The Children's Institute
Pittsburgh, Pennsylvania

Clinical Presentation of Obesity Hypoventilation and Right Heart Failure in Prader-Willi Syndrome

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INTRODUCTION

Obesity Hypoventilation (OHS)

Virtually every kind of sleep disordered breathing has been described in persons with PWS (Vela Bueno 1984; Hertz, 1993; Hertz; 1995; Schluter, 1997; Klift, 1994; Richards, 1993; Vgontzas, 1995) including the non obese child or adult. Hypoventilation is characterized by "constant or slowly diminishing oxyhemoglobin desaturation without the cyclic, episodic or repetitive changes in oxygen saturation associated with apneas and hypopneas or the arousal that terminates these abnormal breathing events." This sustained hypoxia is the typical pattern seen on sleep pulse oximetry of obese patients with PWS and is relatively uncommon in non- PWS obese persons (Von Boxem 1999; Koenig 2001). OHS persons ultimately develop reduced lung volume (Bedell, 1958; Holley 1967).

Stage 1 Nocturnal hypoventilation and hypoxia

Hypoxia first appears during REM phases of sleep (Hertz, 1993) and, as the obesity worsens, can be demonstrated throughout the night sometimes with profound and prolonged hypoxic episodes. Decreased stamina may be present but not be noted by family members.

Stage 2 Early distress manifest by edema

Edema is a useful early clinical sign of obesity-hypoventilation in PWS but this sign is *frequently missed*. The reason for this appears to be the visual subtlety of edema in the obese child or adult. One useful way to describe this type of edema is that the "fat gets hard" as the turgor (firmness) of dependent tissues increases. *Pitting is usually absent*. In the presence of *any* recognizable edema, nocturnal oxygen desaturations are usually quite extensive. (Figure 1)

Stage 3 Daytime hypoxemia and dyspnea

Ambulatory patients with daytime oxygen desaturations have edema (nonpitting) usually to or above the level of the thighs and hips. Oxygen saturations when the patient is awake and sitting quietly may be well below 85%, dropping still lower with activity. Cardiomegaly on chest X-ray sometimes still appears "mild". In Stage 3 and 4 patients will typically increase their resting respiratory rate but this tachypnea is not accompanied by a visible increase in respiratory effort. At rest the tidal volume is small (Pack, 1998) Resting breath sounds are often barely audible with the stethoscope. With activity, however, increased respiratory effort is more evident and often reported by family members as shortness of breath.

Stage 4 Respiratory Failure

Respiratory failure with CO_2 retention is a life-threatening condition which may be acute or chronic. Obese persons with PWS may continue to survive in a compensated state in stage 3 for years without evidence of respiratory failure *if* their obesity is stable and *if* they remain active. Clinical experience clearly indicates that inactivity and overuse of oxygen therapy both cause worsening daytime and nighttime hypoventilation with worsening CO_2 retention. Overzealous use of O_2 frequently precipitates critical illness, intubation, tracheostomy, all of which can and should be avoided by judicious use of O_2, (max 1 liter/minute or 28%) , immediate ambulation and other physical rehabilitation .

Clinical Presentation

The all too familiar clinical picture of shortness of breath, worsening daytime sleepiness and leg swelling seen in morbidly obese persons with Prader-Willi syndrome is obesity hypoventilation leading to respiratory failure with or without right heart failure. The onset may be rapid or slow but is always insidious, that is, initially unrecognized. The clinical presentation of OHS leading to *cor pulmonale* in PWS has been delineated from a large number of patients with PWS arriving at the Children's Institute with various degrees of severity of the condition. The sequence of events leading to morbidity, disability and death from obesity hypoventilation is fairly stereotyped and can be observed in reverse during rehabilitation. The sequence can develop over a period of months in the face of rapid weight gain and severe nocturnal hypoxia or slowly in patients whose weight is stable but in the obese range for many years.

