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Successful Tracheal Decannulation in a Child with Congenital Central Hypoventilation Syndrome

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Abstract

Congenital Central Hypoventilation Syndrome (CCHS) is a rare genetic disorder that affects the automatic control of breathing. We report the case of a child with late presentation of CCHS at 2 years of age who initially failed noninvasive ventilation (NIV) and received a tracheostomy. Due to a mild CCHS genotype, at 7 years of age it was felt she would be a candidate for tracheal decannulation and NIV.

A stepwise approach to decannulation was utilized that included capping of the tracheostomy tube, mask desensitization, NIV with capped tracheostomy and serial polysomnogram evaluations. She was successfully decannulated and transitioned to NIV before 8 years of age. Although the process we used proved effective in this case, more research is needed to standardize transition from invasive to NIV and to establish firm criteria for tracheal decannulation in children with CCHS. Guidelines for managing CCHS exist but best practices for decannulation are not available.

Keywords: Congenital central hypoventilation syndrome; Tracheostomy; Decannulation; Noninvasive ventilation; PHOX2B; 20/25 genotype

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Introduction

Congenital Central Hypoventilation Syndrome (CCHS) is a rare genetic disorder affecting the automatic control of breathing that was first described by Mellins et al. in 1970 [1]. Prevalence is unknown due to under-diagnosis of mild cases [2]. CCHS is characterized by alveolar hypoventilation primarily during sleep but may also occur while awake, and an impaired response to hypoxemia and hypercarbia [2-4]. Symptoms typically manifest in infancy but can also present in older children as well as adults [2,4].

A diagnosis of CCHS is based on clinical findings in the absence of primary pulmonary, cardiac, neuromuscular disease or brainstem lesion and confirmed with paired-like homeobox 2B (PHOX2B) genetic testing [2-7]. There are two types of PHOX2B mutations: polyalanine repeat expansion mutations (PARMS) and non-polyalanine repeat expansion mutations (NPARMS), with PARMS representing over 90% of cases [2-4]. The normal genotype is referred to as 20/20 whereas PARMS produces genotypes of 20/24 – 20/33 [2,3]. Disorder in patients with genotypes 20/24 and 20/25 is the least severe and they typically need only nighttime ventilator support; in patients with 20/26 genotype, it varies depending upon activity level and patients with 20/27 - 20/33 genotypes require continuous support[2-4]. Milder genotypes may present as a late onset condition and be manifested after exposure to anesthesia, respiratory depressants or with severe respiratory infection [2,4,8,9].

CCHS is a lifelong disorder that does not resolve spontaneously or respond to pharmacological stimulants [2-4,6]. The level of required ventilator support depends on severity of the disorder. Positive pressure ventilation can be administered by either tracheostomy or noninvasive interface. In those requiring ventilatory support only at night, noninvasive ventilation (NIV) is preferred. In younger children, NIV is often ineffective due to difficulty ensuring adequate ventilation. Once the child is older, successful decannulation and transition to mask ventilation may be possible. We present the case of an 8 year old female with CCHS who was successfully decannulated and transitioned to NIV.

Case Presentation

A previously healthy 22 month old female with a history of difficult behavior was admitted to

the hospital following a choking episode. The child had been eating chicken nuggets when she began choking, became apneic and lost consciousness. CPR was administered and by the time emergency medical services arrived, spontaneous respirations had returned and she was conscious. The child was admitted to the local hospital for overnight monitoring and was observed to have desaturation and cyanosis upon falling asleep. She was then transferred to a pediatric facility.

No foreign body was observed with laryngoscopy and bronchoscopy. However, tonsils and adenoids were enlarged so they were removed. After the procedure she developed apnea and desaturation which rapidly progressed to respiratory failure requiring intubation and mechanical ventilation. She was later extubated to nasal cannula and weaned to room air during the daytime. Desaturation episodes in the 80% range were noted during sleep when the cannula became inadvertently dislodged. The child was discharged home with instructions to use oxygen by nasal cannula during sleep and monitor with pulse oximeter. A polysomnogram (PSG) was scheduled and was done at 2 years of age.

PSG results were abnormal with significant hypoventilation. Off oxygen, the Apnea Hypopnea Index (AHI) was 6.6, average oxygen saturation by pulse oximeter (SpO_2) was 81-96% with the lowest reading of 80%, and the average end-tidal carbon dioxide ($ETCO_2$) range was 57-61 mmHg with a highest recorded value of 65 mmHg. After adding ¼ liter per minute of oxygen, the AHI was 1.1, average SpO2 improved to 93-100% with a low of 82%, and the $ETCO_2$ averaged increased to 54-78 mmHg with a high of 83 mmHg. Supplemental oxygen had been started due to desaturation but was not increased because of CO_2 retention.

Hospital admission was arranged for genetics and neurology evaluation as well as initiation of bi-level positive airway pressure (BPAP) during sleep. Settings were titrated at bedside based upon SpO_2 and confirmed by PSG prior to going home. Magnetic Resonance Imaging of the brain was found to be normal. Although there was report of some behavioral issues from caregivers, no additional neurological evaluation was done at this time. Laboratory analysis for CCHS was done but results were not available before discharge home. Subsequently, genetic testing was positive for the PHOX2B mutation with the 20/25 genotype which confirmed a diagnosis of CCHS. There were no gastrointestinal complaints suggestive of Hirschsprung's disease, which is generally not associated with this specific genotype [2].

