CASE REPORT

Treatment of respiratory insufficiency in arthrogryposis multiplex congenita with non-invasive ventilation

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Summary
Arthrogryposis Multiplex Congenita (AMC) consists of multiple joint contractures at birth. Chest wall deformities and respiratory and muscle weakness causing altered chest wall mechanics can lead to respiratory failure, which is often present from birth and is usually fatal in early life. Increasingly, patients are surviving into adulthood. We describe an adult presenting with acute hypercapnic respiratory failure (AHRF) and obstructive sleep apnoea on a background of AMC and obesity who responded to chronic domiciliary non-invasive ventilation.

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Arthrogryposis Multiplex Congenita is usually fatal during childhood. It is characterised by fixed stiffness of articular joints secondary to reduced antenatal foetal movements. Respiratory problems are secondary to thoracic cage deformity, neurogenic muscular atrophy, myopathy1–3 or a combination.

Adults rarely present with respiratory insufficiency due to AMC.4,5 We report an adult with AMC presenting with respiratory failure and sleep apnoea hypopnoea syndrome (SAHS) who responded to non-invasive ventilation (NIV).

Case report
A 33-year-old man presented with a two-day history of breathlessness, productive cough and confusion. He was a non-smoker with no known chest disease. During childhood, he had undergone multiple orthopaedic
operations — culminating in bilateral lower limb amputations aged 7 due to multiple joint contractures. There was no family history of note. Further questioning revealed a two-year history of progressive tiredness, early morning headaches and daytime somnolence — symptoms he blamed for losing his office job.

Examination revealed the “classical” (amyoplasia) AMC variant. He exhibited marked kyphoscoliosis, increased anterio-posterior chest diameter and reduced chest expansion without abdominal paradox. His adjusted Body Mass Index was 37.8 kg/m². His Epworth Sleepiness Score (ESS) was 14/24.

Arterial blood gas tensions on air revealed: pH 7.27, pO₂ 4.1 kPa, pCO₂ 9.2 kPa, HCO₃⁻ 30.7 mmol/l. A chest radiograph showed bilateral lower lobe consolidation (Fig. 1). Sputum culture yielded a significant growth of Pseudomonas aeruginosa. He responded to intravenous antibiotics, hydration and controlled oxygen therapy. At discharge, arterial blood gases on air were pH 7.37, pO₂ 8.1 kPa, pCO₂ 6.7 kPa, HCO₃⁻ 28 mmol/l.

Lung function tests revealed a forced expiratory volume in 1 s (FEV₁) 1.15 L and forced vital capacity (FVC) 1.51 L, 32% and 36% predicted for his age and (adjusted) height. His total lung capacity was 2.2 L and residual volume 0.59 L, both 35% predicted for his age and height. The carbon monoxide transfer coefficient (Kco) was 138% predicted. His FVC did not fall in the supine position. Maximum inspiratory mouth pressures were reduced at 53 cm H₂O (normal 120 cm H₂O).

At review three weeks post-discharge, he complained of worsening daytime somnolence with an ESS of 16/24. Sleep limited channel studies (Embletta, Flaga, Reykjavik, Iceland) confirmed low baseline mean oxygen saturations of 86.5% and a mean 4% dip rate of 27.4/h, with associated pulse rises.

Fluoroscopy showed normal diaphragmatic movement. However, inspiratory and expiratory thoracic cage movements were greatly reduced, particularly at costo-vertebral joints. Chest computed tomography (CT) scan showed fusion of the costo-vertebral joints at various levels (Fig. 2).

Otto first described Arthrogryposis Multiplex Congenita (AMC) in 1841. Subsequently, in 1905, Rosencranz reported an antenatal syndrome presenting in the immediate postnatal period with multiple joint contractures. Dislocation of hips and other joints, marked flexion contractures of the knees, wrists and elbows, talipes equinovarus and scoliosis are usual. The disease may be localised to either upper or lower limb. AMC is diagnosed when two or more joints in more than one limb are fixed from birth.

Contractures are usually nonprogressive, improving over time with physiotherapy. The causes of AMC include neurological, myopathic, skeletal and external causes. Reduced active movement in utero provokes retraction of muscles and tendons leading to deformity. Respiratory insufficiency is seen frequently in affected newborn babies and is invariably fatal in childhood.

AMC occurs sporadically, affecting 1 in 3000 live births. Survival into adulthood was rare. With diagnostic advances and early treatment of the condition, patients are now living beyond 30 years of age.

We are aware of one other case report of a patient with this disease presenting with chronic respiratory insufficiency requiring domiciliary ventilation at the age of 35 years.
The cause of respiratory insufficiency in this case was multi-factorial. Poor in utero development of the thoracic cage probably led to kyphoscoliosis. His lowered vital capacity and reduced inspiratory mouth pressures suggest a significant component of respiratory muscle weakness, compounded by obesity. Additionally, fluoroscopic analysis of thoracic cage movement revealed a paucity of rib movements with minimal increase in lateral and dorsoventral rib cage diameters with inspiration. Radiographically, there was an element of costo-vertebral fusion, compounding muscle weakness and obesity. We also demonstrated evidence of moderate SAHS, probably due to obesity and possible weakness of oropharyngeal dilator muscles in combination. The increased $K_{CO}$ probably reflects increased pulmonary flow, observed in obese subjects.

With increasing survival, more patients are likely to present with its protean ways of causing respiratory insufficiency. These patients may benefit from NIV in an acute and domiciliary setting potentially further improving patient survival and long-term quality of life.

Conflict of interest statement

None of the authors have a conflict of interest to declare in relation to this work.

References