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Anesthetic Management of Scoliosis Correction in Mainzer-Saldino Syndrome: A Case Report

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Abstract

Introduction: Mainzer-Saldino Syndrome is a rare autosomal recessive disease with mutations in genes that encode components involved in ciliary transport. This syndrome is characterized by chronic renal failure, severe retinal dystrophy and skeletal abnormalities including spinal and chest wall deformity resulting in severe respiratory failure.

Case presentation: We report the first successful anesthetic management of a 26-year-old man with Mainzer-Saldino Syndrome who underwent scoliosis deformity surgical correction. The severity of respiratory problems and renal dysfunction that characterize this syndrome require a multidisciplinary preoperative assessment and careful planning of intraoperative management, also in relation to surgical complications.

Conclusions: A careful preoperative assessment is essential for the correct anesthetic management. General anesthesia was safely administered; however a constant modulation of controlled ventilation is required to avoid barotrauma and an invasive hemodynamic monitoring is critical to allow adequate fluid management. Lastly, intraoperative dialysis can be planned in long-lasting surgery with a significant loss and replacement of fluids.

Keywords: Mainzer-Saldino Disease; Ciliopathy; Anaesthesia

Introduction

Mainzer-Saldino Syndrome (MZSDS) is a rare genetic disorder that was first described in 1970 by Mainzer and Saldino, characterized by chronic renal failure, severe retinal dystrophy and skeletal abnormalities [1].

A small number of individuals with this disorder have additional problems affecting other organs. These can include liver disease resulting in hepatic fibrosis, cerebellar ataxia, and mild intellectual disability.

It is a type of ciliopathy caused by altered cilia assembly, maintenance, or function with marked variability of clinical expression [2,3].

We present the successful management of one case in a 26-year-old man posted for a scoliosis deformity correction.

Considering the insufficient data of such cases, we take this opportunity as a first description for a successful anesthetic management. Written informed consent was obtained from patient for this report.

Case Report

A 26-year-old male (weight 47.5 kg; height 152 cm) was proposed for a correction of a severe scoliosis deformity with a Cobb's angle curvature of 84° (Fig. 1A). The patient underwent routine preoperative evaluation which included a complete physical and medical history, a 12 lead ECG and routine blood tests. He had a Class III Mallampati airway. Because of his scoliosis, pulmonary and cardiologic examinations were requested. Spirometry showed a severe restrictive pattern with a forced expiratory volume in 1 s and a forced vital capacity corresponding to 36% and 41% of the predicted values, respectively. In addition, the single-breath diffusion lung capacity for carbon monoxide showed a reduced diffusion capacity of 37% of predicted value. The cardiological examination showed a good hemodynamic compensation. Transthoracic echocardiography revealed a right atrium and ventricular sizes at upper limits of normal, minimal tricuspid regurgitation with a pulmonary artery systolic pressure of 43 mmHg and left ventricular ejection fraction of 60%. The preoperative blood pressure and heart rate were normal. Blood tests revealed a serum creatinine of 8.27 mg.dl-1. Considering his history of chronic renal failure and intermittent dialysis, an intraoperative hemodialysis was planned.

In the preoperative room, ECG and pulse oximetry were established and an arterial catheter was inserted into the radial artery under local anesthesia.

A FloTrac catheter (Edwards Lifesciences, Irvine, CA, USA) was connected to the intrarterial access and a EV1000/Volume View (Edwards Lifesciences, Irvine, CA, USA) for continuous arterial pressure, cardiac index (CI), stroke volume index, and stroke volume variation (SVV) monitoring.

Anesthesia induction was performed with fentanyl 100 mcg, propofol 80 mg and cisatracurium 12 mg. Laryngoscopy and tracheal intubation were made with the aid of a video laryngoscope with no problems.

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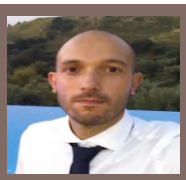

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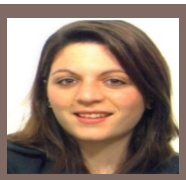

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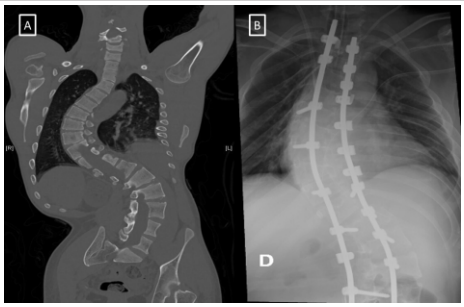

Figure 1: (A) Preoperative computed tomography scan showing scoliosis. (B) Post-operative study demonstrating marked improvement in degree of scoliosis.