Initially she did not do well with BPAP at home. Discussions with caregivers regarding the need for long term ventilation were difficult due to coping with the diagnosis. Adherence did finally increase with the use of clonidine and psychology involvement. Her behavior also improved with better compliance. Despite this, the PSG remained abnormal and adequate ventilation could not be achieved with BPAP. AHI was 59.1, average SpO2 89%, and the $ETCO_2$ remained above 50 mmHg for the almost the entire study regardless of BPAP settings. Good control of ventilation could not be achieved on any of the settings tried.

Therefore, at 2.5 years of age, a tracheostomy was performed for invasive ventilation during sleep. Soon afterwards, she was able to tolerate a speaking valve during the day while awake. Supplemental oxygen was not required. She continued to be fed orally after receiving the tracheostomy but swallowing analysis revealed the need for thickened liquids. Swallowing had not been evaluated previously.

Significant improvements in both development and behavior were seen after tracheostomy placement. Prior to this, her speech had been delayed. A trial of tracheostomy capping during awake time was attempted and well tolerated. By age 3 years, she was able to maintain a capped tube during awake hours and continued ventilator support while asleep.

A follow-up PSG done at 5 years of age to assess ventilation was found to be mildly abnormal. $ETCO_2$ was above 50 mmHg for 18% of the time with a maximum of 58 mmHg. Increased tidal volume settings did not improve ventilation. However it was later noted during the study when the ventilator began auto-cycling due to tracheostomy leak, the $ETCO_2$ normalized. Ventilator adjustments were made to increase the respiratory rate and a repeat study was normal. In the interim she began kindergarten and did very well in school.

At 7 years of age, plans were made to transition to NIV. Since she was already capping her tracheostomy while awake, the next step was mask desensitization. She was fitted with a nasal mask in the clinic setting with her tracheostomy capped while using the ventilator. She was instructed to trial this at home only while awake for desensitization. She had no problem tolerating the mask and was admitted to the hospital one week later to begin nighttime use under observation. NIV with her tracheostomy capped during sleep was well tolerated so the tracheostomy tube was then downsized. Ventilator settings were adjusted after a follow-up PSG. She was discharged home with instructions to use NIV with her tracheostomy capped during sleep.

One month later, just a few days prior to her 8th birthday, she was admitted for removal of the tracheostomy tube. PSG results 3 months post decannulation using NIV initially showed both abnormal ventilation and oxygenation but normalized with a change in ventilator settings and use of a full face mask. Her quality of life has improved due to reduced risk of infection and ability to participate in activities that were previously prohibited with the tracheostomy.

Discussion

CCHS is typically diagnosed in the newborn period; however, more cases are now discovered in late infancy and childhood as well as in adults. The 20/25 genotype is one of the most common and continuous ventilator support is usually not required [2-4]. A late onset of CCHS has been associated with use of respiratory depressants or severe respiratory infection [9]. In this case, symptoms of CCHS presented as inability to respond well to hypoxemia/ hypercarbia related to a choking episode, followed by detection of hypoxemia during sleep and an episode of progression to apnea following use of anesthesia.

While there are reports of successful use of NIV in infants with CCHS [10-12], it is not recommended until 6-8 years of age in stable children for those requiring only night time support [2]. Weaning from ventilator support should never be a consideration given the underlying pathophysiology of CCHS [2]. Initial attempts with NIV in this child were difficult due to non-adherence and ultimately failed. Non-adherence may have been due to young age and behavior issues. It is possible that her difficult behavior was related to chronic night time hypoventilation as it improved after ventilation was initiated. Although her behavior was better once adherence increased with use

of NIV, a tracheostomy was ultimately necessary to obtain adequate support.

A limited number of children with CCHS have successfully transitioned from invasive to NIV [13]. While the European Congenital Hypoventilation Syndrome Network offers some guidance in this area [14], evidence based practice is lacking for the process of approaching tracheal decannulation and changing to NIV. Additionally, firm criteria need to be established to determine candidates for decannulation in those with CCHS. Continuous observation and/or monitoring are of utmost importance in managing CCHS as complete respiratory arrest or severe hypoventilation may occur at the onset of sleep [2]. When NIV is utilized, an actual ventilator should be used rather than a BPAP device since these machines are generally not approved for providing life support in the home setting [2].

We utilized a stepwise approach beginning with mask desensitization, as she already tolerated a capped tracheostomy tube while awake since 3 years of age. The next step was use of NIV during sleep with the tracheostomy tube capped under observation followed by downsizing of the tracheostomy and PSG evaluation. Subsequently, she was admitted for decannulation with additional PSGs planned. The use of multiple PSGs played a vital role in determining the adequacy of ventilator settings. Although this child was successfully decannulated and transitioned to NIV, more guidance is needed to assist practitioners taking care of individuals with CCHS who are candidates for NIV.

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