THE LUNGS IN MARFAN SYNDROME

Many people with Marfan syndrome and some related disorders experience pulmonary complications, or problems with their lungs. If you have Marfan syndrome, it is important for you to see a pulmonologist (lung doctor) if you suspect that you have any problems with your lungs.

About 70 percent of people with Marfan syndrome also have restrictive lung disease.

Lung problems in people with Marfan syndrome can be the result of many different factors including:

- Scoliosis (curved spine) or pectus (chest bone) abnormalities that can restrict lung function and cause soft tissue inflammation
- Weak respiratory muscles that can restrict lung function
- Underdeveloped lungs with enlarged air spaces that can cause sudden lung collapse (spontaneous pneumothorax) and emphysema
- Dysfunctional airways that cause asthma or chronic bronchitis

What are the common types of lung problems in people with Marfan syndrome?

Lung problems that can develop include:

Restrictive Lung Disease

About 70 percent of people with Marfan syndrome also have restrictive lung disease. Restrictive lung disease makes it difficult for the chest to expand and for you to take full breaths. That, in turn, makes it difficult for your body to

take in the amount of oxygen it needs. If you have Marfan syndrome, restrictive lung disease can be a result of muscle weakness or a result of structural issues such as scoliosis, kyphosis, or severely indented chest bone that can reduce the space available for the lungs and interfere with their ability to expand. Restrictive lung disease makes breathing more difficult, and may cause shortness of breath during mild activity, coughing, wheezing, or chest pain.

Sudden lung collapse (spontaneous pneumothorax)

One possible effect of Marfan syndrome is lung collapse (pneumothorax). While this condition is usually not life-threatening, it is an emergency.

Lung collapse happens when air escapes the lung into the space between the lung and the inner lining of the chest. When there is too much air inside the chest cavity, pressure builds up on the lungs, which can cause the lung to collapse. Typically, spontaneous pneumothorax is caused by the bursting of over-expanded lung air sacs call apical blebs or bulia. These blebs are weakened areas of the lungs. If the bleb breaks open (ruptures), it can send air into the space around the lung, causing the lung to collapse. In the Marfan lung, sudden lung collapse can be recurrent, present in both lungs, and/or associated with emphysema.

The symptoms of lung collapse are shortness of breath, a dry cough, and often sudden onset of pleuritic chest pain (pain that gets worse when you take a deep breath). The pain may be confused with an aortic dissection or heart attack, and requires evaluation in an emergency room. In a large pneumothorax, a person may have a bluish skin color due to the lack of oxygen, chest tightness, easy fatigue, and rapid heart rate.

Emphysema

Emphysema is a condition in which the walls of the tiny air sacs in the lungs are damaged so they cannot push all the used air out of the lungs. Approximately 10–15 percent of people with Marfan syndrome have emphysema, but it may be under-diagnosed. Symptoms include shortness of breath during activity, frequent bronchitis (often as a result of common colds or viruses settling in the chest), and low blood oxygen. The diagnosis can be confirmed by a chest x-ray, CT scan, pulmonary function test, or arterial blood test.

Asthma

Asthma is a chronic (long-term) lung disease that inflames and narrows the body's airways. A correct diagnosis of asthma is important for a person with Marfan syndrome because many of the drugs that are used to treat asthma (beta-agonists) can counteract the effects of other drugs that people with Marfan syndrome need to use, such as beta-blockers, to control and slow aortic growth. If you suspect you may have asthma, it is important for you to see a respiratory specialist who can coordinate treatment with your other doctors.

Sleep apnea

Some people with Marfan syndrome have sleep-disordered breathing (sleep apnea), which can have a number of causes. One seems to be laxity of the connective tissue of the airways, which then further relax during sleep and cause partial obstruction to air flow.

Among people who are the same age, weight, and height, sleep apnea is more prevalent in people who have Marfan syndrome than in those who don't. Unfortunately, sleep apnea is often underdiagnosed in the Marfan community. In the general population, the majority of people who are

diagnosed with sleep apnea are overweight. People with Marfan syndrome usually aren't overweight, so sleep apnea is frequently overlooked or not suspected as a diagnosis.

