Journal Scan

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1. Am J Phys Med Rehabil. 2013 Oct;92(10):930-41.

A historical perspective on expiratory muscle aids and their impact on home care.

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Mechanically assisted coughing is the combination of mechanical insufflation-exsufflation to the airways in conjunction with an exsufflation-timed abdominal thrust. It has permitted in-home long-term survival of respirator-dependent users of continuous noninvasive intermittent positive pressure ventilatory support in the United States since shortly after it became available in 1952. By contrast, the first European patients to benefit from continuous noninvasive intermittent positive pressure ventilatory support were not described until institutionalized continuous noninvasive intermittent positive pressure ventilatory support users with Duchenne muscular dystrophy were described in Belgium in 2006, 2 yrs after mechanically assisted coughing was approved for sale by the European Union. Domiciliary management with continuous noninvasive intermittent positive pressure ventilatory support and mechanically assisted coughing has subsequently been described in Europe. This work describes the historical development of mechanically assisted coughing in the framework of aiding the respiratory muscles to prevent respiratory failure and avoid invasive airway intubation. Some current mechanical in-exsufflators include oscillation as a surrogate for physiologic mucociliary transport along with their assisted coughing function. Noninvasive management prolongs life while promoting cost containment and preserving quality-of-life for patients with respiratory muscle impairment.

2. PLoS One. 2013 Sep 18;8(9):e75324.

Nocturnal hypoxia in ALS is related to cognitive dysfunction and can occur as clusters of desaturations.

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BACKGROUND: Amyotrophic lateral sclerosis (ALS) is a neurodegenerative disease that leads to progressive weakness of the respiratory and limb muscles. Consequently, most patients with ALS exhibit progressive hypoventilation, which worsens during sleep. The aim of this study was to evaluate the relationship between nocturnal hypoxia and cognitive dysfunction and to assess the pattern of nocturnal hypoxia in patients with ALS.

METHOD: Twenty-five patients with definite or probable ALS underwent neuropsychologic testing, nocturnal pulse oximetry, and capnography. Patients were grouped according to the presence of nocturnal hypoxia (SpO2<95% for e”10% of the night) and their clinical characteristics and cognitive function were compared.

RESULTS: Compared to patients without nocturnal hypoxia, those with nocturnal hypoxia (n = 10, 40%) had poor memory retention (p = 0.039) and retrieval efficiency (p = 0.045). A cluster-of-desaturation pattern was identified in 7patients (70%) in the Hypoxia Group.

CONCLUSIONS: These results suggest that nocturnal hypoxia can be related to cognitive dysfunction in ALS. In addition, a considerable number of patients with ALS may be exposed to repeated episodes of deoxygenation-

reoxygenation (a cluster-of-desaturation pattern) during sleep, which could be associated with the generation of reactive oxygen species. Further studies are required to define the exact causal relationships between these phenomena, the exact manifestations of nocturnal cluster-of-desaturation patterns, and the effect of clusters of desaturation on ALS progression.

3. J Clin Sleep Med. 2013 Sep;9(9):879-84.

Diagnostic predictors of obesity-hypoventilation syndrome in patients suspected of having sleep disordered breathing.

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INTRODUCTION: Obesity-hypoventilation syndrome (OHS) is associated with significant morbidity and mortality and requires measurement of arterial pCO2 for diagnosis.

OBJECTIVE: To determine diagnostic predictors of OHS among obese patients with suspected obstructive sleep apnea/hypopnea syndrome (OSAHS).

METHODS: Retrospective analysis of data on 525 sleep clinic patients (mean age 51.4 ± 12.7 years; 65.7% males; mean BMI 34.5 ± 8.1). All patients had sleep studies, and arterialized capillary blood gases (CBG) were measured in obese subjects (BMI > 30 kg/m2).

RESULTS: Of 525 patients, 65.5% were obese, 37.2% were morbidly obese (BMI > 40 kg/m2); 52.3% had confirmed OSAHS. Hypercapnia (pCO2 > 6 kPa or 45 mm Hg) was present in 20.6% obese and 22.1% OSAHS patients. Analysis of OHS predictors showed significant correlations between pCO2 and BMI, FEV1, FVC, AHI, mean and minimum nocturnal SpO2, sleep time with SpO2 < 90%, pO2, and calculated HCO3 from the CBG. PO2 and HCO3 were independent predictors of OHS, explaining 27.7% of pCO2 variance ($p < 0.0001$). A calculated HCO3 cutoff > 27 mmol/L had 85.7% sensitivity and 89.5% specificity for diagnosis of OHS, with 68.1% positive and 95.9% negative predictive value.

