Case Report

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A child diagnosed with rigid spine syndrome complicated by ventilatory disorders: a nursing case report

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Abstract

Rigid spine syndrome is a rare myopathy in children and has a poor prognosis because of its lack of treatment and eventual ventilatory failure. We report the case of a 10-year-old child with RSS and ventilatory disorders. We provided care to the child using bilevel positive airway pressure (BiPAP) non-invasive mechanical ventilation and continuous monitoring of transcutaneous carbon dioxide pressure. A 10-year-old Han Chinese girl presented (6 April 6 2016) to the Shanghai Children's Medical Center with ventilatory disorders, including hypoxia and hypercapnia. Transcutaneous oxygen saturation with mask oxygen inspiration was 90%. BiPAP non-invasive ventilator-assisted ventilation was continuously used. Through continuous non-invasive ventilation and carbon dioxide monitoring, the symptoms of dyspnea in this child were effectively controlled and improved. She was discharged on April 19 with instructions to continue using BiPAP at home and transcutaneous oxygen saturation was maintained at 94% to 98%. This case highlights that nursing of patients with rigid spine syndrome and ventilatory disorders should focus on evaluating the effect of non-invasive mechanical ventilation, prevention of complications, and continuous nursing after discharge. Additionally, continuous monitoring of transcutaneous carbon dioxide pressure is feasible for evaluating the effect of BiPAP.

Keywords

Rigid spine syndrome, nursing, non-invasive ventilation, transcutaneous carbon dioxide, partial pressure monitoring, bilevel positive airway pressure, dyspnea

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Introduction

Rigid spine syndrome (RSS) is a rare myopathy in children.[1,2] RSS can be considered as a subtype of congenital muscular dystrophy.[1,3,4] RSS has no effective clinical therapies. Early diagnosis, symptomatic supportive treatment, and rehabilitation physiotherapy are the main interventions for RSS. Ventilatory disorders are regarded as an early complication of RSS.[5] Early intervention by routine assessment of tidal volume and forced expiratory volume can prevent rapid progression of this disease to respiratory failure.

We report the case of a child with RSS and ventilatory disorders. We describe the nursing care of this child using bilevel positive airway pressure (BiPAP) non-invasive mechanical ventilation and continuous monitoring of transcutaneous carbon dioxide pressure ($TcPCO_2$).

Case report

A 10-year-old Han Chinese girl with full-term birth (from a gravida 2, para 2 mother) presented (6 April 2016) to the pediatric intensive care unit of Shanghai Children's Medical Center. She had the chief complaint of fast breathing during sleep at night, lip cyanosis for 2 months, and breath-holding attacks, which stopped when her family members woke her up. Additionally, the child could not lie flat when sleeping at night and had symptoms of chest tightness and palpitations. Her perinatal history was uneventful and development was normal, without previous treatment. Her sister was healthy, as were all other family members. The child was diagnosed with severe pneumonia and asphyxiating thoracic dysplasia syndrome by Anhui Provincial Children's Hospital in March 2016. The girl's symptoms were not completely relieved after antibiotics and mechanical ventilation. In March 2017, the child was admitted to the Orthopedic Clinic of our hospital for scoliosis. A magnetic resonance imaging examination indicated lateral curvature of the spine with obvious dilatation of the spinal cord central canal.

The patient was 150 cm, weighed 30.5 kg (third percentile), and had normal mental development. With regard to the thoracic deformity, the degree of head and neck activity was 0° of flexion and 60° of back extension. Limb muscle strength was grade IV. Electromyography showed myopathy. A muscle biopsy was refused. She had irregular and laborious breathing, as well as repeated breath-holding attacks, especially after falling asleep. Transcutaneous oxygen saturation ($TcSpO_2$) with mask oxygen inspiration was 90%. There were no abnormalities in a routine blood test. The serum creatine kinase level was l83 U/L (normal range: 30–135 U/L) and creatine kinase-MB level was 31 U/L. Arterial blood gas (ABG) analysis showed that pH was 7.25, arterial partial pressure of oxygen was 65 mmHg, arterial partial pressure of carbon dioxide was 71 mmHg, and there was obvious retention and respiratory acidosis. BiPAP non-invasive ventilator-assisted ventilation was continuously used for improvement of oxygenation and the mask fitted to the skin around the mouth and nose of the patient. The head of the bed was raised 30° to 45° during nocturnal sleeping. An indwelling oral–nasal gastric tube was used in the early stage of treatment. The patient was fed a liquid diet or semi-liquid diet, and she ate little and often to prevent gastric distension or aspiration. The skin was kept clean and the body position was changed every 3 hours to prevent pressure sores. Continuous monitoring of $TcPCO_2$ was used and ABGs were regularly monitored. A chest X-ray and lung computed tomography showed that the child had pulmonary infection. Cefotaxime 1.5 g every 12 hours intravenously and cefoperazone 1.5 g every 12 hours intravenously were

