

Anesthetic Management of a Child with Cerebro-Oculo-Facio-Skeletal Syndrome

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Serebro-Okülo-Fasiyo-Skeletal Sendromlu Çocukta Anestezi Yönetimi

ABSTRACT

Cerebro-oculo-facio-skeletal syndrome (COFS, Pena-Shokeir syndrome Type II, Cockayne syndrome Type II) is a rare progressive syndrome with autosomal recessive transition that may cause death in childhood.

We aimed to present the anesthesia management in congenital cataract surgery in a patient with COFS in the present study.

Keywords: General anesthesia, rare disease, pediatrics, Pena-Shokeir syndrome Type II

ÖZ

Serebro-okülo-fasiyo-skeletal sendrom (COFS, Pena-Shokeir Syndrome Type II, Cockayne Syndrome Type II); çocukluk çağında ölümlere neden olabilen otozomal resesif geçişli, nadir görülen progresif bir sendromdur.

Bu olgu sunumu ile COFS tanılı hastamızda konjenital katarakt cerrahisi için anestezi yönetiminin sunulması amaçlanmıştır.

Anahtar kelimeler: Genel anestezi, nadir hastalık, pediatri, Pena-Shokeir sendromu Tip II

Alındığı tarih: 21.11.2018

Kabul tarihi: 19.12.2018

Yayın tarihi: 31.01.2019

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INTRODUCTION

Cerebro-oculo-facio-skeletal syndrome (COFS, Pena-Shokeir syndrome Type II, Cockayne syndrome Type II) is a rare progressive syndrome with an autosomal recessive transmission that may cause death in childhood.

The syndrome has many components such as facial dysmorphism, micrognathia, microcephaly, microphthalmia, cataract, blepharophimosis, kyphoscoliosis, hypotonia, hyporeflexia, arthrogyrosis, generalized osteoporosis, congenital flexion contractures, growth retardation, convulsion, severe mental retardation, and progressive degeneration of the brain-spinal cord⁽¹⁾. In this study, we aimed to present the anesthesia management in congenital cataract surgery in a patient with COFS.

CASE PRESENTATION

The patient, who was a 7-month-old girl weighing 4500 g (<3rd percentile), and 59 cm (<3rd percentile) in height, presented with symptoms of decreased vision in both eyes to ophthalmology outpatient clinic. Her history revealed that she was born by cesarean section (2800 grams) at the 39th week of gestation due to non-progressive delivery and deceleration. Because she had coarse crying and unable to open her eyes widely at the age of 3 days, a general practitioner ordered a karyotype analysis. According to analysis, her karyotype was found as 46XX karyotype.

She had mental, and motor retardation, coarse crying, typical facial dysmorphic appearance, retro-micrognathia, nystagmus, distinctive nose root, small and deeply located eyes, hollow palate structure,



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upper lip covering the lower lip, camptodactyly, split nipples, flexion contractor in upper and lower extremities, and rocker-bottom feet. She was diagnosed with COFS and followed-up by both pediatric genetics and pediatric cardiology departments. Transthoracic echocardiography revealed left ventricular dilatation, patent foramen ovale, patent ductus arteriosus, and first- and second-degree tricuspid valve insufficiency.

She could not follow the light and had bilateral leukocoria. She was also diagnosed with bilateral congenital cataract. Surgical treatment with bilateral lensectomy and anterior vitrectomy was scheduled. Her general condition was normal in the perioperative period. Her disease severity was classified as The American Society of Anesthesiology (ASA) grade III, her body temperature (36.5°C), heart rate (132 min⁻¹), respiratory rate (30 min⁻¹) were within physiologic limits and she had physical symptoms of COFS. Patient's family gave informed consent for anesthesia. Departments of Pediatric Genetics and Pediatric Cardiology approved the surgery with the recommendation of subacute bacterial endocarditis prophylaxis. Preoperatively whole blood count, biochemical tests, and telecardiogram results were within normal limits. Premedication was performed by using 0.05 mg kg⁻¹ intravenous midazolam after proper period of fasting and intravenous hydration. Anesthesia management was initiated in the operating room following intravenous 1 mg kg⁻¹ lidocaine and 2 mg kg⁻¹ propofol induction. A laryngeal mask airway (LMA) no: 1 was properly inserted after oxygenation with easy mask ventilation. Endocarditis prophylaxis was administered. Intraoperatively, maintenance of anesthesia was achieved by using intravenous remifentanyl (0.025-0.1 µg kg⁻¹ min⁻¹) and propofol infusion (25-100 µg kg⁻¹ h⁻¹). The basal fluid deficit was corrected with administration of NaCl and 5% dextrose solution during the total surgery time of 60 minutes. Intravenous paracetamol infusion (15 mg kg⁻¹) was administered 15 minutes before the end of surgery for pain management, and the patient was properly awakened. Her vital findings in the recovery room were within normal limits, and then she was transferred to the ward.