CONCLUSION: We confirmed a high prevalence of OHS in obese OSAHS patients (22.1%) and high calculated HCO3 level (> 27 mmol/L) to be a sensitive and specific predictor for the diagnosis of OHS.

4. Eur Respir Rev. 2013 Sep 1;22(129):365-75.

Sleep disorders in COPD: the forgotten dimension.

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Sleep in chronic obstructive pulmonary disease (COPD) is commonly associated with oxygen desaturation, which may exceed the degree of desaturation during maximum exercise, both subjectively and objectively impairing sleep quality. The mechanisms of desaturation include hypoventilation and ventilation to perfusion mismatching. The consequences of this desaturation include cardiac arrhythmias, pulmonary hypertension and nocturnal death, especially during acute exacerbations. Coexistence of COPD and obstructive sleep apnoea (OSA), referred to as overlap syndrome, has been estimated to occur in 1% of the general adult population. Overlap patients have worse sleep-related hypoxaemia and hypercapnia than patients with COPD or OSA alone. OSA has a similar prevalence in COPD as in a general population of similar age, but oxygen desaturation during sleep is more pronounced when the two conditions coexist. Management of sleep-related problems in COPD should particularly focus on minimising sleep disturbance via measures to limit cough and dyspnoea; nocturnal oxygen therapy is not generally indicated for isolated nocturnal hypoxaemia. Treatment with continuous positive airway pressure alleviates hypoxaemia, reduces hospitalisation and pulmonary hypertension, and improves survival.

5. Eur Respir Rev. 2013 Sep 1;22(129):325-32.

Chronic hypoventilation and its management.

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While obstructive sleep apnoea syndrome dominates discussion of the prevalence of sleep disordered breathing, nocturnal hypoventilation remains extremely prevalent in those with chronic ventilatory disorders and in the natural history of these conditions pre-dates the development of daytime ventilatory failure. In this review the clinical management of chronic hypoventilation in neuromuscular disease will be considered and then compared with that in obesity hypoventilation syndrome. In simple terms these conditions illustrate the polar opposite ends of the spectrum, as in neuromuscular disease the reduced capacity of the respiratory system is unable to withstand a normal respiratory load, and in obesity hypoventilation syndrome the normal capacity of the respiratory system is unable to tolerate a substantially increased ventilatory load.

6. J Can Acad Child Adolesc Psychiatry. 2013 Aug;22(3):235-7.

Nocturnal Anxiety in a Youth with Rapid-onset Obesity, Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation (ROHHAD).

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OBJECTIVE: Behavioral and psychiatric disorders are common in youth with rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (ROHHAD). We outline a rational approach to psychiatric treatment of a patient with a complex medical condition.

METHODS: We report the course of symptoms in a teen with ROHHAD, the inpatient treatment, and review current evidence for use of psychopharmacologic agents in youth with sleep and anxiety disturbances.

RESULTS: A 14-year-old female began rapidly gaining weight as a preschooler, developed hormonal imbalance, and mixed sleep apnea. Consultation was requested after a month of ROHHAD exacerbation, with severe anxiety, insomnia, and auditory hallucinations. Olanzapine and citalopram were helpful in controlling the symptoms. Following discharge, the patient gained weight and olanzapine was discontinued. Lorazepam was started in coordination with pulmonary service. Relevant pharmacologic considerations included risk of respiratory suppression, history of paradoxical reaction to hypnotics, hepatic isoenzyme interactions and side effects of antipsychotics.

CONCLUSIONS: Core symptoms of ROHHAD may precipitate psychiatric disorders. A systematic evidence-based approach to psychopharmacology is necessary in the setting of psychiatric consultation.

7. Pediatrics. 2013 Sep;132(3):e788-92.

Hypocretin-1 deficiency in a girl with ROHHAD syndrome.

