Case report

Anesthetic Management of a 9-Years-Old Child Affected by Al-Raqad Syndrome Scheduled for Cataract Surgery: A Case Report

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Abstract

Al-Raqad syndrome (ARS) is a novel and extremely rare autosomal recessive disorder. This syndrome affects many organs, mainly the central nervous and musculoskeletal systems. Al-Raqad syndrome's manifestations include neurodevelopmental delays and a characteristic phenotype including craniofacial anomalies. This disorder, which is the result of a mutation in the decapping enzyme, scavenger (DCPS) gene, was first described in 2015. We presented a 9-year-old child affected with this syndrome, who suffers from severe neurodevelopmental delays, scheduled for cataract surgery. The anesthesia management focused on neurodevelopmental defects and craniofacial and musculoskeletal abnormalities. The reports of this syndrome are scarce, and to our knowledge, the present case is the first report of anesthesia management of this syndrome.

Keywords: Al-Raqad syndrome, Anesthesia, Cataract

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Introduction

Al-Raqad syndrome (ARS) is an ultra-rare genetic disorder, which is inherited in an autosomal recessive pattern. The manifestations of this syndrome include developmental delay, intellectual disability, and craniofacial abnormalities (1). Other phenotypic implications include neuromuscular defects, skin dyschromia, cardiac septal defect, microcephaly, and skeletal anomalies (2).

ARS was first described in 2015 in a Jordanian family including three affected males, presenting with musculoskeletal disorders and facial anomalies arising from developmental delay (3). Homozygous or compound heterozygous mutations in the DCPS gene located on chromosome 11q24.2 result in this phenotype (3). Functional defects of DCPS have adverse effects on neural cell function (4). DCPS mutation also causes scavenger mRNA decapping enzyme loss and results in syndromic intellectual disability with neuromuscular defects (4).

The case reports of this syndrome are scarce, and to our knowledge, there is no case report describing the anesthetic management of this syndrome so far. Here we present a 9-year-old boy affected by ARS with severe neurodevelopmental delay scheduled for cataract surgery, emphasizing the anesthetic considerations.

Case Report

A 9-year-old boy weighing 10 kg was diagnosed with ARS and was scheduled for cataract surgery. He was affected by a homozygote variant of DCPS single nucleotide polymorphism. He was born in a second-degree consanguineous marriage and had a similarly affected younger sister. Genetic studies showed that both paternal and maternal grandparents were heterozygous for this gene.

The patient was suffering from severe neurodevelopmental disorders and could not stand up, walk or speak. The limbs were atrophic, and myopathy was obvious. He was microcephalic, had a short stature, and was affected by craniofacial abnormalities such as a high-arched palate and lack of tooth growth. Figure 1 shows the physical characteristics of the patient.

The brain MRI revealed dilation of all ventricular system and subarachnoid spaces and inflammatory changes at paranasal sinuses. The video electroencephalography study was not diagnostic of seizure disorder, but because of periods of weakness and lethargy, the patient was treated with Levetiracetam. Echocardiography showed normal left ventricular ejection fraction as well as mild mitral tricuspid regurgitation. No other significant finding was observed. Airway examination, including the open mouth, thyromental distance, head extension, and Mallampati score, were all normal. There were no spine deformities or scoliosis.

The primary anesthetic concerns included neurodevelopmental delay, musculoskeletal malformations, potential risk of malignant hyperthermia, residual respiratory depression after extubation, and probable altered response to muscle relaxants. Due to the similarities of this syndrome with other syndromes caused by neurodevelopmental delay, it was also reasonable to consider the risk of mitochondrial myopathy in the anesthesia management of the patient.

For anesthesia preparation, we first washed the breathing system with 100% high-flow O2 for 10 minutes. In the operating room, the child was monitored using electrocardiography (ECG), capnography (ETCO2), and pulse oximetry (SPO2). Also, the bispectral index (BIS), blood pressure (BP), and temperature (T) were constantly monitored. After injection of Atropine 0.03 mg/kg and Fentanyl 2 microgram/kg, anesthesia induction was performed