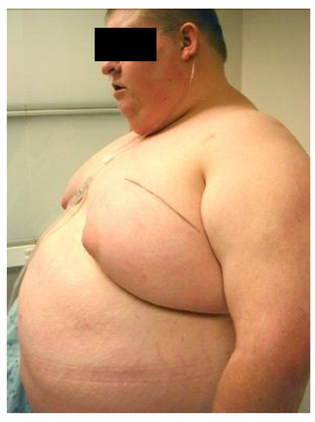
Stage 1 Asymptomatic nocturnal hypoxia

Stage 2 Fluid retention; decreased endurance

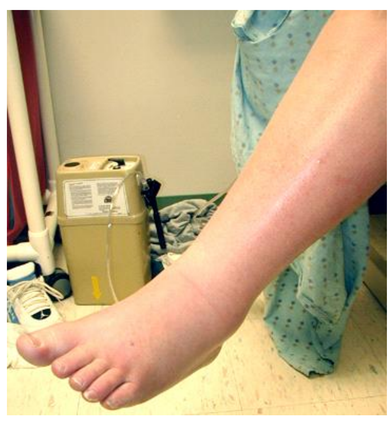
Stage 3 Daytime hypoventilation and hypoxia

Stage 4 Respiratory failure

Adipose tissue of the chest wall increases the work of breathing; cardiomegaly is a late finding.



Patient in Stage 3: Edema appears to follow adipose tissue and may be palpated to the the level of the thighs, waist or chest even when leg edema is visually unimpressive; this is non pitting edema and is best appreciated by palpation of tissue.



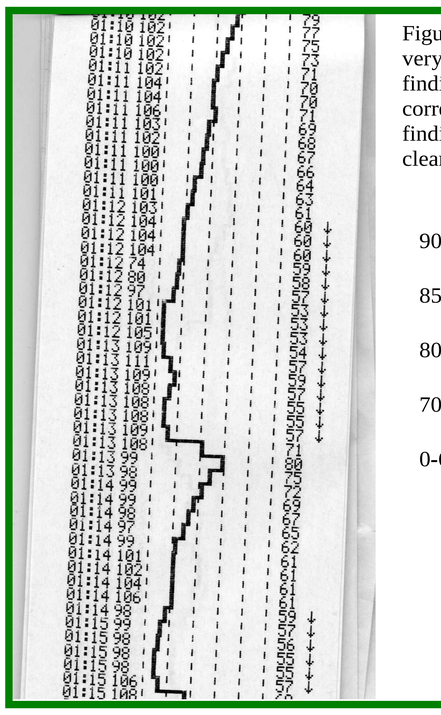
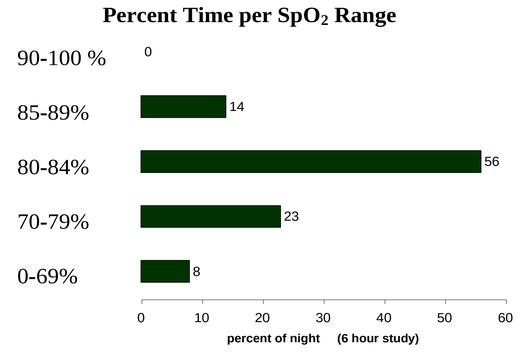
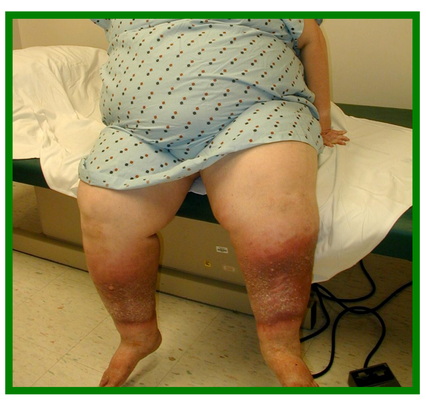
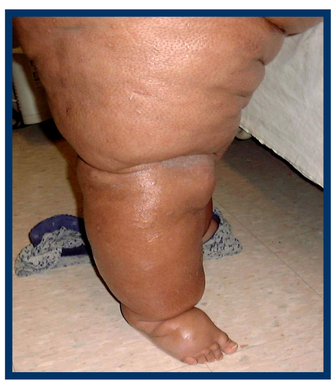


Figure 1 Pulse oximetry studies during sleep are very useful in identifying hypoxemia, the most common finding in obese children and adults with PWS. BMI correlates very imperfectly with the severity of these findings but weight loss and moderate daytime exercise clearly improve nighttime ventilation and hypoxia.



Marked hypoxia lasting most of the night was present in this obese teenager who had mild shortness of breath with exertion and a subtle increase in tissue turgor of her lower body.