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Rapid-onset obesity with hypothalamic dysfunction, hypoventilation, and autonomic dysregulation (ROHHAD) is a rare and complex pediatric syndrome, essentially caused by dysfunction of 3 vital systems regulating endocrine, respiratory, and autonomic nervous system functioning. The clinical spectrum of ROHHAD is broad, but sleep/wake disorders have received relatively little attention so far, although the central hypothalamic dysfunction would make the occurrence of sleep symptoms likely. In this case report, we expand the phenotype of ROHHAD with a number of striking sleep symptoms that together can be classified as a secondary form of narcolepsy. We present a 7-year-old girl with

ROHHAD who displayed the classic features of narcolepsy with cataplexy: excessive daytime sleepiness with daytime naps, visual hallucinations, and partial cataplexy reflected in intermittent loss of facial muscle tone. Nocturnal polysomnography revealed sleep fragmentation and a sleep-onset REM period characteristic for narcolepsy. The diagnosis was confirmed by showing an absence of hypocretin-1 in the cerebrospinal fluid. We discuss potential pathophysiological implications as well as symptomatic treatment options.

8. Crit Care. 2013 Jul 26;17(4):R167.

Continuous positive airway pressure titration in infants with severe upper airway obstruction or bronchopulmonary dysplasia.

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INTRODUCTION: Noninvasive continuous positive airway pressure (CPAP) is recognized as an effective treatment for severe airway obstruction in young children. The aim of the present study was to compare a clinical setting with a physiological setting of noninvasive CPAP in infants with nocturnal alveolar hypoventilation due to severe upper airway obstruction (UAO) or bronchopulmonary dysplasia (BPD).

METHODS: The breathing pattern and respiratory muscle output of all consecutive infants due to start CPAP in our noninvasive ventilation unit were retrospectively analysed. CPAP set on clinical noninvasive parameters (clinical CPAP) was compared to CPAP set on the normalization or the maximal reduction of the oesophageal pressure (Poes) and transdiaphragmatic pressure (Pdi) swings (physiological CPAP). Expiratory gastric pressure (Pgas) swing was measured.

RESULTS: The data of 12 infants (mean age 10 ± 8 mo) with UAO (n = 7) or BPD (n = 5) were gathered. The mean clinical CPAP (8 ± 2 cmH, O) was associated with a significant decrease in Poes and Pdi swings. Indeed, Poes swing decreased from 31 ± 15 cmH, O during spontaneous breathing to 21 ± 10 cmH, O during CPAP ($P < 0.05$). The mean physiological CPAP level was 2 ± 2 cmH2, O higher than the mean clinical CPAP level and was associated with a significantly greater improvement in all indices of respiratory effort (Poes swing 11 ± 5 cm H, O; $P < 0.05$ compared to clinical CPAP). Expiratory abdominal activity was present during the clinical CPAP and decreased during physiological CPAP.

CONCLUSIONS: A physiological setting of noninvasive CPAP, based on the recording of Poes and Pgas, is superior to a clinical setting, based on clinical noninvasive parameters. Expiratory abdominal activity was present during spontaneous breathing and decreased in the physiological CPAP setting.

9. Respir Care. 2013 Aug;58(8):1367-76.

Behind a mask: tricks, pitfalls, and prejudices for noninvasive ventilation.

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It is difficult to exactly date the beginning of mechanical ventilation, but there are no doubts that noninvasive ventilation (NIV) was the first method of ventilatory support in clinical practice. The technique had a sudden increase in popularity, so that it is now considered, according to criteria of evidence-based medicine, the first-line treatment for an episode of acute respiratory failure in 4 pathologies (the Fabulous Four): COPD exacerbation, cardiogenic pulmonary edema, pulmonary infiltrates in immunocompromised patients, and in the weaning of extubated COPD patients. The so-called emerging applications are those for which the evidence has not achieved level A, mainly because the number or sample size of the published studies does not allow conclusive meta-analysis. These emerging applications are the post-surgical period, palliation of dyspnea, asthma attack, obesity hypoventilation syndrome, and to prevent extubation failure. Potentially "risky business" uses include for respiratory failure from pandemic diseases and ARDS, where probably the "secret" for success is early use. Healthcare is rich in evidence-based

innovations, yet even when such innovations are implemented successfully in one location, they often disseminate slowly, if at all, so their clinical use remains limited and heterogeneous. The low rate of NIV use in some hospitals relates to lack of knowledge about or experience with NIV, insufficient confidence in the technique, lack of NIV equipment, and inadequate funding. But NIV use has been increasing around the world, thanks partly to improved technologies. The skill and confidence of clinicians in NIV have improved with time and experience, but NIV is and should remain a team effort, rather than the property of a single local "champion," because, overall, NIV is beautiful!

10. Adv Exp Med Biol. 2013;788:167-73.

Efficacy of noninvasive mechanical ventilation in obese patients with chronic respiratory failure.

