Discoordinate Pharyngolaryngomalacia Supported by Non-invasive Positive Pressure Ventilation in a Girl with Wolf-Hirschhorn Syndrome

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Abstract

Discoordinate pharyngolaryngomalacia (DPLM) refers to a group of patients with severe pharyngomalacia and laryngomalacia caused by discoordinate muscles of the upper airway. DPLM is considered to be associated with neurologic disorders, so it does not respond to laser supraglottoplasty as expected. Such patients used to require further surgical intervention, such as a tracheostomy, to maintain a patent airway. We present a case of DPLM with underlying Wolf-Hirschhorn Syndrome, who was able to properly maintain respiratory function by utilizing non-invasive positive pressure ventilation at home. (J Pediatr Resp Dis 2014;10:8-12)

Key words: DPLM, laryngomalacia, Wolf-Hirschhorn syndrome, laser supraglottoplasty, NIPPV

INTRODUCTION

Laryngomalacia is the most common cause of stridor in infants.¹ It has already been well-established that patients with severe laryngomalacia respond well to carbon dioxide laser supraglottoplasty.²,³ However, a portion of these patients have persistent respiratory distress despite receiving laser therapy, and were found to have co-existing pharyngomalacia. Hence, the concept of discoordinate pharyngolaryngomalacia (DPLM) was introduced by Froehlich P. et al in 1997.² DPLM was considered to be the result of a discoordinate process of the upper airway muscles resulting from a neurologic disorder. Studies and findings in the related literature are relatively scarce, and the treatments to maintain respiratory function are usually invasive, such as tracheostomy. On the other hand, non-invasive positive pressure ventilation (NIPPV) has been used increasingly in children with respiratory failure in the recent years, due to its non-invasive nature and growing evidence of its effectiveness.⁴,⁵ Herein, we report a DPLM girl with underlying Wolf-Hirschhorn Syndrome, who was effectively supported by NIPPV.

CASE REPORT

A full-term female at gestational age of 37 weeks and birth body weight of 1,930 grams was delivered at our hospital. Birth was performed via Casarean section due to prenatally diagnosed Wolf-Hirschhorn Syndrome, which was studied due to intrauterine growth retardation and oligohydramnios. She received endotracheal tube intubation immediately after birth due to respiratory distress. Respiratory distress syndrome and pneumothorax were revealed by chest plain film. She was extubated on day 6 and discharged...
without oxygen support after 46 days of hospitalization. During the admission, she was also diagnosed with a moderately-sized secundum type atrial septal defect, hypoplastic kidneys, and neurological defects, including hypoplasia of corpus callosum and aqueduct stenosis complicated with hydrocephalus.

The girl was admitted to our hospital more than ten times before she was two years old due to status epilepticus, central line infection, and aspiration pneumonia with respiratory distress. She had been intubated four times, with the durations of 4, 6, 4, and 18 days, respectively. Hoarseness and stridor, especially in the inspiratory phase, were more and more prominent after repeated intubation. Tendency towards respiratory failure due to pulmonary infection was also noted during this period of time. Therefore, flexible bronchoscopy was arranged and it was shown that the pharynx (Fig. 1) and larynx collapsed in the deep inspiratory phase, with glottis narrowing by two pieces of granulation tissue over the patient’s vocal cords (Fig. 2). DPLM with co-existing glottic stenosis was impressed.

Under the impression of glottic stenosis due to vocal cord granulation resulting from repeated intubation, laser therapy to resect the granulation tissues was performed. However, inspiratory stridor and respiratory distress were still noted as being severe after the procedure. Follow-up flexible bronchoscopy showed that the glottic stenosis had improved significantly. However, DPLM was still evident since pharynx collapse and larynx narrowing persisted during the inspiratory phase (Fig. 3). DPLM was then considered as the main cause of the deteriorating respiratory function of this patient.

Instead of performing a tracheostomy, we chose NIPPV to help maintain her respiratory function. The ventilator was variable positive airway pressure (VPAP), which could provide bi-level positive airway pressure (BiPAP) or continuous positive airway pressure (CPAP). As the nature of her disease involved the collapse of the upper airway, we believe that CPAP could help maintain a steady pressure to keep the patency of the airway. We had to customize her facial mask since there were no available masks for patients her age. She was irritable and initially had cold sweats in addition to chest wall retraction after extubation. After 9 days of training and adjustment, she could be maintained in stable condition under 8 mmH₂O of CPAP without additional oxygen support (Fig. 4). Inspiratory stridor was also less prominent. She was then discharged from our hospital, and tolerated the setting well at home, with intermittent ventilator-free periods as part of her rehabilitation. In the following 9 months, there were no further incidents of respiratory failure solely caused by DPLM.

**DISCUSSION**

This patient had Wolf-Hirschhorn Syndrome with neurologic defects, and was suffering from progressive...
inspiratory stridor after repeated intubation. Flexible bronchoscopy was performed to evaluate her airway condition, identify the etiology and to select an appropriate treatment strategy. DPLM and co-existing glottic stenosis due to granulation tissue over the vocal cords were diagnosed. She initially received laser therapy but respiratory distress persisted. Instead of invasive tracheostomy, we chose NIPPV and successfully maintained respiratory function.

