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CASE REPORT

Anaesthesia for a patient with Friedreich's ataxia undergoing emergency tibia interlocking nail insertion

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ABSTRACT

Although being rare, Friedreich's ataxia is the most common inherited ataxia. Friedreich's ataxia causes muscle weakness, dysarthria, scoliosis and cardiac symptoms which can cause even death. These are challenging to the anaesthetist and even gets worse with the progression of the disease. Patients are sensitive to non-depolarising muscle relaxants and even regional anesthesia can be difficult due to scoliosis with also medicolegal risk. This is a case report of a 36-year-old wheel-chaired man with Friedreich's ataxia, who underwent a right interlocking nail tibia insertion for tibia fracture. General anaesthesia with laryngeal mask airway without muscle relaxant was used and SIMV mode was the ventilation mode of choice. The patient recovery was smooth with no pain and he was transferred to the ward after 1 h in the recovery room.

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Ataxia; Friedreich's; weakness; cardiac; scoliosis

1. Introduction

Friedreich's ataxia is an autosomal recessive neurodegenerative disease that was first described by Nicholaus Friedreich since 1863. Friedreich described the condition in nine members of three families. His initial reports noted that the age of onset was around puberty with a clinical picture of ataxia, dysarthria, sensory loss, muscle weakness, scoliosis, foot deformity, and cardiac symptoms. The absence of deep tendon reflexes was first described by Friedreich's student, Erb, in 1875 [1,2].

Friedreich's ataxia is a disorder that affects a gene (FXN) on chromosome 9, which produces an important protein (frataxin). Low frataxin levels lead to insufficient biosynthesis of iron–sulfur clusters that are needed for mitochondrial electron transport and iron metabolism. This leads to cell damage and degeneration. Degeneration occurs in sensory nerves more than motor nerves. Similar degenerative changes occur in cardiac cells and pancreatic cells causing left ventricular hypertrophy and dilatation and diabetes mellitus. Friedreich's ataxia is the most common inherited ataxia with a prevalence of 1 in 30,000–50,000 and a carrier frequency of 1 in 90–110. The classic Friedreich's ataxia phenotype is due to a homozygous GAA (guanine, adenine, adenine) triplet repeat expansion in intron 1 of the frataxin gene [3–6].

This is a case report of a 36-year-old wheel-chaired man with Friedreich's ataxia, who underwent a right interlocking nail tibia insertion for tibia fracture.

2. Case report

A 36-year-old wheel-chaired man with a weight of 70 kg and a height of 173 cm was planned for a right interlocking nail tibia insertion for tibia fracture. The patient was suffering from gait disturbances, lower limb weakness, mild scoliosis and mild cavovarus foot deformity. He had slurring of speech for the past 5 years. He is not a diabetic. On examination, cardiorespiratory system was clinically normal. He had dysarthria, nystagmus, generalised areflexia, weakness of skeletal muscles more in the lower limbs, absent plantar reflex and impaired position and vibration senses. Echocardiography revealed mild left ventricular hypertrophy with preserved ejection fraction. ECG showed frequent atrial premature beats. The patient has a history of ESWL 3 years ago for a ureteric stone under sedation.

A written informed consent was signed after discussing the plan of anaesthesia and the expected risks. Patient was planned for right interlocking nail tibia insertion with general anaesthesia using laryngeal mask airway (LMA).

Premedication was done with midazolam 2 mg prior to the surgery. Basic monitors were applied including pulse oximetry, electrocardiogram (ECG), capnography and non-invasive blood pressure (NIBP). After preoxygenation with 100% O_2 for 5 min, fentanyl 3 µg/kg, propofol 2.5 mg/kg and xylocaine 1 mg/kg were given i.v., xylocaine 10% spray for the airway, LMA size 4 was inserted. Air entry was adequate and the position of the LMA was further confirmed by capnography. Volume-controlled synchronized intermittent mandatory ventilation (SIMV) was used with a tidal volume of 500 and frequency of 12.