Intraoperative maintenance of anesthesia included total intravenous anesthesia (TIVA) with a combination of propofol and remifentanil.

A dual-lumen right internal jugular dialysis catheter and a dual-lumen left central venous catheter were inserted with an ultrasound guide. A chest radiograph was performed after catheters placement.

A Bispectral index (BIS) monitor was used to monitor the depth of anesthesia. The BIS value was mantained between 40 and 45.

Cisatracurium bolus rate was titrated under the guidance of neuromuscular transmission monitoring.

Temperature monitoring was established with a single-use sensor placed on the patient's forehead. A warming system was positioned to ensure uniform convective warming.

The patient was placed in a prone position and attention has been paid to pressure areas. Tidal volume was mantained between 6-7 ml.kg-1 of predicted body weight with a plateau pressure at less than 28 cmH2O.

The intraoperative fluid and vasopressor/inotrope administration were guided by the EV1000/Volume View parameters. The goal-directed fluid therapy was put in place to mantain SVV < 15%, CI > 3,0 l.min-1.m2-1 and central venous pressure < 5 cmH2O. Hemogasanalysis was performed every hour. He was transfused with five units packed red blood cell and five units fresh frozen plasma. A total of 2250 ml of normal saline solution was administered during surgery.

The patient underwent dialysis for a total of 2 hours and 45 minutes. Urine production was absent and fluid was removed during hemodialysis.

The surgery proceeded uneventfully for 11 hours and 30 minutes. The surgical approach was a posterolateral arthrodesis from D2 to L5 with intraoperative neurophysiologic monitoring (Fig. 1 B). At the end of the surgery, the patient was transferred to the ICU.

A good analgesic plan with opioids was managed. After an adequate weaning, the trachea was extubated the next day. Postoperative period was uneventful, vital signs and mental status were stable.

Discussion

MZSDS is a rare autosomal recessive disorder characterized by mutations in genes that encode components of the intraflagellar transport complex A, which drives retrograde ciliary transport [4]. These genetic mutations cause abnormal function or formation of cell cilia that characterize a group of diseases called ciliopathies. These include a wide range of phenotypes resulting from developmental or functional defects of unique or multiple systems. The main features of ciliary disease are: retinal dystrophy, renal disease and cerebral anomalies. Additional manifestations include hepatic/pancreas fibrosis, diabetes, and skeletal dysplasia. Syndromic ciliopathies that affect bone development are classified as skeletal ciliopathies [2,3]. Primary clinical features of MZSDS are phalangeal cone-shaped epiphyses, renal disease, severe retinal dystrophy, and hip abnormality [4]. In addition, the patient had severe scoliosis which resulted in restrictive respiratory failure. Pulmonary dysfunction due to chest deformity can be present in ciliopathies, especially in the Jeune syndrome, also called asphyxiating thoracic dystrophy [5,6]. These defects cause variations in normal physiology and can have anesthetic implications in the management of ciliopathies (Table 1).

For these reasons, our patient with MZSDS require a careful preoperative assessment.

Pulmonary function should be assessed through a physical examination, radiological investigation, and blood gas analysis. The presence of thoracic malformation could lead to lung restriction, so spirometry may be needed. In this case, a narrow chest associated with scoliosis resulted in severe restrictive ventilatory failure. During anesthesia, positive pressure ventilation could result in barotrauma or pneumothorax if peak airway pressure is not maintained as low as possible. Additionally, increased thoracic pressure can decrease venous return and diminish cardiac output [5].

Pressure-controlled ventilation has been suggested in Jeune syndrome patient [6].

In neurosurgery, the surgeon compresses and modifies the spine determining volume and pressure airway variations. Hence close collaboration is needed between the surgeon and the anesthesiologist. Based on the surgical stages and blood gas analysis, we alternated a volume-controlled and pressure-controlled ventilation, maintaining a tidal volume of 6-7 ml.kg-1, a plateau pressure at less than 28 cmH2O and a low peak airway pressure.

So, MZSDS patient ventilation settings and regular blood gas analysis follow up are very important.

In addition, a careful airway assessment should be done preoperatively as facial bone deformities are present in ciliopathies.

Due to the limited literary knowledge of MZSDS and the pulmonary function anomalies described above, a preoperative cardiological evaluation with echocardiography would exclude any unknown cardiac malformations as well as right heart hypertrophy and pulmonary vascular hypertension. The latter two findings are typically present in patient with scoliosis.