Wolf-Hirschhorn Syndrome is a well-known syndrome with multiple congenital anomalies and mental retardation. The disease results from the partial loss of the distal portion of the short arm of chromosome 4. Typical features include a “greek helmet” face due to ocular hypertelorism, prominent glabella, and frontal bossing. Intrauterine growth retardation and epilepsy are two major complications. The other clinical manifestations include growth retardation, mental retardation, muscle hypotonia, congenital heart disease, antibody deficiencies, and feeding difficulties. According to a national survey in the United Kingdom in 2001, such patients are prone to severe respiratory tract infection in early childhood, and lower respiratory tract infection accounted for 25% of the causes of death.

Laryngomalacia is the most common etiology of upper airway obstruction in infancy and early childhood, and is usually benign in nature. Carbon dioxide laser supraglottoplasty was considered an effective procedure to treat severe laryngomalacia. However, 0-29% of patients with severe laryngomalacia did not respond to laser supraglottoplasty, especially those with co-existing pharyngomalacia. Therefore, the concept of DPLM was introduced by Froehlich P. et al. in 1997. It was considered as a result of the discoordinate process of the upper airway muscles, and therefore persistent respiratory distress after laser supraglottoplasty.

In the case series by Froehlich P. et al., DPLM was found in 27 of 82 children with severe laryngomalacia. Some of these patients required additional surgical intervention, including partial endoscopic epiglottectomy, adenotonsillectomy, and tracheostomy. Only 2 of these patients were successfully treated with...
BiPAP. On the other hand, 56% of DPLM patients had an associated named syndromic abnormality, and 67% had central neurological symptoms, most frequently axial hypotonia. In a retrospective review, 92% of the patients with CHARGE association, which is an association of malformation, including coloboma, heart defects, choanal atresia, developmental and growth retardation, genito-urinary malformation, and ear anomalies, had DPLM, leading to respiratory obstruction and prolonged tube feeding of such patients.

Although DPLM results from neurologic disorder and brain images might show cortical atrophy or microcephaly, no specifically location with regards to brain lesions could be identified. The diagnosis of DPLM relies on the findings of flexible bronchoscopy. Since flexible bronchoscopy can evaluate the dynamic change of the airway, it is the standard diagnostic modality to evaluate such airway problems, identify the etiology and to help to select the appropriate treatment strategy. Pharyngeal and supraglottic collapse during inspiration is the standard picture of DPLM in flexible bronchoscopy. According to Froehlich P. et al, it was defined as “severe laryngomalacia with complete supraglottic collapse during inspiration, without shortened aryepiglottic folds or redundant mucosa, and with associated pharyngomalacia”. Since differentiating DPLM from laryngomalacia alone and other etiologies of upper airway obstruction is important when selecting an appropriate treatment strategy, we must pay close attention to DPLM when we perform flexible bronchoscopy, especially when the patient is associated with neurologic defect.

Tracheostomy was the treatment of choice for upper airway obstruction for decades, but it is considered to be an intimidating and invasive procedure in the pediatric population. Serious complications may occur, including decannulation, wound complication, granuloma, pneumomediastinum, pneumothorax, and even mortality. On the other hand, NIPPV has growing use in the pediatric patients in recent years. It is less invasive than endotracheal tube intubation and preserves the ability of speech. Furthermore, it requires no sedation and is hence easier to pause, which makes it a practical choice for patients with sleep apnea or neuromuscular diseases, who may only require ventilator support intermittently. It has also been considered as safe for children with acute exacerbated asthma. Studies have suggested NIPPV can prevent extubation failure and reduce the length of hospital stay in critically ill children with heart disease. The downside of NIPPV includes the limited choices of pediatric-sized mask, the necessity of additional caregiver training, the possibility of eye irritation and nasal bridge necrosis, gastric distention, and the lack of airway protection.

This present patient was prenatally diagnosed as having Wolf-Hirschhorn Syndrome. She had axial hypotonia and feeding difficulty since birth, and corpus callosum dysgenesis was noted according to her image studies. Intractable seizure was finally controlled with the use of four different kinds of anti-epileptic drugs. However, she had progressive stridor and respiratory distress after four incidents of endotracheal intubation. DPLM and glottic stenosis caused by vocal cord granulations were diagnosed by flexible bronchoscopy. Although the granulation tissues were successfully excised by laser, stridor and respiratory distress remained severe due to DPLM. Instead of performing an invasive tracheostomy, we chose NIPPV to maintain her respiratory function and satisfactory results were achieved.

CONCLUSION

DPLM should be considered in cases of inspiratory stridor with co-existing neurologic defects, and can be diagnosed by flexible bronchoscopy. NIPPV is an alternative treatment method other than traditional intervention, such as laser supraglottoplasty, surgical uvulopharyngopalatoplasty, or tracheostomy. With an appropriate interface, NIPPV maintain long-term respiratory support for DPLM children, and improve the quality of life for patients and their families.

REFERENCES

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