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Hypoventilation? Obstructive Sleep Apnea? Different Tests, Different Treatment

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People with neuromuscular disorders may be misdiagnosed and mistreated when they encounter breathing and sleep problems. Many general practitioners, and even some pulmonologists, neurologists, and sleep physicians, may not fully understand respiratory insufficiency and physiology in this group.

Hypoventilation. Generally, in people with neuromuscular disorders who are having breathing problems, the main problem is hypoventilation (underventilation) - not breathing deeply and/or often enough. Muscle weakness, scoliosis and/or chest wall stiffness make it difficult or impossible to fully inflate the lungs.

Hypoventilation results in an imbalance in the carbon dioxide (CO₂) and oxygen (O₂) exchange in the blood - too much CO₂ is retained, too little O₂ is taken in. Because hypoventilation usually first occurs during sleep and because several of

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the signs and symptoms overlap, it can be misdiagnosed as obstructive sleep apnea (OSA).

Although any trained health care professional can perform simple pulmonary function tests (PFTs) of breathing ability during an office visit, the tests are most likely to be performed by a pulmonologist, neurologist, nurse, or respiratory therapist. The challenge lies in understanding the results of these tests in the context of a person with neuromuscular disease.

Two important measurements of your ability to breathe deeply are the forced vital capacity (FVC) and maximum inspiratory pressure (MIP or PiMax). The SNIP (sniff nasal inspiratory pressure) test has been shown, in some studies, to be a more sensitive test of respiratory muscle weakness, but it is not widely used in the USA.

Forced vital capacity measures the volume of air you can breathe in and then blow out quickly and completely through a device called a spirometer. It should be measured in both the upright and supine (lying face-up) positions, because you can't breathe as efficiently lying down.

Another simple test that measures the strength with which you can breathe in is the MIP. A mouthpiece is attached to a negative pressure gauge via a narrow tube. With a noseclip pinching off the nostrils, you exhale and then suck on the mouth-piece as hard as possible; the gauge registers the pressure.

A result of <50% predicted FVC or a MIP <60 cm H₂O may signal that it's time to get some assistance with breathing.

However, the most important factor in diagnosing hypoventilation is an elevated level of CO₂ (above 45 mm Hg). This can be measured invasively with an arterial blood gas (ABG) analysis or noninvasively using exhaled end-tidal CO₂ monitoring or transcutaneous CO₂ monitoring.

The pattern seen on an overnight oximetry tracing may also be helpful for identifying early hypoventilation often seen first during the deepest rapid-eye movement (REM) sleep stage.

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Signs and symptoms of nocturnal hypoventilation may include one or more of the following:

- fatigue or exhaustion after normal activity;
- excessive daytime sleepiness;
- shortness of breath, breathlessness with minimal activity;
- claustrophobia or feeling that air in room is somehow bad;
- difficulty in speaking for more than a short time;
- quiet speech with fewer words per breath;
- inability to lie flat while awake due to shortness of breath;
- inability to lie flat during sleep/ need to sleep sitting up (orthopnea);
- trouble falling asleep and trouble staying asleep;
- anxiety about going to sleep.

Other signs and symptoms, which may also be seen in OSA, include:

- excessive daytime sleepiness and need to nap during the day;
- nightmares, night sweats, bedwetting, or need to urinate frequently;
- morning headaches;
- restless/fragmented sleep with frequent awakenings;
- shallow breathing or cessation of breathing for 10 seconds or more;
- awakening from sleep with choking sensation;
- worsening mental status, impaired memory, concentration, cognition.

Do not ignore these signs and symptoms hoping they will go away. They are serious. You may need evaluation and treatment immediately!

The treatment for hypoventilation is NOT oxygen but assisted ventilation, generally at night, with a bilevel ventilator. Bilevel units that offer the S/T mode (the unit operates in a spontaneous -S- mode, meaning the user can spontaneously initiate each ventilator breath, but switches to a timed -T- mode, referred to as the backup rate, when breaths are not initiated by the individual) are recommended for people with neuromuscular disorders.

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Bilevel ventilators provide pressure support ventilation which is achieved by the difference in two set pressures: IPAP (inspiratory positive airway pressure) and EPAP (expiratory positive airway pressure). The IPAP and EPAP pressure settings can be adjusted separately.

People with neuromuscular disorders have more trouble breathing in. They generally need IPAP that is set at least 5-10 cm H₂O higher than EPAP and EPAP that is set at the minimum level. Higher EPAP makes it too difficult for them to exhale. “In my home care company, we start out people new to bilevel with ‘training wheels’ - a minimum span of 5 cm H₂O. After they become acclimated to the treatment, we increase the span if the individual is more comfortable and/or needs more volume,” says Diana Guth, RRT.

For reimbursement of a bilevel unit in the USA by Medicare, the requirements are a diagnosis of a progressive neuromuscular disorder, absence of chronic obstructive pulmonary disease (COPD) or if present it does not significantly contribute to the individual’s respiratory limitations, and one of the following test results:

- FVC <50% of predicted,
- MIP <60 cm H₂O,
- PaCO₂ arterial blood gas >45 mm Hg,
- Nocturnal SpO₂ (oxygen saturation) <88% for five continuous minutes while asleep.

Obstructive sleep apnea (OSA).

Apnea is the cessation of airflow for more than 10 seconds. OSA occurs when tissues in the throat collapse, intermittently blocking airflow during sleep. Snoring is often a major indicator of OSA, but not always.

A sleep study (polysomnogram test or PSGT) is primarily used to determine and design treatment for individuals with OSA. A sleep study is not absolutely necessary for the diagnosis in people with neuromuscular disorders but it may be helpful when first introducing the bilevel treatment.

The main breathing problem is almost always hypoventilation, although people

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with neuromuscular disorders early on may also have undiagnosed OSA. Most sleep labs are not equipped to measure CO₂ levels, and therefore cannot diagnose hypoventilation.

The standard treatment for OSA is continuous positive airway pressure (CPAP) to help keep the airway open or a bilevel unit without a backup rate.

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