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Anaesthesia was maintained using O_2, air, isoflurane 2% and propofol infusion of 0.05 mg/kg/minute. Local infiltration of skin incision sites was done using 10 ml of lignocaine 2% and 10 ml of bupivacaine 0.5% 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 the recovery unit for an hour, he was shifted to the ward later got discharged to home after a week.

3. Discussion

Friedreich's ataxia is a rare progressive neuro-degenerative disease that implies many risks for patients undergoing surgical interventions under anaesthesia. These patients may show sensitivity to nondepolarizing muscle relaxants and succinylcholine administration may carry the risk of hyperkalemia [5–7].

Neuraxial and regional anaesthesia have been used in several cases with Friedreich's ataxia for different procedures and showed no reported side effects but on the other side the fear from medico-legal responsibility and the probability of worsening of neurological manifestations would limit their use [8–10].

To avoid the previous probable risks, muscle relaxants were not used at all either in the induction or maintenance with the use propofol infusion to augment the depth of anaesthesia. LMA was used to decrease the stress of airway manipulation although intubation was used safely without muscle relaxation in previous case reports. SIMV was used instead of spontaneous respiration to guard against carbon dioxide retention due to hypoventilation under deep anaesthesia. [10,11,12,13]

Friedreich's ataxia has many effects on various systems of the body especially the nervous system and the heart that makes delivery of anaesthesia such a critical situation that should be dealt with caution.

Disclosure statement

No potential conflict of interest was reported by the author(s).

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References

[1] Friedreich N. Uber degenerative Atrophie der spinalen Hinterstrange. Virchow's Arch Pathol Anat. 1863;26:391–419.
[2] Harding AE. Friedreich's ataxia: a clinical and genetic study of 90 families with an analysis of early diagnostic criteria and intrafamilial clustering of clinical features. Brain. 1981;104(3):589–620.
[3] Klockgether T. Update on degenerative ataxias. Curr Opin Neurol. 2011;24(4):339–345.
[4] Koeppen AH. Friedreich's ataxia: pathology, pathogenesis, and molecular genetics. J Neurol Sci. 2011;303(1–2):1–12.
[5] Taroni F, DiDonato S. Pathways to motor incoordination: the inherited ataxias. Nat Rev Neurosci. 2004;5(8):641–655.
[6] Sahdeo S, Scott BD, McMackin MZ, et al. Dyclonine rescues frataxin deficiency in animal models and buccal cells of patients with Friedreich's ataxia. Hum Mol Genet. 2014 Dec;23(25):6848–6862.
[7] Friedman LS, Farmer JM, Perlman S, et al. Measuring the rate of progression in Friedreich ataxia: implications for clinical trial design. Mov Disord. 2010;25(4):426–432.
[8] Schmitt HJ, Wick S, Münster T. Rocuronium for muscle relaxation in two children with Friedreich's ataxia. Br J Anaesth. 2004;92(4):592–596.
[9] Bell CF, Kelly JM, Jones RS. Anaesthesia for Friedreich's ataxia. Case report and review of the literature. Anaesthesia. 1986;41(3):296–301.
[10] Hanusch P, Heyn J, Well H, et al. Peridural anaesthesia with ropivacaine for a patient with Friedreich's ataxia. Caesarean section after dorsal stabilisation of the spinal column (Th5-L1. Anaesthesist. 2009;58:691–694.
[11] Wyatt S, Brighouse D. Anaesthetic management of vaginal delivery in a woman with Friedreich's ataxia complicated by cardiomyopathy and scoliosis. Int J Obstet Anesth. 1998;7(3):185–188.
[12] Ganesan I. Anaesthesia for a patient with Friedreich's ataxia. Indian J Anaesth. 2011;55(4):418–420.
[13] Levent K, Yavuz G, Kamil T. Anaesthesia for Friedreich's ataxia. Case report. Minerva Anestesiol. 2000;66(9):657–660.