

A unique solution to obesity hypoventilation in a patient with Noonan's syndrome

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Abstract

We report the difficulties of managing a patient with obesity hypoventilation syndrome (OHS) with the problem of a 'floppy' neck during sleep kinking his trachea and causing upper airways obstruction⁸. We also describe a unique but simple solution to this patient's problem by using a neck collar, as normally used for a neck injury.

Case Report

A 46 year old male with Noonan's syndrome was admitted to A&E with a presumed accidental overdose of lithium and sodium valproate. On admission his observations were stable (T-36.5, BP- 147/92, HR-78, sats-91% on room air). He had a BMI of 33 and was also known to have epilepsy and a bipolar disorder. Two days post admission he went into respiratory failure due to possible aspiration pneumonia and was started on non-invasive ventilation (NIV) via full face mask (pH-7.27, HCO₃⁻ 41.3, PaO₂- 6.3, PCO₂- 12.0). The chest x-ray showed right basal diffuse shadowing. Despite the NIV, the arterial blood gas (ABG) quickly deteriorated (pH-7.04, oxygen saturation - 88% on 10L of O₂, PaO₂ -8.5, PCO₂ and HCO₃⁻ were non recordable) and was transferred to intensive therapy unit (ITU) for intubation. After three days he had a tracheotomy done to facilitate weaning off the ventilator and transferred to the high dependency unit (HDU). During the stay in HDU attempts to decannulate were unsuccessful and the patient's condition deteriorated due to a new chest infection. He was transferred back to ITU. After a stay of 14 days in ITU, the patient was transferred to HDU and finally to the respiratory unit (RU). While in the RU several attempts to decannulate him failed as the patient kept on desaturating. Tracheomalacia was questioned

by the ENT surgeons due to prolonged intubation and the patient was transferred to University Hospital of North Tees (UHNT) for a bronchoscopy with a view to stenting. The bronchoscopy showed only inflammation so UHNT returned the patient to our care.

Due to the patient's high BMI and short webbed neck (characteristic of Noonan's syndrome), obesity hypoventilation syndrome (OHS) or simple obstructive sleep apnoea (OSA) with a normal trachea was thought to be the cause of respiratory failure. The patient had his tracheostomy tube changed to a fenestrated one and a sleep study was attempted by blocking the "trachy" tube^{7,8}. This had to be terminated as the patient quickly desaturated during the procedure. The patient was placed on a *Resmed Elisee* ventilator for full ventilation in the RU. After a prolonged stay of a further four months the patient was weaned off NIV during the day, but still required ventilation during the night. It was then noticed by the nursing staff that when the patient went to sleep his head flopped over and this seemed to obstruct his breathing. A simple foam neck collar was applied and this improved his breathing when asleep to such an extent that NIV could be discontinued. A proper neck collar with a custom opening for the tracheostomy was acquired. With the new neck collar being used at night it was possible to de-cannulate the patient. Unfortunately before he could be discharged he once again developed pneumonia and had to be re-cannulated. He came through this event quickly and once again we managed to wean him off the ventilator and de-cannulate him with the help of his special neck collar. Two of the authors, (BR and JN), then trained the nursing home staff to manage the trachy tube until the patient was well enough to attempt

a decannulation. The patient continued to sleep with his neck collar. Approximately four weeks later final de-cannulation was achieved. The patient is starting to loose weight now and is a very happy person again.

Noonan's syndrome is a relatively common congenital syndrome, equally affecting males and females, although it is referred to as male version of Turners syndrome. Characteristic features are congenital heart malformation, short stature with typical webbed neck, learning problems, impaired blood clotting¹⁰ and characteristic facial features.

Obesity hypoventilation syndrome (OHS) is defined as obesity (BMI >30)¹ and hypoventilation (PCO₂ >6) in the absence of a coexisting pulmonary, chest wall or neuromuscular pathologies. This was previously known as the Pickwickian syndrome⁹. The decline in FVC, TLC and RV are linear whereas the decline in expiratory reserve volume (ERV) and FRC is exponential (ERV= FRC-RV) with increasing BMI^{2,3}. Oxygen consumption is also increased in obesity and respiratory muscle function is impaired in obesity which is thought to be due to a myopathy. The ventilatory response to induced hypercapnia is reduced in patients with obesity as compared to lean controls⁴ and this suggests that obesity impairs the drive to correct hypercapnia. Sleep events also contribute to reduced central drive. A study showed that 87% of the OHS had obstructive sleep apnoea (OSA) and 6% had hypothyroidism⁵. Although not common, severe OSA, especially in this group, may be fatal¹¹.

Hospitalized patients with OHS can initially be managed with NIV and usually converted to simple continuous positive airway pressure (CPAP) therapy quite easily before being discharged home. Medical weight loss has been associated with some correction of the hypoventilation⁶, but sustaining the weight loss

can be a difficult problem. In selected patients bariatric surgery is nowadays a preferred treatment option.

The idea of the neck collar is a novel solution to what initially had seemed to be an insoluble problem. Although collapsing upper airways of varying degrees are pathognomonic of OSA we can find no other report of a similar kind.

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