI. Respiratory infection, poor oxygen delivery, and other respiratory disorders may lead to respiratory failure and death particularly in people with OI Type III, or in people with chest wall deformities, or reduced respiratory function.

Viral infections (colds and flu), bacterial infections (bronchitis and pneumonia) and allergies are common respiratory problems. Even when lung problems are not directly caused by OI, they can be more severe in people who have OI. It is a sobering fact that respiratory failure is the leading cause of death for people with OI.

Causes of Lung Problems in OI

There are two main causes for lung problems specifically related to OI:

1. Abnormal chest wall architecture (the size and shape of the chest cavity)
2. Abnormalities of lung collagen

Chest wall and spinal deformities are caused by:

- Kyphoscoliosis and or scoliosis – spine shape problems
- Rib fractures
- Pectus deformities – protruding or sunken breast bone or sternum

Short stature can make the impact of changes in chest shape more serious. Deformities of the spine and ribs limit the space available for the lungs to expand. This decreases their ability to effectively inhale and exhale.

Abnormal lung collagen has not been thoroughly studied and is not well understood. We know that type I collagen is a major component of lung connective tissue just like it is a major part of bones and other connective tissue. We know that abnormalities in lung collagen occur in OI. So far only one or two studies have looked at the effects of abnormal lung collagen. In one study, people with OI and no chest wall deformities still had abnormal lung function. (Widmann R.F., F.D. Bitan et al. (1999). "Spinal deformity, pulmonary compromise, and quality of life in osteogenesis imperfecta." Spine 24(16): 1673-8). This information helps explain why people with even relatively mild OI seem to have higher rates of asthma and pneumonia than people who do not have OI.

Low Oxygen Causes Severe Problems

The lungs transfer oxygen from the air you breathe into the blood. Oxygen rich blood circulates throughout your body. Without oxygen, other parts of your body cannot work correctly. In addition, persistent low oxygen in the lungs can cause some severe problems including the following:

- Pulmonary hypertension (high blood pressure in the pulmonary blood vessels)
- Right heart failure (inability of the right side of the heart to move blood forward through the lungs, often leading to fluid collection in legs and abdomen)
- Cor pulmonale (the combination of pulmonary hypertension and right heart failure)

What You Can Do to Minimize Your Risk of Lung Problems

- First and foremost **do not smoke, and avoid second hand smoke.** Children with OI should never be exposed to second hand smoke.
- Promptly seek care for all respiratory infections and aggressively treat all respiratory infections. When there is poor lung function, a "cold" can progress quickly to bacterial bronchitis or pneumonia.
- Seek prompt treatment for any breathing difficulties.
- Prevent infection. Washing your hands and avoiding contact with people who obviously have a cold or "the flu" have been proven to be simple but effective ways to stay healthy.
- Keep the upper body strong. Effective breathing requires strong muscles. Talk to your doctor and physical therapist about an exercise program to help you improve your lung capacity.
- Get an annual flu shot, and talk to your doctor about whether you're a candidate for the pneumonia vaccine.

- Promote good secretion clearance by staying well hydrated and speaking to your doctor or respiratory therapist about special coughing techniques and equipment.
- Ask your doctor about whether you should have a sleep study. People with OI seem to have a very high frequency of sleep-related breathing problems.
- Consult with your doctor about preventing and treating chest wall deformities and spine curves.
- Attentively manage your other underlying lung problems such as asthma.
- Monitor your lung function and the amount of oxygen in your blood (oxygenation). These tests should be part of your regular physical exams. Most commonly oxygen levels are tested by doing an “oximetry,” placing a special sensor on the finger. When lung disease is severe, it may be more appropriate to draw blood from an artery in the wrist and do an arterial blood gas or ABG test. Sometimes supplemental oxygen or a positive pressure breathing device such as CPAP or BiPAP will be prescribed to help manage breathing problems.

Pay attention to your lungs! Effectively managing lung problems in OI will improve quality of life and survival.

This fact sheet is based on information provided by Dr. Robert Sandhaus, MD, PhD. Dr. Sandhaus is a respiratory specialist who has worked for many years with people who have Alpha-1, a genetic disorder that causes severe breathing problems. He is a member of the OI Foundation Board of Directors, Medical Director for the Alpha-1 Foundation and works out of the National Jewish Medical and Research Center in Denver, CO.