Anaesthesia for a patient with Friedreich's ataxia

INTRODUCTION

Friedreich's ataxia is an autosomal-recessive, neurodegenerative disease that primarily affects the nervous system and heart with an incidence of 1 in 50,000 individuals.[1] Initial symptoms of Friedreich’s ataxia usually appear in the second decade of life.

The condition is characterised by progressive gait and limb ataxia, dysarthria, lower-limb areflexia, decreased position and vibration sense, muscular weakness in the legs, and a positive extensor plantar response, as well as non-neurological signs including hypertrophic cardiomyopathy and diabetes mellitus.[2]

This is a case report of a 42-year-old man with Friedreich’s ataxia, who underwent wide local excision for squamous cell carcinoma of the scrotum. Though few Friedreich’s ataxia patients undergoing general anaesthesia and central neuraxial blockade are reported in the literature, the concerns of increased sensitivity to muscle relaxants in the former and fear of worsening of neurological symptoms in the later, has made this patient to be anaesthetised under scrotal field block with general anaesthesia using laryngeal mask airway (LMA) and spontaneous ventilation.

CASE REPORT

A 42-year-old man with chronic non-healing ulcer over the scrotum, diagnosed as a case of squamous cell carcinoma, was planned for wide local excision under anaesthesia. He was suffering from gait disturbances since 10 years of age. He had tremors of hands and slurring of speech for the past 5 years. He was having palpitations on and off. He is a diabetic, on treatment with oral hypoglycaemic agents since 3 years. On examination, cardiorespiratory system was clinically normal. He had pes cavus (arched foot) and hammer toes (flexion deformity of proximal interphalangeal joint with hyperextension of metatarsophalangeal and distal interphalangeal joints). He had scanning dysarthria (jerky, explosive, slurred and loud speech with separated syllables), horizontal nystagmus, generalised areflexia, weakness of skeletal muscles (power 3/5), absent
plantar reflex and impaired position and vibration senses. Euglycaemia was achieved with insulin. Echocardiography revealed mild left ventricular hypertrophy. With all these features, he was diagnosed to have Friedreich’s ataxia.

After explaining about the surgical procedure, anaesthesia and the risks involved, a written informed consent was obtained. Patient was planned for scrotal field block with general anaesthesia using LMA and spontaneous ventilation. After overnight fasting, morning dose of insulin was skipped. Fasting blood sugar was 116 mg% and urine ketones were negative. Premedication was done with tab. diazepam 5 mg, inj. glycopyrrolate 5 μg/kg intramuscularly, inj. ranitidine 50 mg, and inj. ondansetron 4 mg intravenously (i.v.) 1 hr prior to the surgery. electrocardiogram (ECG), pulseoximetry, non invasive blood pressure (NIBP), and capnography were monitored. After preoxygenation with 100% O₂ for 5 min, size 4 LMA was inserted, using fentanyl 2 μg/kg, and propofol 2.5 mg/kg i.v. Air entry was adequate and the position of the LMA was further confirmed by capnography.

Anaesthesia was maintained using O₂, air and isoflurane. Scrotal field block was given using 7 ml of lignocaine 1% and 13 ml of bupivacaine 0.25% without exceeding the toxic dose. Intraoperative period was uneventful. At the end of the surgery, patient was awakened and LMA was extubated. After being observed in recovery unit for an hour, he was shifted to post anaesthesia care unit for a day. He was shifted to the ward and later got discharged to home after a week.

**DISCUSSION**

Friedreich’s ataxia is a rare, recessively inherited, spinocerebellar degenerative disorder associated with hypertrophic cardiomyopathy and diabetes mellitus. This disease is steadily progressive and many patients survive more than 20 years after the onset of symptoms. Death is usually due to arrhythmias or congestive cardiac failure. The primary pathology is demyelination and degeneration of posterior columns, corticospinal, ventral and lateral spinocerebellar tracts and peripheral nerves. Frataxin is a mitochondrial protein found in the brain, heart and pancreas, which is involved in iron homeostasis and is an antioxidant. Mutation in the gene for frataxin results in the expansion of trinucleotide GAA (guanine, adenine, adenine trinucleotide) repeats in the intron 1. Reduced frataxin production leads to mitochondrial iron accumulation with impaired mitochondrial respiratory function especially in the nerve and muscle cells. The main anaesthetic implications of Friedreich’s ataxia are due to heart disease, restrictive lung disease (in patients with kyphoscoliosis), diabetes and neuromuscular degeneration. Patients with similar disorders such as amyotrophic lateral sclerosis are vulnerable to hyperkalemia after succinylcholine administration and are reported to be sensitive to non-depolarising muscle relaxants. Central neuraxial blockade has been safely used as an anaesthetic technique in patients with Friedreich’s ataxia for various procedures as per the literature available. However, it was not chosen as the technique of choice in this patient fearing worsening of neurological symptoms and medicolegal issues. Similarly, general anaesthesia using various non-depolarising muscle relaxants and volatile anaesthetics were used with normal or near normal responses, though earlier reports showed sensitivity to some of these agents. Levent et al. has used general anaesthesia without muscle relaxants for a Friedreich’s ataxia patient undergoing total hip replacement.

As the procedure did not necessitate muscle relaxation, general anaesthesia with spontaneous ventilation was chosen as the technique of choice. LMA was used instead of endotracheal tube to scale down the stress response and its deleterious effects on the myocardium. scrotal field block was given for pain relief. Maintenance of anaesthesia with isoflurane gave cardiac stability.

**CONCLUSION**

When the procedure does not warrant, it is better to avoid central neuraxial block and muscle relaxants in these group of patients, as this would simply prevent many potential complications.

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