Restrictive Chest Bellows Disease and Frontometaphyseal Dysplasia*


We describe a patient with frontometaphyseal dysplasia (FMD), restrictive chest bellows disease, hypercapnic respiratory failure, and cor pulmonale. Treatment with intermittent supplemental oxygen, nocturnal nasal volume ventilation, and posture modification was successful in partial resolution of chronic hypoventilation and excessive daytime somnolence.  

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FRONTOMETAPHYSEAL DYSPLASIA

Frontometaphyseal dysplasia (FMD) is a congenital syndrome characterized by cranial hyperostosis, abnormal tubulation of cylindrical bone, and additional skeletal and extraskeletal abnormalities.1,4 Restrictive lung disease and pulmonary hypertension were observed only in the first patient described with this syndrome,4 a 19-year-old man. Other congenital thoracic anomalies associated with FMD include subglottic narrowing, pectus carinatum, scoliosis, and a predisposition to upper respiratory tract infection. Treatment of a patient with FMD, restrictive chest bellows disease, and hypercapnic respiratory failure, with nocturnal nasal volume ventilation, posture modification, and intermittent supplemental oxygen, is described. This is the first report, to our knowledge, of management of this problem with nasal ventilation.

CASE REPORT

A 30-year-old man with FMD presented with six months of progressive dyspnea on exertion, three-pillow orthopnea, excessive daytime somnolence, and a two-week history of pedal edema. Pertinent medical history included surgical repair of pectus excavatum, at the age of 10 years, at which time the patient had complained of dyspnea and fatigability. The patient drank six beers per day and smoked one pack of cigarettes per day. Physical examination revealed a man in mild respiratory distress, with intermittent cyanosis relieved by rest. Prominent supraorbital ridges, poor dentition, micrognathia, and long extremities, typical of his known FMD, were seen (Fig 1). Thoracic lordosis and mild scoliosis (Fig 2), respiratory accessory muscle hypertrophy and use, and a loud split P2 were noted.

An arterial blood gas determination, while breathing room air, revealed hypoxemia and respiratory acidemia (pH, 7.37; PaO\(_2\), 64 mm Hg; and PaCO\(_2\), 55 mm Hg). Moderately severe restrictive chest bellows disease and mild obstructive airways disease were demonstrated by pulmonary function testing (FEV\(_1\), 0.77 L or 19 percent predicted; FVC, 0.96 L or 20 percent predicted, VC, 0.99 L or 20 percent predicted; FRC, 20 L or 81 percent predicted; MVV, 28 L/min or 17 percent predicted; and TLC, 3.55 L or 54 percent predicted). Fluoroscopy of the diaphragms revealed normal bilateral diaphragmatic movement. Oximetry, conducted at 0.5 mph for 4 min, without an incline, and without supplemental oxygen, revealed marked O\(_2\) desaturation, from 84 percent to 78 percent. A subsequent test, conducted with O\(_2\), 2 L/min per nasal cannula, under otherwise equivalent conditions, revealed baseline and exercise oxyhemoglobin saturations of 95 percent and 94 percent, respectively. Daytime supplemental oxygen was prescribed.

The patient underwent two consecutive nocturnal polysomnograms. Polysomnography was conducted using a multichannel

**FIGURE 1.** Prominent supraorbital ridges and micrognathia, typical of frontometaphyseal dysplasia. L or 20 percent predicted; RV, 2.56 L or 150 percent predicted; FRC, 2.60 L or 81 percent predicted; MVV, 28 L/min or 17 percent predicted; and TLC, 3.55 L or 54 percent predicted. Fluoroscopy of the diaphragms revealed normal bilateral diaphragmatic movement. Oxytmery, conducted at 0.5 mph for 4 min, without an incline, and without supplemental oxygen, revealed marked O\(_2\) desaturation, from 84 percent to 78 percent. A subsequent test, conducted with O\(_2\), 2 L/min per nasal cannula, under otherwise equivalent conditions, revealed baseline and exercise oxyhemoglobin saturations of 95 percent and 94 percent, respectively. Daytime supplemental oxygen was prescribed.

**FIGURE 2.** Mild scoliosis and lordosis in this patient with frontometaphyseal dysplasia (FMD). and less striking than those typically associated with hypercarbic respiratory failure in kyphoscoliosis (roentgenogram taken before therapeutic intervention).

