

**Take home point:** *Start getting a baseline FVC at age of 6 years, this means a pulmonary referral. Get an FVC every year to track if there are any breathing problems. Educate the person performing the test that you would like the FVC to be checked in 2 positions: sitting and lying down.*

In CMD, there may be an early onset of breathing problems. This is not asthma, but rather a weakness of the chest wall muscles and diaphragm. When breathing problems go undetected they can lead to a slow gradual rise in carbon dioxide that can cause the following symptoms: daytime headaches, restless sleep, difficulty concentrating. However these are late findings and the onset is so gradual that most are unaware.

To detect a breathing problem early: it is important to see the pulmonologist annually for a specific test called the Forced Vital Capacity (FVC). The FVC is one part of pulmonary function testing. It is the key test to track annually to determine if there is a change in your or your child's breathing status. The FVC is either reported in *liters* or as a *% of the predicted FVC* for someone with the same height and weight. It is easy to follow the FVC%. An FVC greater than 60% is normal, less than 60% is abnormal.

Some forms of CMD have diaphragm involvement. The best way to detect breathing problems when the diaphragm is involved is to perform the FVC in 2 positions, sitting and lying down (supine). When a person is sitting, gravity assists the diaphragm in pulling down and FVC may appear normal. However, when a person is lying down, gravity cannot help, and the FVC will drop in the setting of diaphragmatic weakness. Depending on the % difference between sitting and supine FVC, a sleep study may be ordered to confirm difficulty with breathing and identify a need for noninvasive ventilation at night (bipap NOT CPAP). Starting bipap young is possible. It requires patience and finding the right bipap mask interface. Most kids tolerate nasal interface better than full face mask interface, yet, some pulmonologists are hesitant to prescribe the nasal interface.

For individuals with Ullrich CMD, Merosin Deficient CMD, L-CMD and SEP1 we recommend the breathing test is always done in both the sitting and lying down position to detect an early drop in FVC.

A sleep study or polysomnogram requires a night in a hospital where an individual's sleep will be monitored. The doctors check the oxygen level and carbon dioxide level. In some countries, a decision to start BiPAP is made without a sleep study as it may be difficult to obtain. This decision is made upon how low the FVC is and a blood gas measurement of carbon dioxide.

It can be difficult to get an FVC in kids who have poor oral motor tone or have a difficult time forming a seal around the mouthpiece. In these children, the FVC test can be adapted to using a face mask.