Congenital central alveolar hypoventilation syndrome (Ondine's Curse): effectiveness of early home ventilation for normal development

Y.L. Chang and P.W.D. Meerstadt

Department of Child Health, Westminster Children's Hospital, Vincent Square, London SW15 2NS, UK

Summary: An 8 month old Caucasian girl, with congenital central alveolar hypoventilation syndrome (Ondine's Curse), was discharged with her home ventilation managed by her parents. Her subsequent neurophysical development assessed at 22 months of age was satisfactory.

Introduction

Long term hospitalization is known to affect the normal development of infants. Congenital central alveolar hypoventilation syndrome (Ondine's Curse), a condition requiring ventilation, often results in prolonged hospitalization before eventual discharge can occur. We report a case that was discharged early, because intelligent parents were able to manage the ventilation, and the child's subsequent progress. In the United States, discharge as early as 2 months of age has occurred. In the United Kingdom, as far as we are aware, this is the earliest discharge to date.

Case report

Following a pregnancy complicated by polyhydramnios a female child was born to unrelated Caucasian parents, at term, by rapid vaginal delivery. She was in a poor condition at birth (Apgar scores were 4 at 1 minute, 5 at 5 minutes and 8 at 10 minutes) and required endotracheal intubation and intermittent positive pressure ventilation (IPPV).

On examination, she was a term infant weighing 4 kg, with an occipitofrontal head circumference of 36 cm and a length of 52 cm. Apart from mild hypotonia no other abnormality was noted.

After initial resuscitation in the labour ward, where she received endotracheal intubation and IPPV for 18 minutes, she did not require any further assistance and was extubated and then

discharged to the post-natal ward. However, she developed cyanosis during the first feed and became more unresponsive. An arterial stab revealed a marked respiratory acidosis (pH 6.98, PCO_2 14.5 kpa, PO_2 10.06 kpa, HCO_3 25.6 mmol/l) so ventilation was initiated. Extensive investigations for an infective, neurological, metabolic or myopathic cause were all negative. Imaging techniques, including ultrasound computed tomographic (CT) scan and nuclear magnetic resonance scan, were also negative. Respiratory function tests showed a rise in the arterial CO_2 during sleep states but normal values were obtained when she was awake. Oxygen saturation could be maintained both when asleep and awake. Breathing movements of the chest wall also occurred whilst asleep and awake. This was in keeping with a diagnosis of congenital central alveolar hypoventilation syndrome (Ondine's Curse).

A tracheostomy was performed at one month of age in preparation for long term ventilation and the child was gradually weaned off the ventilator such that it became necessary only to ventilate during periods of sleep. During periods awake she breathed spontaneously. Other avenues of treatment were explored. She had a trial of negative pressure ventilation, but this failed to maintain adequate gases. Drug therapy, in the form of respiratory stimulants, was tried but was found to be unsuccessful.

Because of the problems known to be associated with hospitalization on an infant's normal development¹ we sought to provide the equipment and nursing support to enable ventilation at home, a more normal environment. The articulate, intelligent professional parents were taught to manage the ventilator and basic cardio-pulmonary resuscitation technique. Provisions were made for rapid hospital admission in case of emergency, along

Correspondence: Y.L. Chang, Department of Paediatrics, Kingston Hospital, Wolverton Avenue, Kingston upon Thames, Surrey KT2 7QB, UK
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with an open access to discuss any problem or worry.

Prior to her discharge the child was developmentally delayed. At 7 months of age, she functioned at the 5–6 month level for gross motor skills; fine motor skills were at the 4 month level; social skills were age appropriate. Since her discharge home, she has caught up in these areas. When assessed at 22 months of age, by a Ruth Griffiths Profile, she had a developmental age at the 18 month level, with a general quotient of 81. She has needed two admissions due to chest infections that compromised her ventilation requirements.

Discussion

Ondine's Curse (congenital central alveolar hypoventilation syndrome) has its roots in German literature² and the term was applied in medicine by Severinghaus in 1962.³ It represents a failure of automatic control of respiration by the central nervous system.^{4–6}

Its management requires some method of maintaining respiration whilst asleep. Drugs have been tried, but the most promising, almitrine, has been limited by the side effect of neuropathy.⁷ Diaphragmatic pacing has been used but the experience is limited in paediatric practice and complications occur.⁸ There is therefore the need for some form of ventilation, be it by negative pressure tanks or by positive pressure ventilation. Prolonged positive pressure ventilation will require the formation of a tracheostomy. This will, however, hinder the child's ability to develop speech, even though at times, they can speak around the tube. The use of speaking tubes will further aid speech development. A programme of voice, speech and language stimulation will help lessen the delay in acquiring the receptive and expressive communication skills.^{9,10}

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The establishment of home ventilation for a child requires long term planning and this may lead to prolonged in-patient care, often in the intensive care unit of the hospital. Not only may this impair the child's normal development¹ but the cost of intensive care management, as compared to community care, is significantly greater.¹¹

The impact on the family of a child who is ventilator-dependent has been studied. In the United States, where there has been more experience, it has been found that parents, with the help of therapists, could come to terms with their child's illness. The parents were also able to use the therapist to help voice their fears and concerns to the ever changing sea of medical attendants.^{12,13} Other family members could also be affected by the new arrival. Siblings tended to be more protective to the child with the tracheostomy. Some were more 'clingy' whilst others showed more mature behaviour. Some of the sibs could help in basic tracheostomy care.¹⁴ Children with tracheostomies appeared to cope well with their ventilator dependence, but their emotional reactions will need to be explored as they develop and reach a stage to express themselves appropriately.

The attempt at early discharge, in this case, was successful because suitable funding and intelligent parents were at hand. The normal community services were allocated some additional support to help carry out their task. In the United States, the system has been well developed so that discharge as early as two months of age has been achieved.¹⁰

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