administered sequentially after admission. Re-examination of thoracic vertebrae magnetic resonance imaging on 10 April showed that the muscles around the spinal column had fatty infiltration, with muscular atrophy, and spinal physiological curvature was not obvious (Figure 1).

A gene panel for genetic muscular atrophies was performed and a mutation in the *SEPN*1 gene was detected, which suggested rigid spine muscular dystrophy 1.[1,3,4] Therefore, RSS was considered as the diagnosis. Non-invasive ventilation was only used when sleeping at night 1 week after admission. A chest X-ray re-examination showed that the infection was partially resolved. Another ABG analysis showed that the pH was 7.4, arterial partial pressure of carbon dioxide was 41.3 mmHg, and arterial partial pressure of oxygen was 89.3 mmHg. Using these treatments, the child's breathing gradually stabilized to 16 to 25 breaths/minute, and $TcSpO_2$ without oxygen inhalation was maintained at 96% to 100% in wakefulness during the day. $TcPCO_2$ was maintained at 33 to 45 mmHg. With non-invasive ventilation throughout the sleep period, $TcSpO_2$ was maintained at 96% to 100% and $TcPCO_2$ at 45 to 55 mmHg.

The child was discharged on 19 April with instructions to continue using BiPAP at home. The child and her patients were instructed in using a non-invasive ventilator and pulse oximeter, as well as for related home nursing and cleaning. Currently, the child can go to school normally during the day, but she is recommended not to participate in intense physical activity. Non-invasive assisted ventilation is still required at night, without obvious breath-holding

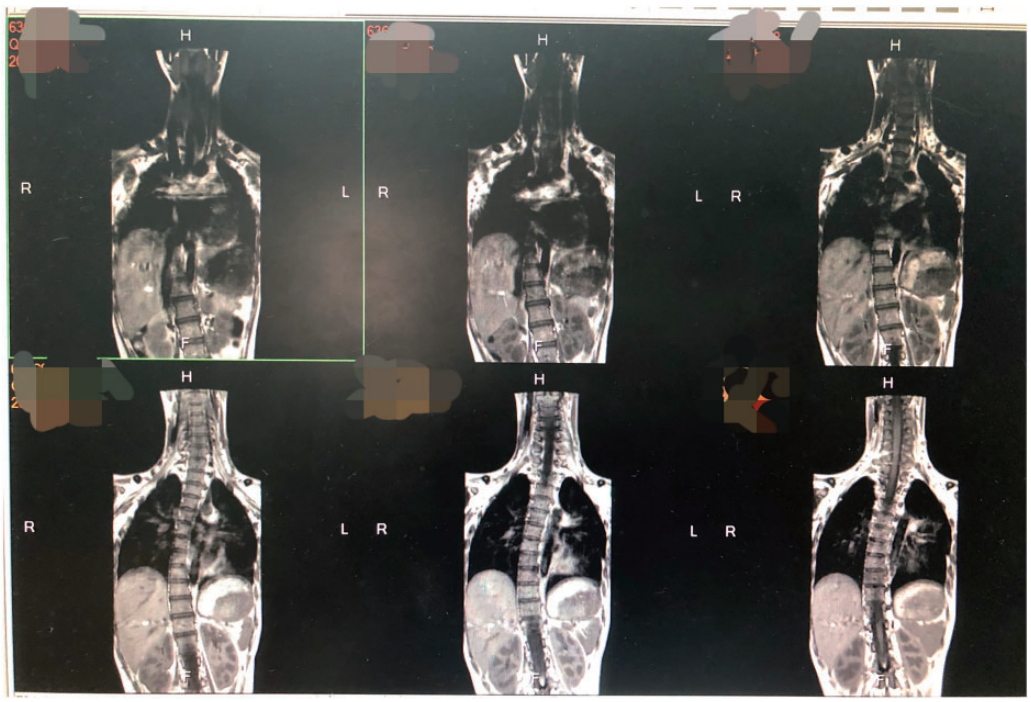

Figure 1. Thoracic vertebrae magnetic resonance imaging on 10 April showing that the muscles around the spinal column have fatty infiltration with muscular atrophy, and the spinal physiological curvature is not obvious.

attacks and cyanosis. Nevertheless, $TcSpO_2$ is maintained at 94% to 98%. The patient was followed every 6 months at the outpatient clinic. The child and family members also had access to 24/7 nursing counseling through the WeChat mobile app. The latest outpatient follow-up was on 12 May 2018. Body weight was 38 kg and height was 157 cm. Non-invasive ventilation was still used during sleeping at night, with an oxygen saturation of 94%.

