

# Foreign body aspiration in a boy with Prader-Willi Syndrome

Jacob S S, Jacob J J, Paul T V

## ABSTRACT

**A five-year-old boy presented with progressive weight gain with effort intolerance and nocturnal symptoms suggesting obstructive sleep apnoea. A clinical diagnosis of Prader-Willi Syndrome was made. As the initial radiography and computed tomography suggested a foreign body, bronchoscopy was done under general anaesthesia and impacted peanuts were removed from the left main bronchus. His symptoms resolved instantly and the patient was asymptomatic at six months follow-up. This report highlights the need to consider foreign body aspiration as a cause for dyspnoea in children with Prader-Willi Syndrome. The report also focuses on the need to adopt strategies that prevent foreign body aspiration and choking in patients with Prader-Willi Syndrome.**

**Keywords: foreign body aspiration, obstructive sleep apnoea, Prader-Willi Syndrome**

*Singapore Med J 2008; 49(1): e12-e14*

## INTRODUCTION

Prader-Willi syndrome (PWS) is the most common syndromic cause of childhood obesity. In addition to the hyperphagia seen by one year of age, patients with PWS suffer from hypotonia and poor motor coordination. This combination of symptoms predisposes these children to choking and aspiration of food. In addition, patients with PWS have breathing disorders related to obstructive sleep apnoea (OSA) and central alveolar hypoventilation. We report a five-year-old boy with symptoms suggestive of OSA, and who was later found to have foreign body aspiration. Removal of the foreign body resulted in complete resolution of the respiratory symptoms. This case report highlights the need to consider foreign body aspiration as a cause for dyspnoea in children with PWS, and the need to institute measures to minimise the risk of aspiration and choking in children with PWS.

## CASE REPORT

A five-year-old boy presented with a history of progressive weight gain, breathlessness on exertion and nocturnal symptoms suggestive of OSA, which had recently worsened over a period of 1–2 months. Born to non-consanguineous parents, he weighed 1.7 kg at birth, had a poor cry and required oxygen for respiratory distress. He required nasogastric feeding in hospital for the first two weeks after birth. His developmental milestones were all delayed. Although the subject had feeding difficulties initially, after one year of age, this was replaced by increasing weight and a voracious appetite. The patient had suffered from constipation and lethargy at one month of age. He was diagnosed to have primary hypothyroidism, and he has since been on Levo-thyroxine replacement therapy.

On examination, he weighed 29 kg and his height was 107 cm. Head circumference was 49.5 cm. He was obese with almond-shaped eyes, double chin, inverted nipples, clinodactyly of the left fifth finger, enlarged adenoids and hypotonia. He had no goitre and appeared clinically euthyroid upon Levo-thyroxine replacement. Genitalia examination was unremarkable. On respiratory examination, there were decreased breath sounds on the left side with a scattered polyphonic wheeze. He had mild mental retardation with an intelligence quotient of 62. Ophthalmological examination revealed myopic astigmatism. His hearing was normal.

A clinical diagnosis of PWS with OSA was made. An initial sleep study showed significant desaturations at night with multiple episodes of awakening. An ENT appointment confirmed that the adenoids were not causing upper airway obstruction. The initial chest radiograph of the patient showed hyperlucency with loss of lung volume on the left side (Fig. 1). In view of this, a computed tomography (CT) of the chest was done. The CT revealed an abrupt narrowing of the left main bronchus and confirmed the loss of lung volume on the left side (Fig. 2). As this suggested a foreign body, bronchoscopic examination was done and peanuts were found in the left main bronchus. The breathlessness resolved immediately following removal of the impacted peanuts. On a follow-up visit six months later, the boy had no symptoms of dyspnoea.

Department of  
Endocrinology,  
Christian Medical  
College,  
Vellore 632004,  
Tamil Nadu,  
India

Jacob SS, MBBS  
Research Officer

Jacob JJ, MD, DNB  
Tutor

Paul TV, MD, DNB  
Lecturer

Correspondence to:  
Dr Jubbin J Jacob  
Tel: (91) 46 1228 2528  
Fax: (91) 46 1520 3570  
Email: jubbin@india.  
com



**Fig. 1** Anteroposterior chest radiograph taken at presentation shows hyperlucency and decreased attenuation of the left lung, leading to the suspicion of an inhaled foreign body.



**Fig. 2** Axial CT image of the thorax shows narrowing of the left main bronchus (arrow).

## DISCUSSION

PWS is characterised by an insatiable hunger starting from childhood, mental retardation, hypogonadism and growth deficiency; hypotonia, feeding problems and failure to thrive are the predominant features in the neonatal period.<sup>(1)</sup> The clinical diagnosis of PWS is based on the criteria proposed by Holm et al in 1993 and is summarised in Table I.<sup>(1)</sup> The availability of genetic testing (deletion 15q11-13 on high resolution [ $> 650$  bands] or other cytogenetic molecular abnormality of the Prader-Willi chromosome region, including maternal isomy) for PWS has standardised the diagnosis, leading to a more specific diagnosis much earlier than that would have been possible with simply the clinical criteria for reference. Revised criteria published in 2001 proposed a lower clinical threshold to prompt DNA testing and a more definitive diagnosis.<sup>(2)</sup>