Severe chronic changes of the legs with longstanding untreated Obesity Hypoventilation
Early treatment is needed to prevent irreversible damage to veins and lymphatics

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Management and Rehabilitation of Obesity Hypoventilation and Right Heart Failure in Prader-Willi Syndrome

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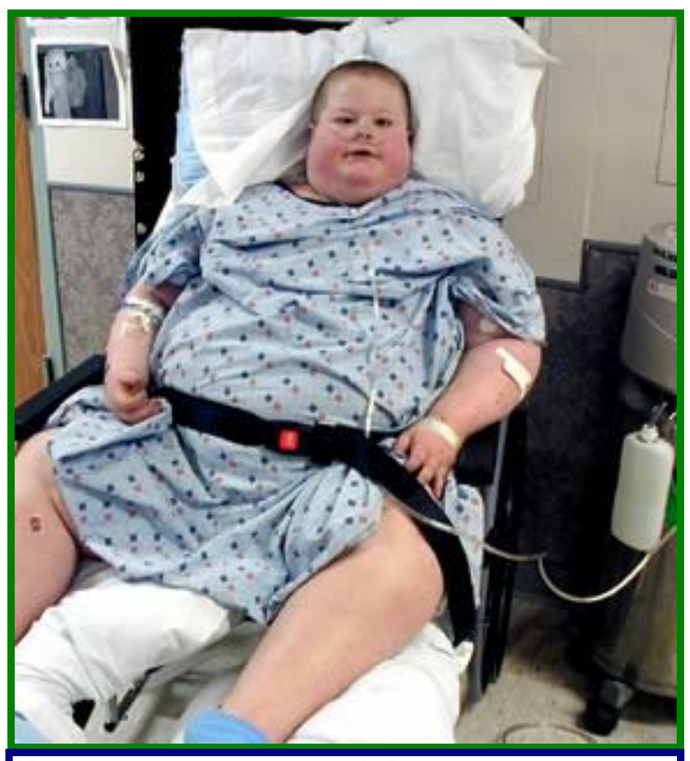
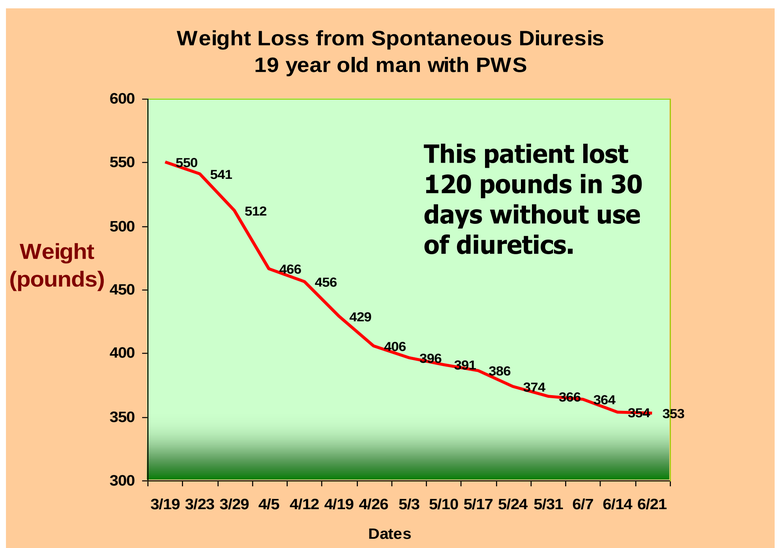
Activity, diet, and spontaneous diuresis

Two primary modalities are effective in reversing the cardiopulmonary deterioration of obesity hypoventilation. These are calorie restriction and ambulation. Rehabilitation to a higher level of physical activity is essential for recovery. Even the most seriously ill patients must begin this process immediately upon hospitalization. **All other medical measures are supportive therapy and will not reverse the condition.**

Hypoxia is not a contraindication for exercise; this patient took his O_2 tank on a hike in the hills with his therapists.



Nine year old boy in stage 4: Proper positioning is vital ; this patient had severe CO_2 retention. His ventilation was restricted by obesity and hard tissue (edema) to the level of his upper chest.



