Melnick-Needles Syndrome with Obstructive Sleep Apnea Successfully Treated with Nasal Continuous Positive Airway Pressure Ventilation

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Melnick-Needles syndrome is a very rare X-linked syndrome, first reported by Melnick and Needles in 1966, and usually diagnosed on the basis of characteristic facial and radiographic findings. Females with the disease survive, but males are lethally affected. We describe a case of Melnick-Needles syndrome complicated with severe obstructive sleep apnea syndrome (OSAS) and restrictive ventilatory impairment that was successfully treated with nasal continuous positive airway pressure (CPAP).

Case Report

A 31-year-old female presented at the plastic surgery clinic complaining of longstanding snoring, witnessed episodes of sleep apnea, excessive daytime somnolence, and dyspnea on exertion. She had a prominent forehead, bilateral exophthalmos, fullness of the cheeks and retrognathia. A spot diagnosis of Melnick-Needles syndrome was made based on these characteristic features.

Three-dimensional computed tomography of the head revealed the characteristic feature and deviation of the left nasal septum (Figure 1). Chest radiography showed bilateral ribbon-like ribs and medial flaring of the clavicles (Figure 2). Pulmonary function tests revealed severe restrictive ventilatory impairment with decreased total lung capacity, forced expiratory volume in 1 second (FEV₁) and forced vital capacity (FVC), but FEV₁/FVC ratio and diffusion capacity were normal.

Polysomnography study led to the diagnosis of severe OSAS with a respiratory disturbance index (RDI) of 76.8/hour. Titration of nasal CPAP (nCPAP) was tried on the second night. The RDI decreased to less than 5/hour after the pressure was decreased to 5/hour.
Discussion

Melnick-Needles syndrome is characterized by craniofacial and skeletal abnormalities. Typically, the mandibular rami are scalloped and eroded bilaterally, and the coronoid process is hypoplastic. Patients with Melnick-Needles syndrome often have a small thoracic cage. Restrictive ventilatory impairments may be apparent as a result, and pulmonary hypertension can develop.

Melnick-Needles syndrome was diagnosed in our patient based on pathognomic facial and skeletal features (Figures 1 & 2). A diagnosis of severe OSAS associated with Melnick-Needles syndrome was then made. Melnick-Needles syndrome associated with OSAS was previously reported by Curran et al in 1993. Tracheostomy was used to treat OSAS in their report. The craniofacial abnormalities of Melnick-Needles syndrome, most of which were present in this case, can all be expected to contribute to upper airway obstruction. As the patient ages, muscle tone decreases, upper airway obstruction during sleep gradually increases, and the small thoracic cage with its restrictive ventilatory effects exacerbates the hypoxemia and increases the severity of OSAS. If obstructive sleep apnea and restrictive ventilatory impairment increased to above 7 cmH₂O, nCPAP (ResMed, Sydney, Australia) was prescribed, but was not tolerated initially. Nasal submucosal resection, uvulopharyngopalatoplasty (UPPP), tongue and soft palate reduction were then performed to improve airway caliber. Despite these treatments, snoring and sleep apnea persisted. Mandibular advancement was suggested, but the patient declined after considering the likely social impact of the change in facial appearance.

An attempt to gradually reintroduce nCPAP was made after further education and a better fitting mask was provided. The patient subsequently managed to successfully use her nCPAP at home, with the pressure set at 7 cmH₂O. Symptoms of sleep apnea, snoring, daytime somnolence and exertional dyspnea improved, including Epworth sleepiness scale score from 16 to 10, and minimal O₂ saturation during sleep from 89.4% to 94.6%. Quality of life, evaluated by Short-Form 36, also improved in five domains (physical function: 85 to 95; role-physical: 75 to 100; bodily pain: 72 to 100; general health: 35 to 40; role-emotional: 33 to 100).

Figure 1. Three-dimensional computed tomography scan of the head shows prominent forehead with decreased anteroposterior diameter, bilateral exophthalmos, mild hypertelorism, bilateral fullness of the cheeks, micrognathia and dental malocclusion.

Figure 2. Chest X-ray shows decreased lung volume and bony abnormalities, including ribbon-like ribs and medially flared clavicles.
are not corrected, repeated hypoxemia and hypventilation can lead to pulmonary hypertension, cor pulmonale and right heart failure. Pulmonary hypertension has been previously reported in patients with Melnick-Needles syndrome. nCPAP is the treatment of choice if the restrictive ventilatory impairment and obstructive sleep apnea cannot be ameliorated with UPPP or mandibular advancement. Unfortunately, patients with craniofacial abnormalities, such as Melnick-Needles syndrome, have difficulty in getting a good mask fit, which affects compliance.

UPPP and nasal submucosal resection can help to widen the oropharyngeal introitus, but skeletal abnormalities will remain that continue to compromise the upper airway during sleep. Ideally, mandibular advancement is performed to improve the caliber of the upper airway, but the restrictive ventilatory impairment will persist.

Despite the dearth of previous reports regarding OSAS in patients with Melnick-Needles syndrome, OSAS should be suspected in all such patients, and nCPAP should be prescribed if the diagnosis is confirmed, in order to improve survival and quality of life.

In conclusion, Melnick-Needles syndrome is rare, with characteristic facial and skeletal abnormalities. In these patients, OSAS and restrictive ventilatory impairment can occur and lead to further pulmonary complications. Surgery may only partially correct upper airway obstruction and fail to prevent further pulmonary complications. nCPAP is ideally suited to treat the unique respiratory problems affecting patients with Melnick-Needles syndrome. Given the potential for nCPAP to improve outcomes and quality of life in such patients, treating specialists should be aware of the need for, and have access to nCPAP when managing these patients.

References