Tracheomalacia in an Adult With Respiratory Failure and Morquio Syndrome

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Patients with Morquio syndrome can develop respiratory failure secondary to reduced chest wall compliance and airway collapse from irregularly shaped vocal cords and trachea. We report the case of a patient with Morquio syndrome whose clinical course was complicated by tracheomalacia. An obese 29-year-old female with Morquio syndrome presented with severe wheezing and tachycardia. One month prior to admission, she underwent elective spinal stabilization surgery, which resulted in fixed head flexion. The surgery was complicated by paraplegia and the need for mechanical ventilation via tracheostomy. Initial bronchoscopy revealed severe tracheomalacia, and the tracheostomy tube was changed to one with an adjustable flange. On 3 occasions over the next 20 days she had labored breathing with dramatically decreased V_T. Each time, bronchoscopy revealed almost complete occlusion of the distal end of the tracheostomy tube. Ventilation became much easier when the tracheostomy tube was advanced past the obstruction. After one month, she became febrile, severely hypoxemic, and her family decided to withdraw care. In patients with Morquio syndrome, close attention must be given to the patient's abnormal airways and malformed chest cage. Mechanical ventilation may be difficult because of upper-airway obstruction or low compliance imposed by the restrictive chest wall. Complete tracheal collapse can occur in these patients, especially with fixed head flexion. *Key words: Morquio syndrome, tracheomalacia, tracheostomy, respiratory failure*. [Respir Care 2007;52(3):278–282. © 2007 Daedalus Enterprises]

Introduction

Morquio syndrome is a rare autosomal recessive disease in a group of disorders called mucopolysaccharidosis.[1,2] It was first described by Morquio in 1929 and is diagnosed in one of every 300,000–500,000 births. Patients with Morquio syndrome have a deficiency in either the enzyme galactosamine-6-sulfate sulfatase in type A, or the enzyme β-galactosidase in type B. This leads to the accumulation of keratin sulfate in the lysosomes, causing malformation of bones, muscles, and connective tissues. Patients with Morquio syndrome often have coarse facial features, macrocephaly, bell-shaped chest, short trunk, kyphoscoliosis, cervical spine instability, and hypermobile joints. Though intelligence is preserved, spinal-cord compression due to vertebral instability leads to progressive weakness and paralysis. Prophylactic cervical fusion is usually recommended. Prenatal diagnosis is made by testing the enzyme level in amniotic fluid. Post-birth diagnosis is via urine chemistry and skin fibroblast culture. Treatment is supportive and symptom-based. Experimental enzyme replacement therapy has been attempted but thus far is unsuccessful. The severity of disease in Morquio syndrome depends on the amount of residual enzyme activity. Though life expectancy is approximately 20 years in the severe forms, mildly affected individuals may survive into the seventh decade. Death usually occurs due to cardiorespiratory failure.

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Respiratory complications may arise in patients with Morquio syndrome due to skeletal restriction, airway compromise, and neurologic compromise. Kyphoscoliosis and chest-wall deformity result in restrictive lung disease. Upper-airway compromise and laryngeal narrowing is common. Cartilage malformation and deposition of mucopolysaccharides result in an abnormally shaped trachea and/or vocal cords, and airway collapse resulting in tracheal and lower-airway obstruction. High spinal cord compression due to atlantoaxial instability and odontoid dysplasia can result in depressed respiration or sudden respiratory arrest.

Because Morquio syndrome is rare, clinicians caring for adult patients will see these patients infrequently. However, because of the very specific airway and respiratory problems encountered by these patients, it is important that these problems are either recognized or anticipated early, and that a plan of action is devised. We report the case of a patient with Morquio syndrome whose clinical course was complicated by tracheomalacia.

Case Summary

A 29-year-old female presented to the emergency department with tachycardia and severe wheezing refractory to bronchodilators. She had a prior medical history of Morquio syndrome and had features common in Morquio syndrome, including macrocephaly, coarsening of facial features, and short stature (Fig. 1).

