Anaesthesia in arthrogryposis multiplex congenita: case report

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The anaesthetic management of an 11-year-old male with arthrogryposis multiplex congenita (AMC) with myopathic features is described. The child presented with an obstructing renal calculus and a pyelolithotomy via an anterior transverse subcostal extraperitoneal incision was performed. In view of the associated hypoplastic musculature, severe scoliosis and expected increased sensitivity to various sedatives and inhalational anaesthetic agents, low-dose ketamine ($10\text{--}15\text{ }\mu\text{g}\cdot\text{kg}^{-1}\cdot\text{min}^{-1}$) was administered by continuous infusion, with a satisfactory result.

Arthrogryposis multiplex congenita (AMC) is a descriptive term for a syndrome presenting with symptoms of multiple persistent joint contractures with myopathic or neuropathic features. There is muscular hypotonia and wasted muscle mass. The myopathic type is more common and is commonly associated with scoliosis and malignant hyperpyrexia. Other associated abnormalities include skeletal deformities, congenital heart disease and genitourinary system anomalies.¹⁻³ Such patients may present for incidental or corrective surgery requiring general anaesthesia.

Case report

An 11-year-old male weighing 11 kg, suffering from AMC, presented with episodic pain in the left flank. Diagnosis of a left obstructing renal calculus was established and the patient was scheduled for pyelolithotomy via an anterior transverse extraperitoneal incision.

On examination he was found to be an intelligent child who was doing well in his studies. He had been delivered

by Caesarean section at full term, with breech presentation and was a "small-for-dates" baby. His six-year-old brother also had generalised features of AMC (Figure 1).

The patient had myopathic features associated with malformation of the joints, causing limitation of bilateral knee movements and a thumb-in-the-palm deformity of both hands. He had facial asymmetry, ptosis of both eyes, orbital hypertelorism, high arched palate and caries of the teeth. No cardiovascular abnormality was noted during the examination.

Preoperative investigations including blood count, serum electrolytes, acid and alkaline phosphatase, serum calcium and phosphate were within normal limits. Serum creatine phosphokinase (CPK) levels were $880\text{ I.U.}\cdot\text{L}^{-1}$ (normal $0\text{--}70\text{ I.U.}\cdot\text{L}^{-1}$).

The patient was not sufficiently cooperative to allow pulmonary function testing. Preoperative blood gas analysis revealed PaO_2 87 mmHg, PaCO_2 of 42.6 mmHg. His chest roentgenogram, taken in the standing position (Figure 2) revealed thoracolumbar scoliosis towards the left with an 80° angle of primary curve. Muscle biopsy taken from a gastrocnemius muscle showed small-for-age muscle fibres, with fibrous and fatty degeneration of the muscle.

Premedication with promethazine 10 mg PO was given one and one half hours before surgery. After pre-oxygenation, anaesthesia was induced using ketamine $1\text{ mg}\cdot\text{kg}^{-1}$ and laryngoscopy was carried out to assess the airway, before administration of pancuronium, 1 mg, to facilitate tracheal intubation.

Pyelolithotomy was performed in the supine position via an anterior transverse subcostal extraperitoneal incision. The procedure lasted two and one half hours and anaesthesia was maintained with an infusion of ketamine at a rate of $10\text{--}15\text{ }\mu\text{g}\cdot\text{kg}^{-1}\cdot\text{min}^{-1}$. Monitoring of ECG: arterial blood pressure (radial artery cannula), and nasopharyngeal temperature was carried out. No further doses of pancuronium were needed. The patient was ventilated with an air/oxygen mixture with a FiO_2 of 0.3–0.4. The anaesthetic course was uneventful and the patient was reversed at the end of the procedure with $40\text{ }\mu\text{g}\cdot\text{kg}^{-1}$ of neostigmine and $20\text{ }\mu\text{g}\cdot\text{kg}^{-1}$ of atropine. The postoperative course was uneventful.

Key words

ANAESTHETICS, INTRAVENOUS: ketamine; GENETIC FACTORS: arthrogryposis multiplex congenita.

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FIGURE 1 Photograph of the patient (postoperative) and his younger brother aged six years.