During rehabilitation:

- Daily intake of 600-800 kcal. will provide adequate nutritional maintenance in the obese, ill patient. Higher caloric intake is clearly not needed even for wound healing.
- Patients with PWS are NEVER fed in bed.
- There are gradually increasing demands for physical activity beginning, if necessary, with walking a few steps to a chair for meals.
- Nurses and therapists work together; consistent behavioral rewards and consequences are implemented by nursing as well as therapy staff. This includes delaying meals until modest therapy goals are achieved.
- Physical and occupational therapists are successful only when they have adequate support from nursing staff and time to wait out the inevitable behaviors of PWS such as whining, crying, delaying, manipulation and frank refusals by reluctant patients.
- Communication with the patient follows the behavioral guidelines to avoid nonproductive efforts.

Avoid overuse of oxygen

Oxygen desaturations may be profound but they are undoubtedly longstanding and patients have adjusted.

Patients are prone to CO2 retention.

Use 1 liter nasal or mask **maximum** (28%).

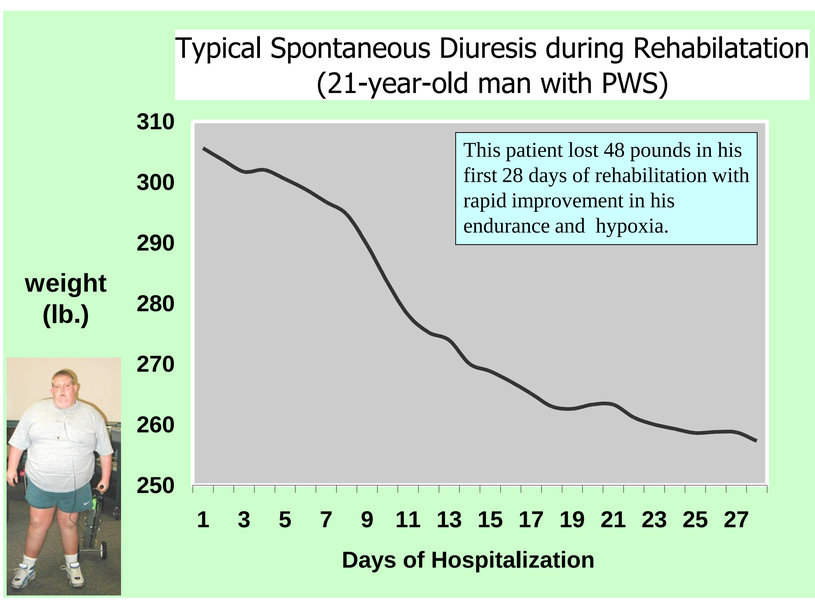
Avoid diuretics

Initiate activity, not diuretics (i.e. treat the hypoventilation not the edema).

Taper diuretics as tolerated.

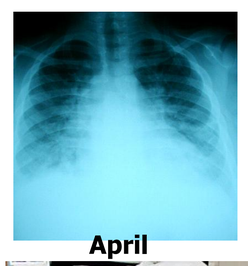
Edema is compensatory; excessive volume depletion decreases a cardiac function.

Normal adult PWS creatinine is typically .5-.8; higher values are abnormal and may indicate overuse of diuretic.

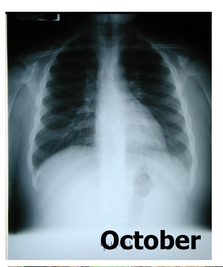


Signs of Recovery

Spontaneous diuresis of edema fluid and improving oxygen saturation are the hallmarks of recovery. The effects of a low calorie diet and increased activity in producing a diuresis are at times dramatic. There is a known natriuretic effect produced by low calorie intake (Katz, 1968; Kreitzman, 1988) which together with exercise (Nichols, 1990) and improved ventilation produces a physiologic response that marks the patients' return to improved cardiopulmonary function. Use of diuretics appears to delay this diuresis rather than assist it even if azotemia is avoided.

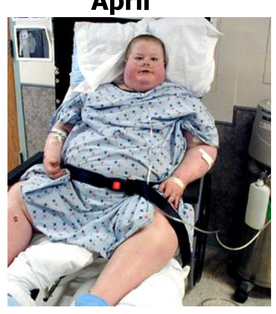


April



October

Full recovery of 9 year old who presented in Stage 4, respiratory failure



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