CASE REPORT

Anaesthetic management of spinal muscular atrophy in a patient with pneumothorax: a case report

Madeeha Rasheed[1], Khalid Ahsan[2]

Abstract

One of the most prevalent hereditary neuromuscular disorders is spinal muscular atrophy (SMA). Progressive muscular weakness and an irreversible loss of alpha motor neurons in the spinal cord are the hallmarks of SMA and are associated with increased sensitivity to opioids and muscle relaxants.

SMA is classified clinically into four categories according to its severity and age of onset, which poses a challenge for the anaesthesiologist. Although general anaesthesia can be given, it is preferable to avoid muscle relaxation.

We are reporting the anaesthetic management of a patient with type 3 SMA, diagnosed with a case of bullous lung disease. The procedure planned was thoracoscopy and pleurodesis/pleurectomy under general anaesthesia at the Aga Khan University Hospital, Karachi, in August 2022. This case report highlights the importance of individualised care in SMA patients, emphasising the need for careful planning and consideration of patient-specific factors.

Keywords: Spinal Muscular Atrophy, Anaesthesia, Thoracoscopy, Pleurodesis.

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Introduction

Alpha motor neurons in the anterior horn cells of the spinal cord degenerate to cause spinal muscular atrophy (SMA), an autosomal recessive condition.

It was initially recorded by Johan Hoffmann and Guido Werdnig in 1893 and 1891, respectively.[1] It occurs due to SMN1 (Survival Motor Neuron) gene depletion. The patient discussed in this report was suffering from recurrent pneumothorax. He was diagnosed with bullous lung disease.

[1]Aga Khan University Hospital, Karachi, Pakistan; [2]Department of Anaesthesiology, Aga Khan University Hospital, Karachi, Pakistan.
Correspondence: Madeeha Rasheed.
Email: madeeha_rasheed@hotmail.com
ORCID ID: 0009-0007-8934-0202

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SMA is one of the most common hereditary causes of infant mortality, affecting about 1 in 10,000 live births globally, Studies conducted in different regions have shown differences in the prevalence and clinical presentation of SMA, with certain groups showing higher frequencies of genetic subtypes. For instance, SMA type I, the most severe form, may be more common in some cultures, whereas milder versions like SMA type III may be more common in others.[2]

Case Report

A 19-year-old male patient presented with shortness of breath and chest pain in August 2022 at the Aga Khan University Hospital, Karachi, with a history of recurrent pneumothorax and chest drain insertion.

The patient had a history of progressive lower limb weakness, including difficulty in walking and climbing stairs, gradually increasing from childhood.

The preoperative assessment showed no respiratory or cardiac function abnormalities. Airway examination and other baseline lab parameters were normal. Chest X-ray showed right-sided pneumothorax. Preoperative investigations showed deletion of the SMN1 gene exons 7 and 8 which is consistent with Spinal Muscular Atrophy.

The procedure planned was video-assisted thoracoscopy, pleurodesis, and pleurectomy under general anaesthesia. Informed consent was taken, and ASA (American Society of Anaesthesiologists) standard monitoring[3,4] was applied. Injection of Glycopyrrolate 0.2mg was given as premedication.

Induction was done with Propofol, a loading dose of 1.5mg/kg, and infusion started at 3mg/kg/h. Intermittent boluses of Propofol 50mg + 50mg were given at the time of intubation. Along with this, Dexmedetomidine 1mcg/kg over 10 minutes and infusion continued at 0.7 to 1mcg/kg/h. Throughout the induction phase, the patient remained haemodynamically stable.

A left-sided double-lumen tube (size 37) was inserted using a McGrath laryngoscope and fixed at 29cm after confirmation with auscultation.

Figure-1: Ventilatory parameters
Image showing ventilatory parameters during the procedure, 50% FIo2 and no spontaneous effort on controlled mode of ventilation, maintaining anaesthetic depth via TIVA (avoiding muscle relaxants)

Before placing the patient in the left lateral position, another bolus of Propofol was given. Intraoperative anaesthesia was maintained with Total Intravenous Anaesthesia (TIVA), using Propofol and Dexmedetomidine, avoiding muscle relaxants. Volume-controlled mode of ventilation (hyperventilated) was started with an air oxygen mixture (FIO2 0.6-1.0). Figure 1 illustrates the use of TIVA technique to maintain the depth of anaesthesia and intraoperatively stabilize the ventilatory parameters of the patient in control mode, thereby eliminating the need for any kind of muscle relaxant.

Before incision, local anaesthetic (Lidocaine 2%) was injected at the site of the incision. Right sided posterolateral thoracotomy was performed. The surgery was uneventful without any need for muscle relaxation and the patient remained haemodynamically stable throughout the procedure.