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Chronic respiratory failure (CRF) develops in a minority of obese patients. Noninvasive mechanical ventilation (NIMV) is a new optional treatment for such patients. The aim of this study was to evaluate the effectiveness of NIMV in obese patients with CRF. The material of the study consisted of 34 obese patients (body mass index 47.3 ± 7.9 kg/m(2)) with CRF (PaO2 = 6.40 ± 0.93 kPa and PaCO2 = 8.67 ± 2.13 kPa) who were hypoxemic despite an optimal therapy. Thirteen patients had an overlap syndrome (OS) - chronic obstructive pulmonary disease (COPD) coexisting with obstructive sleep apnea syndrome (OSAS) and 21 patients had obesity-hypoventilation syndrome (OHS). Ventilation parameters were determined during polysomnography. The efficacy of NIMV was evaluated on the fifth day and after 1 year's home treatment. We observed a significant increase in the mean blood oxygen saturation during sleep in all patients; the increase was greater in patients with OHS (92.6 ± 1.4 %) than in patients with OS (90.4 ± 1.8 %). There was a significant improvement of diurnal PaO2 and PaCO2 on the fifth day of NIMV (mean PaO2 increase 2.1 kPa and PaCO2 decrease 0.9 kPa) and also after 1 year of home NIMV (mean PaO2 increase 1.9 kPa and PaCO2 decrease 2.4 kPa). Only one patient stopped treatment because of lack of tolerance during the observation period (1-3 years). In conclusion, NIMV is an effective and well tolerated treatment option in obese patients with CRF resulting in a rapid relief of respiratory disorders during sleep and a gradual, long-term improvement of gas exchange during the day, particularly in patients with OHS.

11. Acta Neurochir (Wien). 2013 Aug;155(8):1417-24.

Awake craniotomies without any sedation: the awake-awake-awake technique.

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BACKGROUND: Temporary anaesthesia or analgosedation used for awake craniotomies carry substantial risks like hemodynamic instabilities, airway obstruction, hypoventilation, nausea and vomiting, agitation, and interference with test performances. We tested the actual need for sedatives and opioids in 50 patients undergoing awake craniotomy for brain tumour resection in eloquent or motoric brain areas when cranial nerve blocks, permanent presence of a contact person, and therapeutic communication are provided.

METHODS: Therapeutic communication was based on the assumption that patients in such an extreme medical situation enter a natural trance-like state with elevated suggestibility. The anaesthesiologist acted as a continuous guide, using a strong rapport, nonverbal communication, hypnotic suggestions, such as dissociation to a "safe place", and the reframing of disturbing noises, while simultaneously avoiding negative suggestions. Analgesics or sedatives were at hand according to the principle "as much as necessary, but not more than needed".

RESULTS: No sedation was necessary for any of the patients besides for the treatment of seizures. Only two-thirds of the patients requested remifentanil, with a mean dosage of 96 ìg before the end of tumour resection and a total of 156 ìg. Hemodynamic reactions indicative of

stress were mainly seen during nerve blockades and neurological testing. Postoperative vigilance tests showed equal or higher scores than preoperative tests.

CONCLUSIONS: The main challenges for patients undergoing awake craniotomies include anxiety and fears, terrifying noises and surroundings, immobility, loss of control, and the feeling of helplessness and being left alone. In such situations, psychological support might be more helpful than the pharmacological approach. With adequate therapeutic communication, patients do not require any sedation and no or only low-dose opioid treatment during awake craniotomies, leaving patients fully awake and competent during the entire surgical procedure without stress. This approach can be termed "awake-awake-awake-technique".

12. Can Respir J. 2013 May-Jun;20(3):165-70.

Diagnostic practices and disease surveillance in Canadian children with congenital central hypoventilation syndrome.

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OBJECTIVE: To assess the diagnostic and surveillance practices of Canadian pediatric subspecialists for children with congenital central hypoventilation syndrome (CCHS).

METHODS: The present analysis was a prospective cross-sectional study. A web-based survey was sent to 303 pediatric subspecialists in Canada: 85 pediatric respirologists, 77 pediatric neurologists and 141 neonatologists. The survey included 36 questions about the current diagnostic and surveillance management of pediatric CCHS. Differences in responses among respirologists, neurologists and neonatologists were evaluated for each question, where feasible, and responses were compared with the 2010 American Thoracic Society (ATS) Clinical Policy Statement for CCHS.