Discussion

AMC was first described by Otto in 1841, who referred to it as congenital myodystrophy. The current term was suggested by Stern in 1923 from two Greek words meaning "crooking of joints." AMC is monogenically inherited and most likely due to homozygosity for an autosomal recessive gene. A prominent feature of AMC is contractures (mostly flexion type) and the muscles at the affected joints are weaker than those at the normal joints. There can be involvement of the spine, leading to hyperlordosis or scoliosis. About ten per cent of affected patients have associated congenital heart disease, including patent ductus arteriosus, aortic stenosis, coarctation of the aorta and cyanotic congenital heart disease. The patients' appearance has been likened to "wooden doll." There is scant subcutaneous tissue and muscle.^{1,2}

Arthrogryposis has been observed following the treatment of maternal tetanus with tubocurarine in the tenth week of pregnancy. It may also be seen in a number of

FIGURE 2 Anteroposterior chest x-ray showing dorsolumbar scoliosis towards left.

pathological states that immobilize the fetal limbs at some period during or shortly after the formation of joints.⁴

AMC may present as myopathy or neuropathy, with associated hypotonia and wasted muscle mass. There is a decreased number of muscle fibres and in the transverse diameter of the individual fibres. Fibrous and fatty degeneration of the muscle, as well as fatty infiltration between muscle bundles, can be seen. However, even in most severe cases some normal muscle fibres can usually be found.^{1,2}

The most common finding in the central nervous system is degeneration and a reduction in the number and size of anterior horn cells. The diameter of the spinal cord is decreased, usually in the cervical and lumbar regions. Degeneration of the posterior horn cells has also been reported. Demyelination of the pyramidal tracts and motor roots and reduction of number of axons in the peripheral nerves have likewise been reported. There are abnormally high levels of creatine-phosphokinase (CPK) and 1-6-diphospho-fructoaldolase.^{1,2}

Problems arising during anaesthetic management include difficulty in tracheal intubation due to micrognathia and a short neck. These patients are prone to hypotension, respiratory depression, diminution of muscle tone and prolonged action of various inhalational and injected

anaesthetic drugs.⁵ Due to the associated myopathy and the skeletal deformities respiratory problems include alveolar hypoventilation, microatelectasis, a restrictive respiratory pattern, a decreased ability to cough and an increased incidence of aspiration. Presence of scoliosis, with a Cobb angle exceeding 70° leads to a significant reduction of lung volumes, an increased work of breathing, abnormal ventilation perfusion ratios and hypoxaemia, followed eventually by carbon dioxide retention, pulmonary hypertension and cor pulmonale. Usually the convex side of scoliotic chest makes greater contribution to the gas exchange and the lung on the side of concavity is compressed which if kept dependant, as in lateral position, can lead to further impairment of gas exchange. For this reason an anterior surgical approach was selected in this patient and he was kept supine during the procedure.

Administration of inhalational anaesthetics may further aggravate the hypoxia, due to the counteraction of the hypoxic pulmonary vasoconstriction and the added central depression of respiration, especially in spontaneously breathing patients.⁶ Ketamine provides a better choice as it is not associated with significant respiratory or cardiovascular depression, maintains muscle tone and helps maintain high PaO₂ values by keeping the shunt fraction low.⁷ Our patient maintained his PaO₂ and PaCO₂ values within the normal ranges during the ketamine infusion, while breathing spontaneously before intubation. Low-dose ketamine infusion also promotes early recovery, with a lower incidence of emergence phenomenon.⁸ Delayed recovery from anaesthesia in these patients may lead to retention of secretions, atelectasis and pneumonia.

A high incidence of malignant hyperpyrexia (MH) in these patients has been reported.³ Ketamine has been used safely in MH patients and a decrease in CPK levels has been observed during ketamine infusion.^{9,10} Tubocurarine can occasionally cause nonfatal hyperpyrexia in a susceptible patient, but in conjunction with inhalation agents it may aggravate an already established reaction.⁹ Some patients with myopathy exhibit increased sensitivity to curare and administration of even small dose may result in undesired prolonged paralysis.¹¹ Our choice of pancuronium for relaxation for the tracheal intubation and the surgical procedure was based on its reported safe use in MH⁹ and its cardiovascular stimulatory action.¹²

We conclude that low-dose ketamine (10–15 µg·kg⁻¹·min⁻¹) plus pancuronium constituted a safe anaesthetic in a patient with AMC. Ketamine produces analgesia even at this low dose and, if required, a high FiO₂ can be administered, without compromising the quality of the anaesthetic. Early recovery from anaesthesia is promoted.

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Résumé

La conduite anesthésique d'un mâle âgé de 11 ans atteint d'arthrogrypose congénitale (AMC) et myopathie est décrite. L'enfant s'est présenté avec une obstruction rénale lithiasique. Une pyélotomie par une incision antérieure et transverse sous costale et extrapéritonéale fut faite. A cause de l'hypoplasie musculaire, la scoliose sévère et la grande sensibilité aux différents sédatifs et agents anesthésiques d'inhalation, de petites doses de kétamine (10 à 15 µg·kg⁻¹·min⁻¹) ont été administrées par perfusion continue avec succès.