Sleep apnea is thought to be more common in the Marfan community because of head and facial abnormalities, such as floppy airway tissue, high arched palate, and retrognathia (a deformity of the jaw where the jaw is small, or recessed, which can cause the airway to be obstructed).

Studies have shown that untreated sleep apnea can cause aortic wall stress, a particular concern in Marfan syndrome. This makes it extremely important to diagnose and treat sleep apnea if it is suspected. If you suspect you have sleep apnea it is important to see a pulmonologist (lung doctor) to receive the correct diagnosis.

The symptoms of sleep apnea are chronic (ongoing) and loud snoring, pauses in snoring that are followed by choking or gasping, and snoring that worsens when sleeping on your back and lessens when sleeping on your side. Often, family members or a bed partner will notice these symptoms before you do. Other symptoms of sleep apnea are sleepiness during the day, at work, or when driving, morning headaches, and dry mouth or sore throat when you wake up. Sleepiness occurs in a person with sleep apnea because when breathing pauses, or when there are shallow breaths during sleep, a person goes from a deep sleep to a lighter sleep causing a poorer quality of sleep.

How are lung problems treated in people with Marfan syndrome?

Restrictive lung disease

Apart from early correction of curvature of the spine (scoliosis), which does improve respiratory function, other surgical measures to normalize the dimensions of the chest wall do not always help. Thus, decisions about treatment options should be individualized to address a person's specific cosmetic and functional concerns.

Restrictive lung disease can be worse if another airway disease, such as asthma or emphysema, is present. Supplemental oxygen and pulmonary rehabilitation are recommended to improve the quality of life.

Pulmonary rehabilitation is an array of activities and therapies, such as nutritional counseling, energy-conserving techniques, and breathing strategies involving specialists like respiratory therapists, physical and occupational therapists, dietitians or nutritionists, and psychologists or social workers. The rehabilitation program is individualized to optimize physical and social performance and is appropriate for anyone with breathlessness that affects his or her quality of life. The goals of respiratory rehabilitation are typically to reduce symptoms of shortness of breath and increase the ability of the lungs to function properly. Lung tests that measure lung functioning may not change despite an improvement in quality of life.

Lung collapse (pneumothorax)

A doctor who examines the chest with a stethoscope may hear decreased or absent breath sounds over the affected lung. The diagnosis is confirmed by a chest x-ray or chest CT scan, and arterial blood gases (measures the amount of oxygen and carbon dioxide in the blood). The size of the spontaneous pneumothorax will determine the probable course of the condition and the best treatment.

If you experience any of the symptoms of pneumothorax, it is important that you receive immediate medical attention to detect the severity of the lung collapse. You also need to tell the surgeons that you have Marfan syndrome and may need aortic surgery in the future. This will help the surgeon make an appropriate treatment recommendation.

A "small" lung collapse is treated in the hospital with supplemental oxygen and rest. In some cases a health care provider will use a needle to pull the extra air out from around the lung so it can expand fully. Often a small pneumothorax will resolve itself without any treatment.

A "moderate to large" pneumothorax is treated by chest tube evacuation (inserting a tube between the ribs into the space around the lungs to help drain the air, allowing the lung to re-expand) and possibly pleurodesis if the lung does not re-inflate on its own. Pleurodesis is a surgical procedure that involves "scarring" the lung surface to attach the lung to the chest wall. It is helpful in treating pneumothorax and also helpful in preventing another pneumothorax from happening.

The best pleurodesis method for people with Marfan syndrome is mechanical, rather than chemical, pleurodesis. A mechanical pleurodesis is performed manually by a surgeon. The surgeon gently strokes the pleura (the thin tissue that lines the chest cavity and surrounds the lungs) with a piece of gauze. Mechanical pleurodesis roughens up the pleura so that, when the abrasion heals, the lung adheres to the chest wall. Mechanical pleurodesis is the preferred method for people with Marfan syndrome because it makes future cardiac surgery easier if needed in the future. Physicians who care for people with Marfan syndrome should assume that all Marfan patients eventually need aortic replacement.

