

Muscle weakness is one of the problems that people with cystinosis may experience. Occasionally, the muscles used for breathing are weak and cause symptoms. The most common sign of respiratory muscle weakness is becoming short of breath with physical activity. If someone with cystinosis notices this symptom, he/she should report this to his/her doctor. However, there are many problems unrelated to cystinosis that can cause shortness of breath with activity, so an evaluation needs to be performed. One of the ways to detect muscle weakness is pulmonary function testing that measures the amount of air someone can hold in their lungs and how hard they can suck in and blow out. Additional tests may be recommended including standard x-rays or CT scans of the chest to look for other lung problems. Occasionally tests are done such as an electrocardiogram or an echocardiogram to make sure that heart function is okay and not contributing to the shortness of breath.

If it appears that muscle weakness is causing shortness of breath, supportive treatments are available. When the problem is severe (as measured by symptoms, pulmonary function tests, and blood levels of oxygen and carbon dioxide), it may be recommended that the person use a device to assist their breathing at night and during the day as needed. The device is called BiPAP and consists of a face mask that usually goes over the nose and is attached to a machine that blows air under low pressure to help inflate the lungs. This machine works to insure adequate breathing at night and to rest the muscles used for breathing. An exercise program may also be beneficial. Many medical centers have pulmonary rehabilitation programs that provide guidance in types and levels of exercise that are useful for people with pulmonary problems. To help decide if these treatments are needed, the cystinosis doctor can consult with a pulmonary expert who has experience in evaluating and treating patients with respiratory muscle weakness.

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