

Arnold-Chiari Malformation Type 1 as an Unusual Cause of Acute Respiratory Failure: A Case Report

Mohamad Khatib¹, Moustafa Elshafei¹, Amr Shabana¹, Dnyaneshwar Mutkule¹, Dinesh Chengamaraju¹, Nevin Kannappilly¹, Nazeer Alaudeen¹, Ruchir Joshi¹, and Abdulqadir Nashwan¹

¹Hamad Medical Corporation

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Abstract

Chiari malformations are a group of heterogeneous disorders; Chiari I malformation is characterized by a form of structural defect in the cerebellum and the cranial base. The case report highlights the possibility of underlying neurological disease in cases where acute Type 2 respiratory failure is unexplainable.

Key Clinical Message

The authors urge clinicians to consider the possibility of Arnold-Chiari Malformation Type 1 with other central causes especially in cases where acute respiratory failure is unexplainable.

Background

Historically, Hans von Chiari described certain hindbrain abnormalities as postmortem findings in infants; these came to be known as Chiari malformations. Four types of Chiari malformations are described in the literature: types I, II, III, and IV.^{1,2}(Chiari malformation types II, III, and IV are distinct from type I and are not discussed in this article). Chiari I malformation is the most common, having been estimated to occur in 1 in 1000 births.¹

Arnold Chiari type I malformation (Type-I ACM) is characterized by cerebellar tonsils herniation and downwardly displaced below the level of the foramen magnum. Typically, tonsils lying 5 mm or more (normally 3 mm) below the foramen magnum on neuroimaging are consistent with an ACM. However, there is no known direct correlation between clinical severity and the tonsils position.

The actual nature of CM-I has not been fully understood.² In most cases, CM-I does not become symptomatic until adolescence or adulthood³⁻⁵ and the mean age at presentation is approximately 18 years.⁶ Also, symptom onset is often insidious. Usually, Symptoms of Type-I ACM develop as a result of the following mechanisms; cerebellum, medulla, and upper spinal cord compression, and disruption of CSF flow through the foramen magnum. Medulla and spinal cord compression may result in spinal cord injury and lower cranial nerve and nuclear dysfunction. Also, the cerebellum compression may result in various neurological symptoms such as dysequilibrium, dysmetria, ataxia, and nystagmus. Disruption of the Cerebral Spinal Fluid (CSF) flow through foramen magnum probably leads to the most common symptom, which is pain.

Case Presentation

A 37-years-old, healthy, non-smoker male construction worker attended our emergency department with a history of dry cough, fever, and shortness of breath for three days. His respiratory rate was 20/min with O2

Saturation of 94% on a nasal cannula 4 L/m, hypotensive on norepinephrine, looked confused but obeyed commands.

Chest radiography showed Non-homogeneous opacities in the right mid and lower zones. Arterial blood gas on admission revealed hypoxemia (PaO₂ 71 mmHg) and mild hypercapnia (PaCO₂ 43 mmHg), high bicarbonate concentration (23.6 mmol/L) and pH was 7.366 with normal lactate. Initially, he was started on broad-spectrum antibiotics. His oxygenation maintained with Nasal canula initially. Subsequent ABGs showed elevation PaCO₂ and was supported with Bilevel positive airway pressure ventilation (Bi-PAP) alternated with high flow nasal cannula. The respiratory viral panel came positive for Rhinovirus PCR & Human Metapneumovirus PCR, and septic workup were negative.

He improved clinically and weaned from norepinephrine for over 24 hours. However, he continued to require BiPAP due to type 2 respiratory failure with noted episodes of bradypnea (8-10 BPM) and dropped oxygen saturation during sleep. Drug overdose was ruled out, and thyroid functions were normal. He developed mild occasional difficulty of swallowing on 6th day of hospital admission.