Children with PWS suffer from a variety of breathing abnormalities. These include OSA and alveolar hypoventilation related to sleep. Obesity in PWS leads to increased fat deposition in the soft palate, uvula, neck and pharynx. These deposits contribute to the narrowing of upper airway. This, along with pharyngeal muscle weakness during sleep, results in OSA.<sup>(3)</sup> In addition, there is abnormal central ventilatory response to hypoxia in obese and non-obese patients with PWS and decreased responsiveness of the peripheral chemoreceptors.<sup>(4)</sup> The fact that they do not arouse normally to the hypoxia puts them at higher risk for OSA-associated morbidity and sudden death. All children with PWS and breathing abnormalities associated with sleep disorders should undergo sleep study and therapy for the underlying problem. Appropriate therapy may include weight control, adenotonsillectomy and nocturnal ventilation.

**Table I. Diagnostic criteria for Prader-Willi Syndrome. <sup>(1)</sup>**

Major criteria	Minor criteria
1. Characteristic facial features (includes almond-shaped eyes, down-turned mouth, narrow bifrontal diameter, strabismus, thin upper lip).	1. Decreased foetal movement and infantile lethargy.
2. Developmental delay.	2. Esotropia, myopia.
3. Feeding problems/failure to thrive during infancy.	3. Hypopigmentation.
4. Hypogonadism (may include cryptorchidism, hypoplastic scrotum, small testes, hypoplastic labia minora and clitoris, and delayed puberty).	4. Narrow hands with straight ulnar border.
5. Infantile hypotonia.	5. Short stature (compared with family members).
6. Rapid weight gain between one and six years of age.	6. Skin picking.
	7. Sleep disturbance/sleep apnoea.
	8. Small hands and feet.
	9. Speech articulation defects.
	10. Thick, viscous saliva.
	11. Typical behavioural problems.

Score one point for each major criterion and 0.5 point for each minor criterion. A diagnosis of PWS should be suspected in children younger than three years of age with a score of at least 5; and in children aged three years and older with a score of at least 8, with 4 points from the major criteria.

Fatalities among children with PWS following growth hormone (GH) therapy prompted concerns about aggravating OSA with GH therapy. However, prospective short-term evaluation of OSA over six months of GH therapy documented improvement in OSA in two-thirds of the study group and worsening in the remainder.<sup>(5)</sup>

The other potential problem in patients with PWS is choking and death related to choking. In a recent survey that examined the contribution of choking leading to mortality in PWS, 34% of deceased patients were reported to have a history of choking, with choking reported as the cause of death in 8%. Potential causes of increased choking in PWS include poor oral/motor coordination, poor gag reflex, hypotonia, hyperphagia, decreased mastication, and voracious feeding habits.<sup>(6)</sup> The same reasons that predispose patients with PWS to choking would also increase the risk of foreign body aspiration. In patients with preexisting sleep-related breathing disorders, foreign body aspiration may easily be overlooked. In this report, the patient was presented for the first time with a ten-week history of sleep-related dyspnoea and respiratory distress. The initial chest radiograph finding led to the suspicion of a foreign body aspiration; subsequent removal of the impacted peanuts led to a complete reversal of his respiratory symptoms.

Behavioural therapy targeted at dietary control and weight reduction may prevent early deaths in patients with PWS. However, this is almost impossible to achieve in the majority of patients. Additional measures that could prevent

mortality and morbidity related to aspiration and choking causes include:<sup>(6)</sup>

1. Education of care providers on the Heimlich manoeuvre.
2. Supervised meals.
3. Better food preparation.
4. Avoidance of high-risk choking foods, like nuts and lozenges.

In conclusion, this case highlights the need to consider foreign body aspiration in patients with PWS and respiratory distress. The risk of aspiration and choking is related to the motor in-coordination, hypotonia and hyperphagia. Preventive strategies aimed at minimising choking and aspiration may improve mortality and morbidity rates in patients with PWS.

#### REFERENCES

1. Holm VA, Cassidy SB, Butler MG, et al. Prader-Willi syndrome: consensus diagnostic criteria. *Pediatrics* 1993; 91:398-402.
2. Gunay-Aygun M, Schwartz S, Heeger S, O'Riordan MA, Cassidy SB. The changing purpose of Prader-Willi syndrome clinical diagnostic criteria and proposed revised criteria. *Pediatrics* 2001; 108:E92.
3. Deane S, Thomson A. Obesity and the pulmonologist. *Arch Dis Child* 2006; 91:188-91.
4. Arens R, Gozal D, Omlin KJ, et al. Hypoxic and hypercapnic ventilatory responses in Prader-Willi syndrome. *J Appl Physiol* 1994; 77:2231-6.
5. Miller J, Silverstein J, Shuster J, Driscoll DJ, Wagner M. Short-term effects of growth hormone on sleep abnormalities in Prader-Willi syndrome. *J Clin Endocrinol Metab* 2006; 91:413-7.
6. Stevenson DA, Heinemann J, Angulo M, et al. Deaths due to choking in Prader-Willi syndrome. *Am J Med Genet A* 2007; 143:484-7.