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Polysomnography for adults with neuro muscular disease commonly results in misdiagnosis and/or delayed diagnosis

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dults with forms of neuromuscular disease (NMD) commonly first show diaphragm and intercostal muscle weakness as sleep disordered breathing. In the US, sleep professionals are surprisingly unaware of the most important polysomnogram measurement for NMD patients: end-tidal carbon dioxide. Thus it often is unmeasured. Because respiratory muscle weakness can result in under ventilation and blood gas disturbances, lack of CO, measurement gives a partial snapshot of sleep breathing. Further, sleep labs often lack staff to assist with transfers and other needs throughout the study. Home use sleep study equipment is limited in availability. Polysomnogram results are frequently read by clinicians unfamiliar with NMD. Further, equipment does not interpret depth or effort of breathing and interprets the NMD shallow breathing as apnea. Too often this results in misdiagnosis. Generally, the NMD patient shows a restrictive pattern of breathing on pulmonary function test (PFT) and needs help with ventilation for deeper breathing and respiratory muscle rest. More often than not, the sleep study results in no equipment recommendation or an order for continuous positive airway pressure (CPAP). NMD patients with diaphragm weakness do not have the expiratory muscle strength to tolerate the positive pressure needed to support their inspiratory weakness. The NMD patient feels discomfort on CPAP and sees no improvement in sleep deprivation, arousals, and/or morning headaches. The individual must self-advocate to acquire the needed bi-level ventilation with two different positive pressures, one much lower for exhaling. This process often results in confusion, second opinion visits and months to years without needed intervention. Sleep professionals should collaborate with clinicians who specialize in NMD respiratory muscle weakness to offer thorough and accurate polysomnography analysis via home studies, allowing the patient access to their own environment and caregiver(s). Neglecting to do so means NMD patients' quality of life and longevity is negatively impacted.

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