In view of persistent type 2 respiratory failure which is unexplainable by pulmonary pathology and mild occasional difficulty of swallowing and bradypnea Neurological evaluation was done showed absent gag reflex, horizontal nystagmus, and wide base walking. Deep tendon reflexes were exaggerated with impaired sensation in lower limbs. Thin liquid- post swallowing cough was noted, pain while swallowing, so feeding nasogastric tube was inserted. MRI brain and Cervical Spine (see Fig 1 & Fig 2) was done, which revealed Arnold Chiari malformation with upper cervical syrinx suggestive of a Type-I ACM with associated cervico-medullary junction compression. The patient was transferred to the Neuro-surgery Unit for surgical decompression with cervical fixation.

Discussion

Respiration is regulated by two separate neural mechanism – voluntary and automatic.⁷ The automatic process is regulated by the respiratory centers in the brainstem. The chemoreceptors in the carotid and aortic bodies send signals to the medulla which innervates the respiratory muscles. In awake state there is a continuous voluntary mechanism to maintain ventilation. During sleep the voluntary mechanism is lost and anatomical and functional integrity become more important.⁸

Here in this case our patient had a healthy life with no prior history of any chronic respiratory illness until he developed a viral pneumonia. He developed acute type 2 respiratory failure requiring noninvasive ventilation. His arterial blood gas characteristics were suggestive of alveolar hypoventilation. His respiratory failure did not improve even after the treatment of pneumonia. Persistent unexplainable respiratory failure and bradypnea made us think of an alternate diagnosis of central cause for hypoventilation which was supported by his new onset occasional difficulty in swallowing. MRI substantiated our diagnosis. His hypoventilation was masked until he had a normal life. Central hypoventilation became evident when he had increased demand of ventilation due acute illness.

Type-I is the most common form of ACM and has been estimated to occur in 1 in 1000 births and mostly asymptomatic. ACMs cases are often detected coincidentally while undergoing unrelated investigations.⁹ Although ACM is still listed as a rare disorder by the Office of Rare Diseases Research (ORDR), this classification is based on outdated data, mostly before the MRI era. The introduction of MRI has increased the frequency of ACM case discoveries.

The prevalence rate of Type-I ACM is ranged between 0.1-0.5% and less predominant in males.¹⁰ Genetic basis for Type-I ACM has been often suggested.^{11,12} Recent studies favor the presence of genetic linkage to chromosomes 9 and 15.¹³ Other researchers suggested that Type-I ACM is a disorder of primary para-axial mesodermal origin, which could affect the formation of axial skeletal defect and different neurological anomalies.¹⁴

Type-I ACM is a common structural disorder characterized by motor deficiency, sensory loss, lower cranial palsy, and cerebellar syndrome.¹⁵⁻¹⁸ Neurological symptoms associated with Type-I ACM are including mild

to severe sensory deficit, lower cranial nerve palsies, headache, neck pain, and ataxia. Respiratory-related signs and symptoms seems to be unusual¹⁹ or properly unmasked by other conditions,¹⁷ and often associated with central alveolar hypoventilation^{17,19} which is usually the trigger for clinicians to consider ACM not the respiratory failure, and this is typically what led us to make this observation. To the authors' knowledge, respiratory failure as the first manifestation of ACM has only been reported once.¹⁵

Conclusions

In conclusion, we urge clinicians to consider the possibility of ACM with other Central causes in cases where acute respiratory failure is unexplainable.

Abbreviations

ACM: Arnold Chiari malformation

CNS: Central Nervous System

CSF: Cerebrospinal Fluid

Declarations

Ethics approval and consent to participate

The article describes a case report. Therefore, no additional permission from our Ethics Committee was required.

Consent for publication

The consent for publication was obtained.

Availability of data and material

All data generated during this study is included in this published article.

Competing interests

The authors declare that they have no competing interests.

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Authors' contributions

MKA: Data Collection, Literature Search, Manuscript Preparation

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All authors read and approved the final manuscript

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Figure Legends

Figure 1 A&B Sagittal magnetic resonance image of the brain. Note the cervico-medullary junction compression

Figure 2 Axial C-spine T2-weighted magnetic resonance image of the upper cervical spinal cord at the level of C1-2.

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