RESULTS: A total of 83 (27%) participants responded to the survey; the highest survey response rate (40%) was from respirologists. For the diagnosis of CCHS, 25% of respondents did not order genetic testing, either alone or with another test, to make a diagnosis of CCHS. The criteria and tests recommended by the ATS to make a diagnosis of CCHS - genetic testing, diagnosis of exclusion, polysomnogram and plus or minus a hypercapnic challenge - were ordered by 23 (43%) of the 54 respondents. Although polysomnograms were ordered for more than 90% of children with CCHS, only 37% of respirologists aimed for a carbon dioxide range of 35 mmHg to 40 mmHg during polysomnogram titrations.

CONCLUSIONS: The results demonstrate variability in the diagnostic and surveillance practices of pediatric subspecialists in children with CCHS across Canada. The present study provides an initial needs assessment and demonstrated that there are significant deviations in practice from the 2010 ATS guidelines.

13. Aviat Space Environ Med. 2013 Jun;84(6):551-9.

Lung volumes, pulmonary ventilation, and hypoxia following rapid decompression to 60,000 ft (18,288 m).

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INTRODUCTION: Rapid decompressions (RD) to 60,000 ft (18,288 m) were undertaken by six subjects to provide evidence of satisfactory performance of a contemporary, partial pressure assembly life support system for the purposes of flight clearance.

METHODS: A total of 12 3-s RDs were conducted with subjects breathing 56% oxygen (balance nitrogen) at the base (simulated cabin) altitude of 22,500 ft (6858 m), switching to 100% oxygen under 72 mmHg (9.6 kPa) of positive pressure at the final (simulated aircraft) altitude. Respiratory pressures, flows, and gas compositions were monitored continuously throughout.

RESULTS: All RDs were completed safely, but one subject experienced significant hypoxia during the minute at final altitude, associated with severe hemoglobin desaturation to a low of 53%. Accurate data on subjects' lung volumes were obtained and individual responses post-RD were reviewed in relation to patterns of pulmonary ventilation.

The occurrence of severe hypoxia is explained by hypoventilation in conjunction with unusually large lung volumes (total lung capacity 10.18 L).

CONCLUSIONS: Subjects' lung volumes and patterns of pulmonary ventilation are critical, but idiosyncratic, determinants of alveolar oxygenation and severity of hypoxia following RD to 60,000 ft (18,288 m). At such extreme altitudes even vaporization of water condensate in the oxygen mask may compromise oxygen delivery. An altitude ceiling of 60,000 ft (18,288 m) is the likely threshold for reliable protection using partial pressure assemblies and aircrew should be instructed to take two deep 'clearing' breaths immediately following RD at such extreme pressure breathing altitudes.

14. Sleep. 2013 Jun 1;36(6):933-40.

Increased sympathetic and decreased parasympathetic cardiac tone in patients with sleep related alveolar hypoventilation.

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OBJECTIVE: To assess autonomic function by heart rate variability (HRV) during sleep in patients with sleep related alveolar hypoventilation (SRAH) and to compare it with that of patients with obstructive sleep apnea (OSA) and control patients.

DESIGN: Cross-sectional study.

SETTING: Sleep Unit, University Hospital of University of Navarra.

PATIENTS: Fifteen idiopathic and obesity related-SRAH patients were studied. For each patient with SRAH, a patient with OSA, matched in age, sex, body mass index (BMI), minimal oxygen saturation (SatO2), and mean SatO2 was selected. Control patients were also matched in age, sex, and BMI with patients with OSA and those with SRAH, and in apnea/hypopnea index (AHI) with patients with SRAH.

INTERVENTIONS: N/A.

MEASUREMENTS AND RESULTS: Time- and frequency-domain HRV measures (R-R, standard deviation of normal-to-normal RR interval [SDNN], very low frequency [VLF], low frequency [LF], high frequency [HF], LF/HF ratio) were calculated across all sleep stages as well as during wakefulness just before and after sleep during a 1-night polysomnography. In patients with SRAH and OSA, LF was increased during rapid eye movement (REM) when compared with control patients, whereas HF was decreased during REM and N1-N2 sleep stages. The LF/HF ratio was equally increased in patients with SRAH and OSA during REM and N1-N2. Correlation analysis showed that LF and HF values during REM sleep were correlated with minimal SatO2 and mean SatO2.