The study protocol was approved by the Shanghai Children Medical Center Ethics Committee and the child's parent provided their consent for publication of this case.

Discussion

Initiation of non-invasive assisted ventilation rather than invasive mechanical ventilation is based on the patient's emotional state, whether hemodynamics are stable, and whether the child cooperates. Unlike invasive mechanical ventilation, non-invasive assisted ventilation requires children to stay awake and cooperate with the ventilator. Non-invasive ventilation masks close the children's nose and mouth tightly, and the children feel pressure when breathing. Some children will experience excessive vigilance, irritability, and incompatibility, and the treatment needs to be switched to invasive ventilation through a trachea cannula. The key to non-invasive assisted ventilation is to obtain the trust and cooperation of children in the intensive care unit without accompanying family members. The compliance of children and their families is improved after symptoms are improved, which is helpful for continuous treatment of such children. [6,7]

The Guidelines on Respiratory Management for Children with Neuromuscular Disabilities proposed by the British Thoracic Association[7] suggest that, during non-invasive ventilation, the trend of partial pressures of oxygen and carbon dioxide should be monitored continuously to follow progression of the disease. Routine ABG analysis is the gold standard for determining hypoxemia and hypercapnia, but it can interfere with the patients' life, especially during sleep. In our case, a $TcPCO_2$ and transcutaneous oxygen pressure ($TcPO_2$) monitor was adopted. The probe is placed on the skin of the anterior thoracic region or abdominal wall. The electrode temperature is 42°C and its position is changed every 2 hours during continuous monitoring. $TcPO_2$ and $TcPCO_2$ monitoring are non-invasive and continuously monitor partial pressures of oxygen and carbon dioxide, allowing dynamic evaluation of their values. The correlation and consistency of $TcPCO_2$ and $TcPO_2$ with partial pressures of oxygen and carbon dioxide from ABG have been confirmed by previous studies.[8,9] Nevertheless, there is the possibility that these correlations might be affected by a variety of factors, but the values are used to guide day-to-day management in an already diagnosed patient, not to make a new diagnosis. In our case, the data that were obtained from non-invasive monitoring and those obtained by synchronous blood gas analysis showed an error of $< 10\%$, which has good clinical significance for guiding treatment.

Morita et al.[10] showed that respiratory failure in children with RSS was mainly due to restrictive ventilation disorders and that continuous ventilator-assisted therapy was not necessary unless severe pulmonary infections occurred. Therefore, they used intermittent-assisted mechanical ventilation therapy as a supportive therapy, even though there was no respiratory failure in children. The Canadian Nursing Guidelines for non-invasive assisted ventilation and the guidelines for non-invasive and invasive mechanical ventilation for treatment of respiratory failure from the German Society of Pneumology state that

non-invasive assisted ventilation is suitable for home care when used intermittently during the night and alternating with natural breathing during the day.[11,12] The necessity and importance of continuing nursing care are pointed out in terms of the nursing particularities in rare cases of RSS. In the present case, because of using non-invasive ventilation therapy at home, the patient is being continuously followed and still achieves satisfactory results. Continuous nursing care is particularly important in China in the absence of continuity care in community practice. The cooperation of the family members and the follow-up and guidance function in the WeChat mobile app allowed the medical staff to provide some support and reassurance to the child and family members. The use of such apps should be examined in terms of satisfaction and compliance with treatment at home.

In conclusion, RSS is a rare condition. This study reported a nursing case of non-invasive assisted ventilation with BiPAP for managing special restrictive ventilatory disorders of RSS. A good therapeutic effect and compliance were achieved by combining non-invasive assisted ventilation with non-invasive $TcPCO_2$. This nursing approach could prolong survival and improve the quality of life of children with RSS and respiratory distress.

Declaration of conflicting interest

The authors declare that there is no conflict of interest.

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