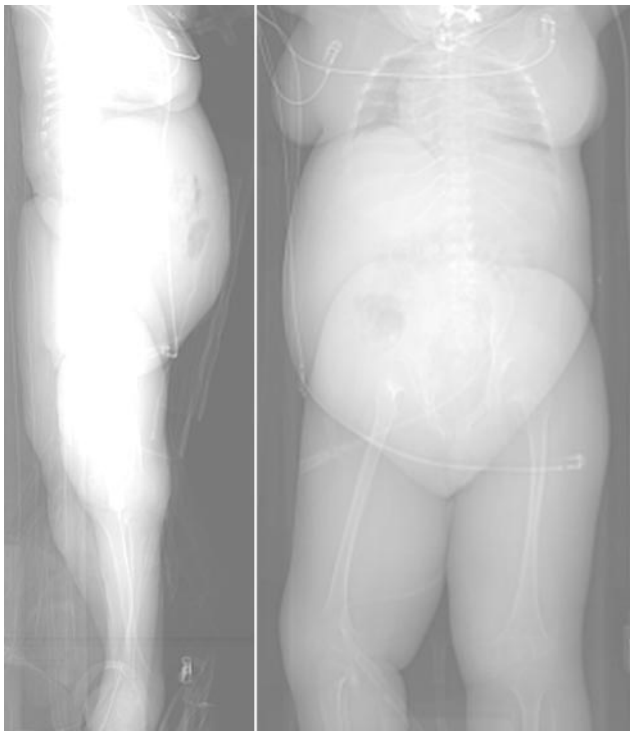

Fig. 1. Scout radiographs illustrating the body habitus of a patient with Morquio syndrome. She has a short neck that is fixed in flexion, causing the chin to hang over the tracheostomy tube, as well as a small chest cavity (due to chest-wall abnormalities), short stature, and obesity.

One and a half months prior to this admission, she underwent elective spinal stabilization surgery for foramen magnum compression. This procedure included foramen magnum decompression, C1 laminectomy, and spinal fusion from occiput to C4, resulting in fixed head flexion. The surgery was complicated by spinal-cord infarction, paraplegia (T4 level), and respiratory failure due to weakness and recurrent pneumonias, which required mechanical ventilation and tracheostomy. Initial tracheostomy placement was difficult because of a short anterior neck that was fixed in flexion, and substernal cricoid cartilage. A cuff leak persisted despite multiple trials with different tracheostomy tubes, and therefore a 6.0-mm Bivona tracheostomy tube with a tight-to-shaft cuff was placed. The tight-to-shaft cuff is a low-volume, high-pressure cuff when inflated during positive-pressure ventilation, but when deflated allows greater air flow around the tube during spontaneous breathing. She was then transferred to the rehabilitation center where she had been undergoing tracheotomy mask trials lasting up to an hour. Because of the relatively short time the high-pressure-cuff tube was in place, we do not believe that clinically important tracheal wall injury occurred with the tight-to-shaft cuff.

She presented to the emergency department receiving manual ventilation and was placed on pressure-controlled ventilation, with settings of pressure control (PC) 24 cm H_2O, positive end-expiratory pressure (PEEP) of 5 cm H_2O, respiratory rate 26 breaths/min, and fraction of inspired oxygen (F_{IO_2}) 0.7, which yielded tidal volume (V_T) of 150–250 mL. Her initial arterial blood gas results revealed a severe acute-on-chronic respiratory acidosis (pH 7.24, P_{aCO_2} 71 mm Hg, P_{aO_2} 93 mm Hg), and she required heavy sedation to allow adequate ventilation. Following stabilization, she was transferred to the medical intensive care unit for treatment of respiratory failure secondary to pneumonia and acute respiratory distress syndrome.

The following day, in the medical intensive care unit, ventilation became very difficult; her V_T decreased to 70–90 mL on PC 30 cm H_2O, PEEP 10 cm H_2O, respiratory rate 28 breaths/min, and F_{IO_2} 0.7. There was no change in her level of sedation and she was not dyssychronous with the ventilator. Bronchoscopy revealed severe tracheomalacia and obstruction of the distal tip of the tracheostomy tube with the membranous posterior trachea. The acute increase in airways resistance was thought to be due to tracheal collapse in combination with a tracheostomy tube that was too short to bypass the collapsed segment of trachea. The tracheostomy tube was changed to a 6.0-mm Bivona with an adjustable flange that allowed positioning the tube beyond the collapsed tracheal segment. The flexible Bivona tube also relieved any component of trache-

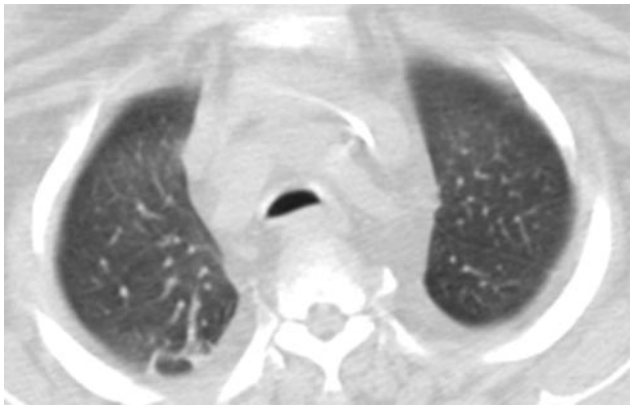

Fig. 2. Thoracic computed tomogram showing an irregularly shaped trachea and tracheal collapse. Although there appears to be an ample lumen for gas flow at the level shown in this image, there may be considerably more airway narrowing during exhalation. A similar tracheal shape, with a relatively elongated posterior membrane, was reported by Walker et al.[6]

