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CASE REPORTS

The Role of Polysomnography in the Diagnosis of a Neuromuscular Disorder

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We describe the first diagnosed case of congenital myasthenia gravis suspected through polysomnogram. A 16-month-old girl, admitted with failure to thrive and apnea, underwent an all-night polysomnogram after common causes for sleep-disordered breathing were ruled out and treated. She was found to have hypoventilation related to respiratory muscle weakness. Further evaluation led to the final diagnosis of congenital myasthenia gravis.

CASE REPORT

This 16-month-old girl with a family history and phenotypic features of branchio-oto-renal syndrome (preauricular pits, hypertension, and hyperechogenic kidneys)[1] was admitted for failure to thrive felt to be secondary to gastroesophageal reflux disease. Physical examination was completely normal. Nocturnal obstructive apnea, without snoring but with severe bradycardia and oxygen desaturation, was noted. A magnetic resonance imaging study of the brain was normal. Ear, nose, and throat evaluation showed normal airways, and tracheostomy was performed to relieve upper-airway obstruction. Postoperatively, oxygen desaturations persisted, requiring continuous positive airway pressure and supplemental oxygen.

An overnight polysomnogram was obtained. Sleep architecture was normal, and no episode of obstructive or central apnea was recorded. Alveolar hypoventilation (mean SaO_2 87% and $TccO_2$ 51 mm Hg) with baseline tachypnea (80-95 breaths per minute) was recorded. Synchronous intermittent mandatory ventilation with a pressure support of 12 cm H_2O and 25 breaths per minute enabled her to wean to room air. A chest radiograph to further evaluate the etiology of the alveolar hypoventilation was normal. Complete neurologic examination showed normal strength and deep tendon reflexes, without focal deficits. An electromyogram performed at 2-Hz stimulation was normal but, when repeated at 10- and 50-Hz stimulation, revealed evidence for a presynaptic defect of neuromuscular transmission consistent with congenital myasthenia gravis; the patient was commenced on pyridostigmine.

DISCUSSION

This case is reported to highlight the role of polysomnography in the diagnosis of neuromuscular disorders related to polysomnographic findings of sleep-disordered breathing. This is the first reported case in the English literature of congenital myasthenia gravis initially suspected because of polysomnogram findings.

The child's failure to respond to tracheostomy ruled out upper-airway obstruction as the sole cause of sleep-disordered breathing. A normal magnetic resonance imaging scan of the brain ruled out any structural abnormalities (eg, Chiari II malformation). Polysomnography revealed adequate ventilation with tachypnea during wakefulness. During sleep, hypoventilation became pronounced, in spite of appropriately increased respiratory effort, notably through rapid eye movement (REM) sleep. Assisted ventilation was sufficiently therapeutic in this setting. Because of the otherwise normal findings on physical examination, these polysomnography results led to the suspicion that respiratory muscles may be weak, as the respiratory drive and chemoreceptors responses appeared to be intact.

In normal subjects, upper-airway resistance increases and chemosensitivity is reduced during sleep, resulting in a fall in ventilation. During REM sleep, ribcage and accessory breathing muscles are suppressed, with a further fall in ventilation.

It is known that sleep-disordered breathing and nocturnal desaturations are common and most severe during REM sleep.[2] Sleep-disordered breathing is common in myasthenia gravis associated with peripheral respiratory muscle weakness, particularly diaphragmatic weakness.[3,4] Significant hypoventilation and overt respiratory failure can occur abruptly,[5] as was seen in our patient.

A high prevalence of sleep-disordered breathing has been reported in patients with neuromuscular diseases, irrespective of the primary disorder. The most common form of sleep-disordered breathing in patients with respiratory muscle weakness is hypoventilation due to reduced tidal volume, particularly during phasic REM sleep. During REM sleep, suppression of the intercostal and accessory respiratory muscles (including the abdominal muscles), combined with inadequate diaphragmatic recruitment, leads to hypoventilation. The degree of muscle suppression, and consequent reduction in ventilation, is proportional to the density of eye movements and often meets the criteria for central hypop-

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nea during bursts of rapid eye movements.[2]

The overnight polysomnogram played the crucial role of eliciting the possibility of an underlying respiratory muscle weakness in the causation of sleep-disordered breathing in this patient. The electromyogram proved it to be a myasthenic syndrome. The patient, as in most cases of congenital myasthenic gravis, did not respond completely to pyridostigmine but is able to function fully with nocturnal ventilatory support alone on room air.

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