Renal involvement has been noted in cilia disease. Progressive renal dysfunction results from a ciliary defect in the nephrons leading to end-stage renal disease. In this case, the patient reported a chronic renal failure with history of periodic dialysis treatments. Surgical arthrodesis from D2 to L5 leads to a significant intraoperative fluids loss and their replacement may be inappropriate, resulting in overload. For this reason and due to the moderate loss of fluids during this surgery, invasive hemodynamic monitoring is necessary to allow adequate fluid management. In this way, hemodynamics is optimized and the overload or deficit of circulating fluids is avoided. Furthermore, . in agreement with the nephrologist, we planned an intraoperative dialysis with a serial control of serum electrolytes.

Liver fibrosis may be present in MZSDS, but in our patient there were no clinical or laboratory findings of liver dysfunction.

In literature, which anesthetic drugs can safely be used in MZSDS is not described. However, in other ciliopathies such as Jeune syndrome, it is reported that non-inhaled anesthetics, inhaled anesthetics, and muscle relaxant able to be used safely [5,6].

The complications of this type of surgery with the greatest impact are nerve injuries. For this reason, intraoperative strategies have

been designed to reduce this risk. Somatosensory and corticomotor evoked potentials, that reflect the sensory and motor pathway functions respectively, were planned [7]. However, anesthetic agents and physiologic perturbations may interfere with somatosensory and corticomotor evoked potentials: hypothermia, hypotension, hypercapnia, halogen, and muscle blocking medication may interfere with the signal transmission [8]. Consequently, general anesthesia with proper depth control and without neuromuscular relaxation is a key for ideal management. The most highly recommended technique is TIVA avoiding neuro-muscular blocking during the anesthesia maintenance phase [9]. BIS helps to prevent the consequences of the anesthetic effect that could influence neurological monitoring directly and indirectly causing dynamic instability. It also helps to prevent the risk of intraoperative movements and awareness. Hypothermia increases bleeding in column surgery [10]. In this case, the body temperature was always under surveillance.

Finally, we believe that postoperative recovery in the ICU is useful for regular monitoring organ functions.

Conclusion:

a multidisciplinary team working is needed to perform a careful preoperative assessment and intraoperative management.

A detailed anesthetic plan is required in the perioperative period in MZSDS patient because the disorder can involve different organs. Several points must be considered including lung and renal function. General anesthesia was safely administered in our patient and controlled ventilation requires constant modulation by the anesthesiologist to avoid barotrauma. Intraoperative dialysis can be planned in long-lasting surgery with a significant loss and replacement of fluids.

Ciliopathic Syndrome	Features	Anesthetic implications
Bardet-Biedl Syndrome	Obesity	Difficult airway management
	Renal anomalies	Hemodynamic instability possible
	Left ventricular hypertrophy	Avoid nephrotoxic drugs and fluid overload
	Congenital heart disease	Administer low doses of drugs with liver metabolism
	Hepatic fibrosis	
	High arched palate/dental crowding/small teeth	
Jeune Syndrome	Chest wall deformity	Avoid or use with caution drugs with liver metabolism if liver disease is present or if transaminases are increased.
	Pulmonary hypoplasia	Avoid high peak pressure during mechanical ventilation. High thoracic pressure can decrease venous return and cardiac output, barotrauma and pneumothorax.
	Pulmonary hypertension	
	Fibrocystic liver disease	
	Pancreatic insufficiency	
	Renal anomalies	
Nephronophthisis	Renal anomalies	Avoid nephrotoxic drugs and fluid overload
	Arterial hypertension	
Joubert Syndrome	Cleft palate, large tongue, small jaw, and laryngomalacia	Difficult airway management
	Abnormal breathing pattern (hyperpnea/apnea)	Use opioids with caution
	Hepatic fibrosis	Avoid or use with caution drugs with liver metabolism if liver disease is present or if transaminases are increased
Meckel-Gruber Syndrome	Abnormality of larynx/tongue/cervical vertebrae	Difficult airway management
	Polycystic kidneys	Avoid nephrotoxic drugs and fluid overload
Oral-Facial-Digital Syndrome	High arched palate, poly-lobed tongue, abnormalities of the epiglottis and larynx	Difficult airway management
	Congenital heart disease	
McKusick-Kaufman Syndrome	Congenital heart disease	Hemodynamic instability possible
	Chronic renal failure	Avoid nephrotoxic drugs and fluid overload
Alstrom Syndrome	Obesity	Difficult airway management
	Scoliosis	Avoid high peak pressure during mechanical ventilation. High thoracic pressure can decrease venous return and cardiac output, barotrauma and pneumothorax.
	Restrictive lung disease	Avoid nephrotoxic drugs and fluid overload
	Chronic renal failure	Hemodynamic instability possible
	Dilated cardiomyopathy	Avoid or use with caution drugs with liver metabolism if liver disease is present or if transaminases are increased
	Hepatic dysfunction	

Table 1: Anesthetic concerns related to main diagnostic features of ciliopathies.

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