

Prolonged expiratory apnoea with cyanosis in Arnold Chiari II malformation

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Lesson

Apnoea associated with Arnold Chiari malformation is a known entity and can be obstructive or central. Differentiating between two types is vital to deciding management pathway and prognosticating disease process.

Keywords

central apnoea, Arnold Chiari II malformation, cyanotic expiratory apnoea

Introduction

Southall et al.¹ first presented the notion of prolonged expiratory apnoea in children in 1985, demonstrating a relationship between upper airway obstruction and severe hypoxemia. These may be obstructive or central in origin. Obstructive apnoea in some patients with Arnold Chiari malformation may be reversed by optimal control of hydrocephalus with or without cervical decompression. These patients may also develop episodes of cyanotic expiratory apnoea of central origin (prolonged expiratory apnoea with cyanosis). This form of apnoea does not respond to surgical or medical treatment and may show progressive worsening over time. Compression with traction on the cranial nerve, particularly of cervical rootlets of the vagal nerve which innervate the cricoarytenoid muscle for vocal cord abduction, produces ineffective vocal cord abduction in these patients.²

Case report

This is a report of a baby girl who was born at 35 weeks' gestation with antenatal diagnosis of spina bifida. She was born to young parents who were non-consanguineous. Postnatal magnetic resonance imaging diagnosed Arnold Chiari malformation type II associated with hydrocephalus. A ventriculoperitoneal shunt was inserted for hydrocephalus at two weeks of age. She was discharged home after recovering from the surgery. At one month of age, she presented to accident and emergency department

with breathing difficulty and distress. She was clinically diagnosed with bronchiolitis at this point. She deteriorated on the fourth day of illness and had to be intubated and ventilated because of an episode of prolonged apnoea. During her stay on paediatric intensive care unit, she had multiple apnoeic/desaturation events and this could have prevented any attempt at extubation.

Failure to extubate prompted investigating for a cause. Micro laryngeal bronchoscopy revealed bilateral vocal cord palsy. Speech and language therapy team reported that her swallow was unsafe on assessment. In view of her apnoeic events and vocal cord palsy, a decision was made to perform a tracheostomy and start non-invasive ventilation. Despite being ventilated, she had multiple profound and life-threatening desaturation episodes. Most were self-terminating, lasting up to 10 s, some requiring stimulation and a few requiring bag-tracheostomy ventilation. These episodes were attributed to her underlying neurological condition, i.e. Arnold Chiari type II malformation. Polysomnography established the apnoeic episodes and the sleep physiologist determined them to be central in origin. She was reviewed by neurologist who formally diagnosed them as prolonged expiratory apnoea with cyanosis and correlated them with type II Chiari malformation. She was sent home with a care package that included overnight non-invasive ventilation by NIPPY in form of pressure support bilevel positive airway pressure, suction equipment and oxygen supply. Non-invasive ventilation is given while she is asleep. She is not on any ventilator support while awake. She has since then been admitted a few times to hospital with recurrence of prolonged apnoeic episodes lasting up to 30 s and requiring bag-tracheostomy ventilation. During these periods while in hospital, she has been given day and night non-invasive ventilation for a day or two, and then weaned down to night time ventilation which is what she gets at home.

Discussion

Patients with Chiari malformation can develop lower cranial nerve disturbances, including absent gag reflex, neurological alterations of upper limbs, cerebellar problems, and respiratory problems (45%–65%), and up to 30% of patients die during the first two decades of life, mainly because of respiratory failure.³ Most (85%–90%) develop hydrocephalus and require decompression of the ventricular system. In severe cases, tracheostomy and nocturnal ventilation may be required, particularly when there is failure of less invasive modes of assistance.⁴ Moreover, while approximately 30% of Chiari malformation type II children have normal sleep-breathing, 20% of these children show severe sleep-disordered breathing, apnoea, hypoventilation and arousal deficits.⁵ These may predispose to prolonged apnoea, which could increase the risk of sudden death during the first two years of life.⁶ It is believed that apnoea may result from central neural dysfunction (centrally mediated expiratory apnoea with cyanosis) and should be distinguished clinically if not etiologically from bilateral abductor vocal cord paralysis which is obstructive as opposed to central apnoea.⁷ Bilateral abductor vocal cord paralysis may be precipitated suddenly with hydrocephalus and increasing intracranial pressure. The patients can develop severe inspiratory stridor and asphyxia. The transient nature of the stridor and the reversibility of the bilateral abductor vocal cord paralysis after the relief of the increased intracranial pressure are seen as an argument against the involvement of a selective structural lesion of the brainstem.

Persistent hydrocephalus is correlated with a less favourable prognosis of vocal cord function return and not an immediate relief is a priority.⁸ Cochrane et al.,⁹ in a small number of patients, have also delineated between obstructive and central apnoea and have found that obstructive apnoea is usually reversed with an optimally functioning ventriculoperitoneal shunt, whereas central apnoea does not respond to cervical decompression. Another lethal respiratory sign is prolonged expiratory apnoea with cyanosis. It manifests itself as a total cessation of expiratory effort with cyanosis, which can also occur in mechanically ventilated patients.

Most common differentials are post sigh central apnoea and prolonged expiratory apnoea of cyanosis.¹⁰ This is a benign condition. Post sigh central apnoea differs from pathological central apnoea in several important ways. Post sigh central apnoea is preceded by an augmented breath and brief (<3 s) electrocortical arousal noted on electroencephalogram. Pathological central apnoea is typically not preceded by an augmented breath or arousal. Our

patient suffered with apnoeic episodes both while awake and during sleep. These episodes happened even when she was ventilated either invasively or non-invasively. They were not preceded by electrocortical arousal or augmented breath. Oxygen saturation rarely fell more than 3% from baseline. They were hence diagnosed as prolonged expiratory apnoea with cyanosis.

Management of respiratory abnormalities in Chiari malformations depends on the extent of problem. Isolated hypoxemia and mild central apnoea will need supplemental oxygen. Hypopnea associated with hypercapnia requires management with bilevel positive airway pressure non-invasive ventilation. From a surgical perspective, craniocervical decompression should be done at onset of brainstem dysfunction associated with severe central apnoea. Ventriculoperitoneal shunt is indicated for hydrocephalus. Finally, tracheostomy mechanical ventilation is needed for irreversible brainstem damage in children who have progressively got worse over the years. In children who had developed particularly severe impairment with bilateral vocal cord paralysis and apnoea, the prognosis was poor.¹¹

Declarations

Competing Interests: None declared.

Funding: None declared.

Ethics approval: Written informed consent for publication was obtained from the patient.

Guarantor: AZ

Contributorship: AZ wrote the article and sought consent from parents. NH helped with literature search for discussion and edited discussion before submission.

Acknowledgements: None

Provenance: Not commissioned; peer-reviewed by Zhe Wu.

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