CASE REPORT

Congenital fibre type disproportion: an unusual presentation of respiratory failure

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SUMMARY

A 38-year-old lady, with a weeklong history of flu-like illness, presented acutely with type-2 respiratory failure needing mechanical ventilation. She had a known history of a rare subtype of congenital myopathy called congenital fibre type disproportion myopathy (CFTDM). This condition most commonly presents as hypotonia in neonates and is characteristically non-progressive. It may present in children as generalised weakness, delayed milestones and skeletal deformities such as scoliosis and myopathic facies. To the best of our knowledge this is the first reported case of CFTDM contributing to respiratory failure in an adult patient. She made good recovery in four weeks and was discharged home on intermittent non-invasive ventilatory support.

Key Words: Congenital fibre type disproportion; Myopathy; Mechanical ventilation.

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INTRODUCTION

Congenital fibre type disproportion myopathy is a condition, first reported in 1973 by M. H. Brooke[1], who described characteristic shortening of type-1 skeletal muscle fibres in 12 children. The chief initial presentation in this case series was weakness and hypotonia in neonates. A positive family history has been reported in 40% of cases. The disease is said to be transmitted as an autosomal recessive trait but there are exceptions. Several reports of CFTDM have disclosed an autosomal dominant inheritance as well[2,3]. Since this term was coined, there have been other case reports describing associated features including delayed motor milestones, scoliosis of the lumbar and thoracic spine, myopathic facies, high-arched palate, multiple joint contractures, and in severe cases, respiratory and bulbar weakness[4,5]. It is very unusual for CFTDM to cause respiratory weakness during adulthood.

CASE REPORT

A 38-year-old lady, who was born of non-consanguineous parents, was admitted as an emergency to the Adult Respiratory Care Unit (ARCU) at St James's University Hospital, Leeds. She presented with a flu-like illness, dry cough and worsening shortness of breath. She also had associated proximal muscle weakness, myalgia and exhaustion. Three months before this admission, she was investigated for iron deficiency anaemia with a gastroscopy and colonoscopy that were essentially normal. As a neonate, she was found to be a 'floppy baby' diagnosed with CFTDM after a muscle biopsy at eight months of age (which depicted selective atrophy of type-1 fibres). She had complaints of tiredness after exertion at school, and moderate proximal muscle weakness at shoulder and pelvic girdle, other than this she had no significant problems during childhood. She worked as a clerk and had never smoked or drunk alcohol excessively. She lived with her husband and two children; her eldest

son was diagnosed with cerebral palsy at birth. There was no family history of congenital myopathies.

On admission to ARCU, a physical examination revealed a small chest with crowded ribs, scoliosis and a prominent lumbar lordosis. She had a pale, myopathic face but examination of muscle tone, power, sensation and co-ordination in all four limbs was unremarkable. The deep tendon reflexes were globally diminished even with reinforcement techniques. She was alert and orientated, eye movements and pupillary reflexes were normal and no cranial nerve pathology or bulbar muscle involvement was noted on examination. A thorough general physical as well as cardiovascular, respiratory and gastrointestinal systemic examination did not reveal any abnormalities. Her temperature was 36.8°C, respiratory rate 12/min, blood pressure 117/80 mmHg, pulse rate 120/min and her oxygen saturation measured by pulse oximetry was 87% on room air. Initial arterial blood gas analysis on air showed a respiratory acidosis and a type-2 respiratory failure (pH 7.28, PCO_2 10.06 kPa, PO_2 6.40 kPa, base excess +8.10 mmol/L, bicarbonate 34 mmol/L, lactate 1.77mmol/L). She was transferred to the critical care unit (CCU) for an initial trial of non-invasive ventilation (NIV), but failed to show any improvement in her clinical condition or blood gases. Therefore, she was intubated and mechanically ventilated. On the third CCU day, her blood gases improved; hence, she was extubated and put on NIV. She was discharged to ARCU on seventh day.

She developed hospital acquired pneumonia a week after discharge from critical care that was successfully treated with broad spectrum antibiotics (inj. vancomycin and inj. meropenem) for seven days until her blood and sputum cultures were negative. She was investigated with spirometry, which showed a severe restrictive pattern. The forced expiratory volume in the first second (FEV_1) was 2.85 L (40% predicted), forced vital capacity (FVC) was 3.3 L (38% predicted), FEV_1/FVC ratio was 82%. The vital capacity was 3.28 L, total lung capacity was 4.90 L, residual volume was 1.54 L, peak expiratory flow rate was 400 L/min, and diffusion capacity for carbon monoxide (DLCO) was 8.6 mmol/kPa.min. Electromyogram and nerve conduction studies were normal. Chest x-ray revealed a right lower lobe consolidation. Parainfluenza and respiratory syncytial viruses were positive from bronchoalveolar lavage and sputum samples. Computed tomogram and pulmonary angiography, autoantibody screen, thyroid function tests and creatine kinase levels were normal. The lady made good recovery and was discharged home on intermittent NIV.

DISCUSSION

Congenital myopathies generally cause weakness and wasting of muscles and present early in life, often at birth, with a variety of symptoms. Congenital fibre type disproportion refers to the significantly shorter length of type-1 muscle fibres in comparison to the type-2 fibres on microscopy. It is usually nonprogressive or slowly progressive and the weakness may improve after the second year of life. The exact pathogenesis of the disorder is unknown. Like other myopathic conditions, there is no specific cure and treatment is mainly supportive.

There has been some controversy regarding the existence of CFTDM as a separate nosological entity because many neuromuscular conditions mimicking the type-1 fibre hypotrophy have been described. Over the last few decades, several central nervous system, peripheral nerve and anterior horn cell diseases have been shown to share a similar histological appearance. Furthermore, there is a lack of consensus on the diagnostic criteria for CFTDM based on the histopathological features and clinical course[6,7].

CFTDM is considered a static muscle disorder and generally carries a benign prognosis, with no significant deterioration after childhood, and even some improvement with age. However, the natural course of CFTDM can be quite variable; in up to 25% of cases the weakness can be severe and associated with recurrent respiratory failure and death in children [8,9]. In this case report, we described the symptoms and clinical signs corroborative of a viral illness precipitating a respiratory failure in a patient with an underlying congenital myopathy. There are no previously reported cases of such a peculiar contribution of CFTDM to respiratory failure needing mechanical ventilation in an adult patient.

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