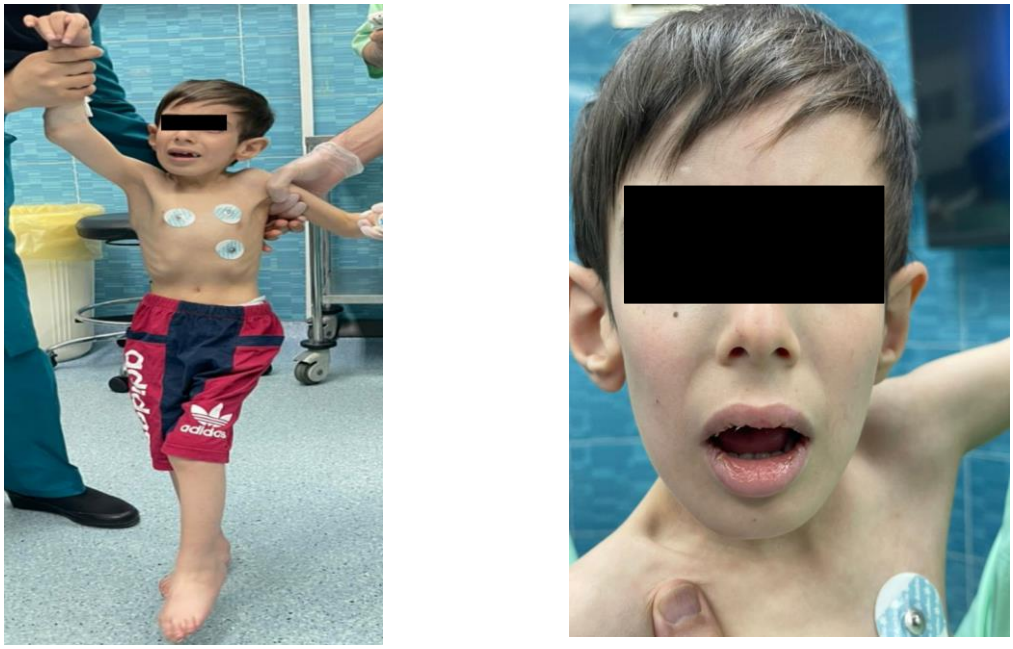

Figure 1. Physical characteristics of the patient with ARS.

using IV Propofol infusion. Besides, we use a Laryngeal Mask Airway (LMA) with a size of 2.5 when BIS reached 40. The child had no bucking and was ventilated with appropriate tidal volume and normal airway pressure. The maintenance dose of Propofol was infused based on the proportional-integral-derivative algorithm to maintain a BIS value of between 40 and 60. Propofol infusion was set according to the BIS level every 5 minutes in a way that if BIS was less than 40, Propofol infusion was decreased by 20%. Tetracaine eye drop combined with 0.25 mcg/kg/min of Remifentanil was used for analgesia. In the state of hypertension or tachycardia more than 20% of baseline, a bolus dose of 0.5 mcg/kg q25min was infused to treat hemodynamic changes. During the operation, capnography and temperature monitoring were performed, and both were stable. The procedure lasted for 1.5 hours. At the end of the surgery, all infusions were stopped and the child was awakened with his baseline condition. He was observed overnight in the pediatric intensive care unit and discharged one day later without any event.

Discussion

ARS is an ultra-rare congenital disorder inherited in an autosomal recessive pattern and is mainly associated with neurodevelopmental delay. This syndrome is caused by DCPS single nucleotide polymorphism, encoding scavenger mRNA decapping enzyme (1). The reports of this syndrome are scarce, and to our knowledge, there is no report about anesthesia management and considerations in ARS so far.

We reported this case to discuss which anesthesia management and considerations are best suited for these patients. We avoided neuromuscular relaxant drugs because of neuromuscular defects and the possibility of altered responses to this class of drugs. Also, malignant hyperthermia may potentially occur in these patients. In addition to the availability of Dantrolene and cooling systems, it is recommended to use cautious monitoring such as thermal monitoring and capnography to avoid this event. Besides, we suggest it is reasonable to avoid using depolarizing muscle relaxants, and if necessary, short-acting non-depolarizing relaxants should be used under careful monitoring. Rocuronium can be a suitable choice when Sugammadex is available. Also, great care must be taken in the prescribed dose and the drug's titration method. Our case was undergoing surgery in a center with facilities for postoperative ventilation and a pediatric intensive care unit. To prevent malignant hyperthermia, we had thermal monitoring and used capnography. Besides, we washed out the breathing system with high-flow oxygen for 10 minutes and extracted any vaporizers from the anesthesia machine. Also, we had a cooling system and Dantrolene available; however, we preferred to avoid neuromuscular paralysis. The LMA provided proper ventilation, so we did not need to intubate the patient.

To minimize the risk of Propofol Infusion Syndrome (PIS), we used a dual proportional-integral-derivative algorithm based on BIS monitoring for IV administration of Propofol, so we had a proper level of anesthesia without any movement (5). We also used Remifentanil infusion as short-acting analgesia with negligible respiratory depression effects. Therefore, the patient was awake at the end of the procedure with the same muscle tone as before, with spontaneous ventilation and normal saturation. Because of the accompanying neurological defects in ARS, it is reasonable to avoid drugs that potentially may cause seizures and increase brain metabolism.

Because of craniofacial disorders in ARS patients, difficulties in airway management should also be considered. Because of this concern, we kept the patient breathing spontaneously. Due to the possibility of delayed awakening and residual respiratory paralysis after anesthesia, ventilation facilities, and intensive care units should also be available.

Conclusion

In conclusion, we tried to use an anesthesia method with reduced risk factors under cautious monitoring to the best of our knowledge, and our ARS patient underwent surgery without significant problems. However, if the patient did not suffer from mental retardation and could cooperate, sedation only, without general anesthesia, could be a safer approach.

Acknowledgment

None.

Conflicts of Interest

The authors declare that there are no conflicts of interest.

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