DISCUSSION

COFS is a rare and progressive disease, which involves the central nervous and musculoskeletal systems. Lowry reported COFS as a new syndrome in 1971⁽¹⁾. Pena and Shokeir described COFS in 1974⁽²⁾. COFS has many components such as facial dysmorphism, micrognathia, microcephaly, microphthalmia, congenital cataracts, blepharophimosis, kyphoscoliosis, hypotonia, hyporeflexia, arthrogyrosis, generalized osteoporosis, congenital flexion contractures, growth retardation, convulsions, severe mental retardation, and progressive degeneration of the brain and spinal cord⁽³⁾. Orphanet November 2016 data reported its prevalence as 1<1.000.000 and 20 cases were reported worldwide⁽⁴⁾. Recurrent aspiration pneumonia, growth, and developmental retardation, and difficulties in nutrition cause early childhood deaths⁽⁵⁾.

Micrognathia might cause difficult intubation, and upper airway obstruction may be seen⁽⁶⁾. Preservation of airway enables adequate ventilation and control of respiratory complications. We took the necessary measures preoperatively in case of a difficult airway, taking into consideration the patient's current symptoms by providing different models and dimensions of the laryngoscope set, LMA devices in different models, pediatric gum elastic bougie, pediatric video laryngoscope, and invasive airway devices. Succinylcholine administration must be avoided in these patients due to the risk of hyperkalemia. Anesthetic drugs may interact with anticonvulsant drugs which are used for patients with COFS, who have an increased risk of pulmonary infection due to pulmonary hypoplasia, so an appropriate approach must be selected in anesthesia management⁽⁷⁾. Cardiac problems should be considered to prevent air embolism during surgery. In our patient, we were very careful due to current cardiac symptoms. Positioning must be performed with care due to secondary symptoms of spinal cord degeneration, hypotonia, hyporeflexia/areflexia, kyphoscoliosis, arthrogyrosis, joint contraction, and osteoporosis. We used protective gel-type equipment for all pressure sites during positioning of our patient, and a normothermic environment was maintained during surgery.

CONCLUSION

Rare syndromes such as COFS are special cases in terms of anesthesia management. We aimed to present our case and review the literature so as to increase awareness regarding anesthesia management in rare diseases ⁽⁴⁾. Preservation of airway ensures adequate ventilation and control of respiratory complications. We took the necessary measures preoperatively in case of a difficult airway, taking into consideration the patient's current symptoms by providing different models and dimensions of the laryngoscope set, LMA devices in different models, pediatric gum elastic bougie, pediatric video laryngoscope, and invasive airway devices. Succinylcholine administration must be avoided in these patients due to the risk of hyperkalemia. Anesthetic drugs may interact with anticonvulsant drugs which are used for patients with COFS, who have an increased risk of pulmonary infection due to pulmonary hypoplasia; an appropriate approach must be selected in anesthesia management ⁽⁵⁾. Cardiac problems should be considered to prevent air embolism during surgery. In our patient, we were very careful due to current cardiac symptoms. Positioning must be performed with care due to secondary symptoms of spinal cord degeneration, hypotonia, hyporeflexia/areflexia, kyphoscoliosis, arthrogryposis, joint contraction, and osteoporosis. We used protective gel-type equipment for all pressure sides during positioning of our patient, and a normothermic environment was maintained during surgery.

As a result, proper anesthetic management is very important in especially rare cases such as COFS. In our case report, we aimed to increase awareness of anesthesia management in patients with COFS.

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