Emphysema

The conventional treatment for emphysema is supplemental oxygen, bronchodilator therapy (medication that opens up the bronchial tubes), and aggressive treatment of infections. Research is underway on other methods that may be useful.

Asthma

Most people with Marfan syndrome take drugs called beta-blockers to help ease the pressure on the aorta. However, beta-agonists, which have the opposite effect, are the conventional treatment for asthma. Since beta-blockers should not be used for children with severe asthma or reactive airway disease, you should speak to your doctor about other options.

Sleep apnea

Sleep apnea is diagnosed by sleep studies and a physical examination. Sleep studies are tests that measure how well you sleep and how your body responds to sleep problems. A doctor may suggest a sleep study called a polysomnogram or a home-based portable monitor.

A doctor's physical exam involves checking for extra or large tissue. Children who are suspected of having sleep apnea may have enlarged tonsils. Adults who are suspected of having sleep apnea may have an enlarged uvula (tissue that hangs from the middle of the back of your mouth) or soft palate (the roof of your mouth at the back of your throat).

A polysomnogram records the amount of oxygen in your blood, the air movement through your nose while you breathe, snoring, and chest movements. A PSG is often done at a sleep center or sleep lab. A home-based sleep test with a portable monitor will measure some of the same things

that a polysomnogram measures, but instead of being done in a sleep center or sleep lab, a home-based sleep test can be done in the comfort of your own bed.

For mild cases of sleep apnea, the treatment may include some lifestyle changes and a mouth-piece, called an oral appliance. The oral appliance lowers your jaw and your tongue to keep your airways open while you sleep.

Continuous positive airway pressure (CPAP) is considered by many experts the most effective treatment for moderate to severe cases of sleep apnea. A nasal CPAP machine provides supplemental oxygen. A mask, worn over the nose and/or mouth while you sleep, is hooked up to a machine that delivers a continuous flow of air into the nostrils to gently blow air into your throat. The positive pressure from air flowing into the nostrils helps keep the airways open while you sleep so that breathing is not impaired.

In some cases, people with Marfan syndrome have trouble finding a CPAP mask that fits comfortably so they have to try several masks to find one that works. They may also require a special type of mouthpiece (mandibular advancement device) or other gadget to help the mask fit properly.

Some people who have sleep apnea can benefit from surgery. Surgery is done to widen breathing

passages. It usually involves shrinking, stiffening, or removing excess tissue in the mouth and throat or resetting the lower jaw. It may be helpful to have surgery to remove tonsils in children if the tonsils are blocking the airway.

What can you do on a day-to-day basis?

There are several things that you can do on a day-to-day basis to care for your lungs when you have Marfan syndrome. And while it is natural to avoid thinking about emergencies like sudden lung collapse, understanding your condition and preparing in advance to handle emergencies can help you be more confident every day that you and your family know what do should the need arise. Taking care of yourself in this way is an important part of living with Marfan syndrome or a related disorder.

Here are some steps you and your family can take on a routine basis, and also when emergencies arise.

Caring for Your Lungs

Routine care

- Visit a pulmonologist to receive a pulmonary function test.
- Follow your treatment plans for the management of any lung problems you have.
- Avoid or quit smoking. If you have Marfan syndrome or a related disorder, the potential for lung problems should be a powerful incentive for you to avoid or quit all forms of smoking.

Sudden lung collapse

- Avoid activities that risk rapid changes in atmospheric pressure. Since the Marfan lung is more prone to collapse, it is important to avoid activities that risk rapid changes in atmospheric pressure, such as skydiving, scuba diving, or traveling in an unpressurized aircraft.

- If you have any of the signs or symptoms of a collapsed lung, seek immediate medical attention to detect the severity of the lung collapse.
- Be prepared in the event of an emergency. Complete our Emergency Preparedness Kit, downloadable from our website, and bring it with you to the emergency department should the need arise.

Sleep apnea

- Avoid alcohol and medicines that make you sleepy. They make it harder for your throat to stay open while you sleep.
- Sleep on your side instead of your back to help keep your throat open.
- If you smoke, quit smoking.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.

Figure 1. The Lung