ostomy tube malposition due to tracheal abnormalities and the patient's fixed neck flexion. With the tracheostomy tube secured at 10 cm from the flange to the distal tip, V_T returned to 215–350 mL on decreased ventilator settings (PC 15 cm H_2O, PEEP 10 cm H_2O, respiratory rate 26 breaths/min, and F_{IO_2} 0.6). Findings consistent with tracheal collapse were present on computed tomogram (Fig. 2). The next day, a persistent cuff leak was noted, so the tracheostomy tube was up-sized to a 7.0-mm Bivona, with the adjustable flange secured 9 cm from the distal tip, with adequate cuff seal and ventilation.

In the subsequent days, her sedation was decreased and she was transferred to the respiratory acute care unit for further ventilator weaning on the 9th hospital day. She was switched from pressure-controlled ventilation to pressure-support ventilation, and she tolerated spontaneous breathing trials of increasing length. Her spontaneous V_T was 150–300 mL and her pulse-oximetry-measured oxygen saturation was 94–96% on F_{IO_2} 0.5. However, she almost always required increased ventilatory support at night. It was also commonly noted that she would develop respiratory distress during spontaneous breathing trials that was relieved with a low level of ventilatory support (eg, 5 cm H_2O pressure support and 5 cm H_2O PEEP). It was difficult to keep the adjustable-flange tracheostomy tube in the proper place because of her severely fixed neck flexion.

After 2 weeks in the respiratory care unit, she developed extreme respiratory distress during a spontaneous breathing trial, with tachypnea, tachycardia, bilateral wheezes, and a V_T decrease to 50–100 mL. Even after resuming ventilatory support of PC 20 cm H_2O, PEEP 15 cm H_2O, respiratory rate 12 breaths/min, and F_{IO_2} 0.5, arterial blood gas analysis showed acute-on-chronic respiratory acidosis

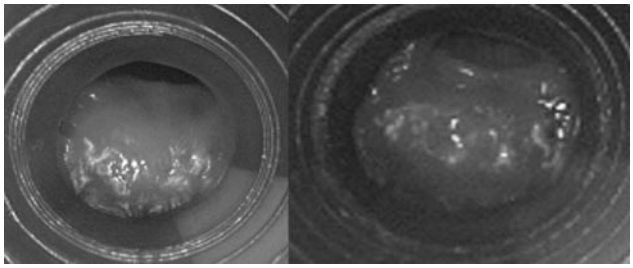

Fig. 3. Bronchoscopy (left: during inhalation; right: during exhalation) shows posterior tracheal collapse and the tracheal wall obstructing the tracheostomy tube. The tube was advanced past the obstruction, which was probably due to both tracheomalacia and airway distortion associated with Morquio syndrome.

(pH 7.24, P_{aCO_2} 94 mm Hg, P_{aO_2} 137 mm Hg). Her inspiratory airways resistance was 40 cm H_2O/L/s using the end-inspiratory-pause method. Bronchoscopy showed tracheostomy tube malposition resulting in almost complete occlusion of her tracheostomy tube by her posterior tracheal membrane (Fig. 3). Likely the high airways resistance was due not only to tracheomalacia but also to airway distortion associated with Morquio syndrome causing the tracheal wall to obstruct the tracheostomy tube. The tracheostomy tube, which had slipped out to 8.75 cm, was advanced back to 9 cm, resulting in increased tracheal patency. After repositioning the tube, V_T increased to 500–600 mL and airways resistance decreased to 18 cm H_2O/L/s. During the subsequent bronchoscopy it was noted that airway patency was improved with the addition of PEEP, with an optimum PEEP of 15 cm H_2O. Since positive airway pressure was required to stent her airways open, we instituted a strategy of pressure-support ventilation during the day and volume-controlled ventilation at night. Her set PEEP ranged from 8 cm H_2O to 15 cm H_2O during this period.