CONCLUSIONS: Patients with SRAH exhibited an abnormal cardiac tone during sleep. This fact appears to be related to the severity of nocturnal oxygen desaturation. Moreover, there were no differences between OSA and SRAH, supporting the hypothesis that autonomic changes in OSA are primarily related to a reduced nocturnal oxygen saturation, rather than a consequence of other factors such as nocturnal respiratory events.

15. Panminerva Med. 2013 Jun;55(2):191-5.

Obstructive sleep apnea/hypopnea syndrome.

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Obstructive sleep apnea/hypopnea syndrome (OSAHS) is characterized by recurrent episodes of partial or complete upper airway collapse during sleep that is highlighted by a reduction in, or complete cessation of, airflow despite documented on going inspiratory efforts. Due to the lack of adequate alveolar ventilation that results from the upper airway narrowing, oxygen saturation may drop and partial pressure of CO2 may occasionally increase. The events are mostly terminated by arousals. Clinical consequences are excessive daytime sleepiness related to the sleep disruption. Minimal diagnostic criteria have been defined for OSAHS. Patients should have excessive daytime sleepiness that can not be better explained by other factors, or experience two or more of the following symptoms, again that are not better

explained by other factors: choking or gasping during sleep; recurrent awakenings from sleep; un-refreshing sleep; daytime fatigue; and impaired concentration. All patients should have more than five obstructed breathing events per hour during sleep. An obstructive apnea or hypopnoea can be defined as an event that lasts for e 10 s and is characterized by an absence or a decrease from baseline in the amplitude of a valid measure of breathing during sleep that either reaches >50% with an oxygen desaturation of 3% or an arousal (alternatively a 30% reduction with 4% desaturation). The American Academy of Sleep Medicine (AASM) recommends these definitions. The Task Force of the AASM also states that there are common pathogenic mechanisms for obstructive apnea syndrome, central apnea syndrome, sleep hypoventilation syndrome and Cheyne-Stokes breathing. It was more preferable to discuss each of these separately; although, they could be placed under the common denominator of "sleep-disordered breathing syndrome". The definition of OSAHS using two components, daytime symptoms and breathing pattern disturbances during sleep, may suggest that there is a tight correlation between the two. However, unfortunately this is not the case. The breathing pattern abnormalities, mostly described by an Apnea/Hypopnoea Index (AHI), only weakly correlate with quantified measures of sleepiness, such as the Epworth Sleepiness Scale (ESS). This probably means that interindividual sensitivity, with some individuals coping better with sleep fragmentation than others, does compromise the relationship between the AHI and daytime sleepiness scores. In addition, epidemiological studies show a broad range of sleepiness in the general population. Obviously, epidemiological studies investigating the prevalence of OSAHS are all biased by the lack of a uniform definition. The prevalence of an AHI of >5 events · h-1 in a general population (without taking into account symptoms of sleepiness) has previously been estimated to be 24% in a male population. When symptoms of sleepiness were also taken into account, the prevalence decreased to 4% in males and 2% in females.

16 Int J Artif Organs. 2013 Jun 25;36(6):434-8.

Reversal of pulmonary hypertension after diaphragm pacing in an adult patient with congenital central hypoventilation syndrome.

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INTRODUCTION: Patients with the congenital central hypoventilation syndrome (CCHS) suffer from life-threatening hypoventilation when asleep, making them dependent on mechanical ventilation (MV) at night or during naps. State-of-art respiratory management consists of intermittent positive-pressure ventilation via a tracheotomy or mask. In some patients hypoventilation is permanent, in which case ventilatory support must be extended to the waking hours. Diaphragm pacing can prove useful in such situations.)

METHODS AND RESULTS: This report describes the case of a 26-year-old woman with CCHS in whom failure to achieve adequate MV led to life-threatening pulmonary hypertension (PH), with a systolic pulmonary artery pressure (PAP) of 80 mmHg and right ventricular hypertrophy, despite optimization of all possible measures and despite extensive therapeutic education efforts. Diaphragm pacing using laparoscopically implanted intradiaphragmatic phrenic nerve stimulation electrodes corrected alveolar hypoventilation and lastingly reversed PH (systolic PAP below 40 mmHg after 2 months, sustained after 2 years). Diaphragm pacing induced shoulder pain, however, involving the chronic use of analgesics. The pacing had to be stopped for tolerance reasons after two years, leading to PH worsening and the need for diurnal MV.)

CONCLUSIONS: Diaphragm pacing appears likely effective to restore alveolar ventilation and reverse PH in adult CCHS patients.