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**Key points:**
- A 30-year-old man with FMD presented with progressive dyspnea on exertion, orthopnea, and daytime somnolence.
- Physical examination revealed supraorbital ridges, micrognathia, and long extremities.
- Arterial blood gas showed hypoxemia and respiratory acidemia.
- Pulmonary function testing demonstrated restrictive chest bellows disease and mild obstructive airways disease.
- Nocturnal polysomnography revealed marked desaturation and improved with supplemental oxygen.
The hypercarbia of nocturnal hypoventilation, and the mechanical negative pressure tracheostomy revealed tamed 25 and 25 grams, and 25 mm Hg; (Puritan Bennett) and 25 mm Hg. Oxyhemoglobin desati,rations were observed above this time oxyhemoglobin saturation was above 95 percent. In the second polysomnogram, sleep-disordered breathing was abolished with nasal volume ventilation (Puritan Bennett Companon 2800 Portable volume ventilator in SIMV mode, frequency 24/min, tidal volume 1,700 ml, with ventilation delivered through a ventilator [Puritan Bennett] Adam circuit nasal delivery system with small nasal pillows) and by restricting the patient's nocturnal posture to the nasal position. Oxyhemoglobin saturation was maintained above 90 percent during sleep, without supplemental oxygen. Nocturnal nasal volume ventilation and nonstop sleep were prescribed.

Six weeks later, the patient was found to be acyanotic, more alert, and without pedal edema, but he still noted mild daytime hyper-somnolence. Fifteen weeks after initiation of therapy, an awake room air arterial blood gas determination demonstrated near complete resolution of chronic hypercapnia (pH, 7.43; PaCO₂, 67 mm Hg; and PCO₂, 46 mm Hg). Oxyhemoglobin, conducted at 4 mph for 2 min, on a 10 percent incline, without supplemental oxygen, revealed baseline and exercise oxyhemoglobin saturations of 94 percent and 89 percent, respectively.

**DISCUSSION**

Therapies for hypercapnic respiratory failure include tracheostomy with volume-cycled ventilation, nocturnal nasal ventilation, mouth intermittent positive pressure ventilation, face mask ventilation, diaphragm pacing, and negative pressure ventilation. These techniques offer their respective advantages and disadvantages. Tracheostomy and mechanical ventilation offer reliable airway control, but they require surgery and poor cosmesis. Mouth intermittent positive pressure ventilation is traditionally reserved for patients who are awake and is used in conjunction with a nasal system for nocturnal use. Complications of face mask ventilation include skin necrosis and aspiration, and some investigators routinely use nasogastric suction, making this a less attractive option. Diaphragm pacing requires a surgical procedure and a tracheostomy. The chief disadvantages of negative pressure devices include bulk, discomfort, confinement, and the induction of upper airway obstruction, possibly through asynchrony with patient respirations.

Sleep-disordered breathing is a recognized sequela of diseases of the chest bellows. Nocturnal nasal ventilation has been used successfully in small groups of patients with restrictive chest wall disease, particularly kyphoscoliosis. The principal complications of this therapy include nasal dryness or congestion, eye irritation from air leaks, and gastric distention. It is otherwise free of many of the disadvantages of other means of ventilation.

To our knowledge, this is the first report of daytime hypercarbia and hypersomnolence secondary to FMD, and its treatment with nocturnal nasal ventilation. Central apneas have been described in association with various chest wall diseases and the presence of central sleep apnea in this patient may suggest an alteration in ventilatory chemosensitivity. However, near complete normalization of daytime hypercarbia with nocturnal assisted ventilation argues against an isolated defect in central respiratory control accounting for the hypercarbia. An acquired and reversible defect in ventilatory chemosensitivity cannot be excluded. Extensive muscle wasting, calf wasting, and wasting of the muscles of the arms and legs, especially the thenar and intraosseous muscles of the hands, are all myopathies reported in association with FMD. There were no physical signs of a generalized myopathy in this patient and diaphragmatic motion was normal. Therefore, an isolated respiratory myopathy is unlikely. We propose that hypersomnolence and hypercarbia occurred as a consequence of sleep-disordered breathing and restrictive chest wall disease. Considering that the thoracic abnormalities were less striking (Fig 2) than those typically associated with hypercarbic respiratory failure in kyphoscoliosis, pulmonary physicians should be aware of the potential association between FMD and sleep-disordered breathing.

**REFERENCES**

13. Leger P, Jennequin J, Gerard M, Robert D. Home positive pressure ventilation via nasal mask for patients with neuromuscular weakness or restrictive lung or chest-wall disease. Respir Care 1989; 34:73-7