At the end of the procedure, the wound was closed, and local anaesthetic (Bupivacaine 0.25%) was infiltrated at the incision site and chest tube site. The patient was extubated uneventfully and shifted to the recovery room with oxygen supplementation. Postoperatively, there was an improvement in symptoms and resolution of pneumothorax. Follow-up of the patient was done till discharge from the hospital.

Discussion

Neuromuscular diseases (NMDs) are considered a clinically and genetically diverse group of diseases that are constituted by dystrophic alterations in the muscle and a decline of muscle strength. Pre-junctional disorders, junctional disorders, and post-junctional disorders are the three categories into which they can be separated. A particular kind of pre-junctional disease is categorised as spinal muscular atrophy.

A null mutation in the SMN1 gene results in the hereditary condition known as spinal muscular atrophy (SMA). Lower α-motoneurons in the anterior horn of the spinal cord irreversibly die due to mutation, while the loss of SMN1 results in low levels of Survival of Motor Neuron (SMN1), a protein essential for neuromuscular development.[5]

Muscle weakening that is gradual and symmetrical is its defining feature. Clinical severity in SMA varies from the most severe form, which manifests at birth, to the mildest form, which manifests in adulthood. SMA affects voluntary muscles, such as those in the shoulders, hips, thighs, and upper back. Deep tendon reflexes are diminished, and the lower limbs appear to be affected more than the upper limbs. Bulbar muscle involvement can also occur. The risk of recurrent chest infections and aspiration pneumonia is also present. Extra neural manifestations also include heart defects.[6-8]

The age of onset and the patient's maximum motor ability are used to classify SMA patients clinically. It is classified as: severe, type I, intermediate, type II, mild, type III, and extremely mild conditions which include adult-onset, type IV. (Table-1)[6-8]

This patient was diagnosed with SMA type III; about 30%

Table-1: Types of Spinal Muscular Atrophy.

Types	Characteristics	Clinical Features
Type 0	Decrease in foetal movements during pregnancy	Lack of reaction to stimuli, facial diplegia and congenital heart defects
Type 1	Infantile onset, Werdnig-Hoffmann disease, onset at birth or by the age of 6 months	Generalised muscle weakness, weak cry and breathing distress. Risk of aspiration and failure to thrive
Type 2	Intermediate onset or Dubowitz disease, onset-3-15 months	Predominantly proximal Muscle weakness. Lower limbs are more involved
Type 3 and Type 4	Adult onset Spinal Muscular Atrophy or Kugelberg Welander disease, onset-18months or adulthood	Inability to walk and stand, develop foot deformities, scoliosis and respiratory muscle weakness

of instances of SMA are categorised as SMA type III, commonly referred to as juvenile SMA or Kugelberg-Welander illness (Kugelberg & Welander, 1956).[1] This is a mild form that appears after the age of 18 months and children who are affected can walk on their own, but over time, their walking skill declines. Type III children and adults rarely have respiratory problems because they have normal lung function and respiratory muscle power, apart from during acute illness. Scoliosis, fatigue, and muscle and joint pain with activity are also common.

Research suggests that patients with neuromuscular illness should not be given Suxamethonium or Succinylcholine, because of the potential for hyperkalaemia.[9] Non-depolarising neuromuscular blockers appear to have varying and, perhaps, protracted sensitivities.

Local reports from healthcare institutions further underscore the importance of tailored anaesthetic approaches for SMA patients undergoing surgical procedures.

Limitations include the single-case nature of the report and the need for further research.

In a recent case report, a two-year-old child with SMA type 1 was scheduled for fundoplication and gastrostomy under general anaesthesia. Even though rapid sequence induction was ideal for this procedure, Suxamethonium and opioids were avoided. Neuromuscular blockade was subsequently maintained with small doses of Cis-atracurium according to the train-of-four stimulus and ultrasound-guided bilateral rectus sheath block was provided for analgesia during surgery.[10] However, in the present case the use of muscle relaxants was completely avoided.

In another case report, Brown AF et al. mentioned giving general anaesthesia with Propofol and Remifentanil without the use of muscle relaxants.[11] Yasemin et al. also used a similar method for vertebral implant removal in a type 2 SMA patient.[12]

Conclusion

The nature of procedure necessitates a sufficient depth of anaesthesia and maintenance of airway with double lumen tube for one lung ventilation along with avoidance of muscle relaxation. This case was difficult to manage, hence, the use of TIVA with propofol in conjunction with dexmedetomidine was very helpful in maintaining adequate depth of anaesthesia. Therefore, Anaesthesiologists and surgeons must work together in a multidisciplinary manner to maximise perioperative care and reduce the risk of complications.

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MR: Performed anaesthesia of the case, consent, writing, review and final approval.
KA: Performed the anaesthesia of the case, editing and final approval.