Ten days later, the patient again developed respiratory distress and was difficult to ventilate. Bronchoscopy again showed obstruction of the distal tracheostomy tube. The tracheostomy tube was advanced to 10 cm and PEEP was increased from 8 cm H_2O to 15 cm H_2O. This need to reposition the tube illustrated that tube position was critical for maintaining the airway patent and that the adjustable-flange tracheostomy tube is prone to slip. Over the next several days, she developed a fever to 40.5°C and required increasing ventilatory support and sedation. She had another episode of respiratory distress, and bronchoscopy again showed occlusion of the tracheostomy tube, and the tube was advanced to 10.75 cm, which placed the tip about 1 cm above the carina. She was transferred back to the medical intensive care unit for sepsis and acute respiratory distress syndrome. Despite multiple ventilator changes, and sedatives and paralytics to allow for adequate ventilation, her arterial blood gas values showed a persistent severe respiratory acidosis and hypoxemia. Care was

withdrawn per family request; she died one month after her initial presentation to the emergency room.

Discussion

This case illustrates respiratory care issues encountered in patients with Morquio syndrome. Specifically, these include chronic respiratory problems due to chest wall deformity and airway problems due to neck flexion and tracheomalacia. As this is a rare disease, clinicians will treat these patients infrequently. However, individuals with Morquio syndrome are increasingly surviving to adulthood, increasing the likelihood of presenting to the hospital in acute respiratory failure. Early recognition of these patients' respiratory and airway problems is imperative.

Tracheal anomalies have previously been reported in patients with Morquio syndrome.[3–5] Pritzker et al[4] reported that patients with Morquio syndrome could suffer from tracheal collapse during head flexion. Shinhar et al[5] reported 3 cases of difficult airway management in patients with Morquio syndrome, and Walker et al[6] reported 2 cases with similar problems. The tracheal anomalies in our patient made airway management very challenging. Though tracheomalacia usually refers to airway collapse due to weakening of the supporting cartilage, it is also characterized by widening of the posterior membrane (see Fig. 2) and reduced anterior-posterior diameter of the trachea. This was the main cause of obstruction found on bronchoscopy in our patient (Fig. 3). Collapse is worsened during forced exhalation.

Shinhar et al[5] described the use of custom-made tracheostomy tubes to stent the trachea in patients with Morquio syndrome. With our patient we used a tracheostomy tube with an adjustable flange (Fig. 4). Unlike a tracheostomy tube with a fixed flange, the adjustable-flange tube allows adjustment of the distance the tube extends into the trachea.[7] In this case we adjusted the tube position under bronchoscopic guidance. A disadvantage of this tube is the tendency of the tube shaft to slide in the flange, which can be devastating in patients in whom the tracheostomy tube length is crucial. This becomes increasingly problematic over time, and, for this reason, this tracheostomy tube is not recommended for prolonged use. Our patient's short, severely flexed neck caused her chin to bear directly onto the proximal tracheostomy tube, necessitating frequent readjustment of the tube position.

In patients with Morquio syndrome, tracheostomy management can be difficult in terms of tube placement, tube type, and tube size. Tracheomalacia can create marked difficulties in management, and this can extend to the carina and beyond. In addition, the major airways can be compromised by abnormal deposits that may require a stent. Early recognition of this problem and a coordinated plan to manage this is imperative. Because of thoracic

Fig. 4. The Bivona adjustable-flange tracheostomy tube, similar to that used in the patient reported here.

cage abnormalities, patients with Morquio syndrome eventually develop respiratory failure. Elective tracheostomy, management of airway issues, and involvement of clinicians who can provide home mechanical ventilation are imperative. This often necessitates referral to a tertiary-care referral center.

Positive airway pressure can effectively prevent airway collapse in patients with tracheomalacia.[8–13] This was the case in our patient. We noted that she could often breathe without distress on low levels of positive-pressure ventilation (eg, 5 cm H_2O pressure support and 5 cm H_2O PEEP) but developed respiratory distress during spontaneous breathing trials when positive pressure was removed. Tracheomalacia can be treated with stents[14,15] or surgery,[16–19] but our patient was too critically ill for either of these treatments. We used the tracheostomy tube and positive-pressure ventilation to stent the airway open.

Conclusion

Respiratory failure can complicate the care of patients with Morquio syndrome. Airway management can be particularly challenging due to macrocephaly, severe neck flexion, and tracheomalacia. This is further complicated by decreased chest wall compliance. Fixed neck flexion can result in external pressure applied to the tracheostomy tube, resulting in malposition of the tube, forcing the distal end into the posterior tracheal membrane. As seen in our patient, tracheomalacia can be extremely problematic. When severe airway collapse is present, the selection and

placement of the tracheostomy tube can be difficult, and the tube requires constant monitoring. Though patients with Morquio syndrome may live symptom-free with one or all of these anomalies, complications may arise when they become ill and a straight, inflexible tube is placed into an abnormally shaped trachea.

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