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EDİTÖRE MEKTUP / LETTER TO THE EDITOR

Anesthesia management in a patient with Bardet-Biedl syndrome undergoing scoliosis surgery

Skolyoz cerrahisi geçiren Bardet-Biedl sendromlu bir hastada anestezi yönetimi

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To the Editor,

Bardet-Biedl syndrome (BBS), also known as Laurance-Moon-Bardet-Biedl syndrome, is a rare multisystem autosomal recessive genetic disorder. BBS is a multisystem immobile ciliopathy primarily characterized by retinal cone-rod dystrophy, obesity, postaxial polydactyly, cognitive impairment, hypogonadotropic hypogonadism, and/or genitourinary and renal malformations[1]. Secondary features of the syndrome include developmental delay, lack of speech, brachydactyly or syndactyly, dental defects, ataxia or poor coordination, lack of smell, diabetes mellitus, hypertension, congenital heart disease, liver abnormalities, musculoskeletal abnormalities[1,2]. Patients with BBS have to undergo multiple operations throughout their lives. This syndrome may cause various problems during anesthesia and postoperative period due to its many clinical symptoms. In cases with BBS, inspiratory and cardiac complications and metabolic problems are high, endotakeal intubation and anesthesia management are difficult. There are very few studies in the literature on anesthesia management during BBS treatment.

This report was made available for publication after informing the family and obtaining their consent, and written informed consent was also obtained for the publication of case details. The patient, who was admitted to the hospital due to scoliosis, was 13 years old, 1.55 cm tall, and weighed 69 kg. Kifectomy and osteotomy of the posterior elements were planned for the patient who was diagnosed with scoliosis as a result of the examinations. The patient who was born prematurely had polydactyly and syndactyly at birth. With the addition of hydronephrosis and rapid weight gain complaints at 6 months, the diagnosis of Bardet Biedl syndrome was confirmed by genetic tests. The patient was operated under general anesthesia for polydactyly and syndactyly at the age of 1 year. She was diagnosed with retinitis pigmentosa at the age of 7 years. The patient had clinical symptoms of the syndrome, such as obesity, hypogonadotropic hypogonadism, hypothyroidism, and mild mental retardation. Physical examination revealed Mallampati class II, short neck, high palate, wide tongue, no murmur on cardiac auscultation, other systemic examinations were normal. All laboratory tests were within normal limits. Urine output was sufficient. There were no abnormal findings in the electrocardiogram (ECG) and echocardiography. Vertebral X-ray showed scoliosis.

Invasive blood pressure, electrocardiography, pulse oximetry, end tidal carbon dioxide monitoring, neuromuscular monitoring were performed intraoperatively. CVP catheter was opened. Before the application of anesthesia, devices were prepared in anticipation of airway obstruction. Anesthesia induction was achieved with iv 2mg/kg propofol, 1 mcg/kg fentanyl and 0.6 mg/kg rocuronium bromide. The patient's mask ventilation was comfortable. Endotracheal intubation was performed with a spiral cuffed endotracheal tube numbered 6.5

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without any difficulties. Anesthesia was maintained without hemodynamic complications by intravenous administration of propofol and remifentanil. During the operation, normothermia was provided with a heating blanket, and precautions such as heated intravenous infusion were taken against hypothermia. The operation was completed in 7 hours. Decurarization was achieved with 2 mg/kg sugammadex. Postoperative acute pain control was provided with 15 mg/kg paracetamol and 1.5-2 mg/kg tramadol. The patient was extubated and transferred to the pediatric intensive care unit. The patient was taken to the service 1 day later and discharged on the 3rd day.

BBS is a rare autosomal recessive disease with a prevalence of 1:160,000 in Northern European populations and 1:13, 500 in the Middle East[2]. BBS is a ciliopathy secondary to basal body dysfunction[3]. Ciliopathies are a group of genetic diseases caused by defects in the function or structure of cellular primary cilia, caused by mutations in more than 20 different genes[4]. In clinical practice, ciliopathies are characterized by immobile cilia defect, retinitis pigmentosa, polydactyly, situs inversus, learning disability, cystic kidney, liver and pancreatic cysts. Skeletal dystrophy may be present in 4% of patients with BBS. Impairment of the function of BBS proteins can lead patients to develop osteoporosis and appears to predispose to interfere with bone health[5].

In patients with BBS, facial dysmorphism, high palate, morbid obesity, and dental anomalies may complicate mask ventilation during anesthesia, and abnormalities in the epiglottis may cause difficult intubation. Bifid epiglottis, an extremely rare congenital anomaly, has been reported in relation to this syndrome[6]. In the preoperative anesthetic evaluation of our patient, mallampati class II, truncal obesity, short neck, wide tongue were present. Advanced airway devices such as video laryngoscope, fiberoptic laryngoscopy were prepared for the patient in anticipation of airway obstruction. Mask ventilation was comfortable and intubation was performed with direct laryngoscopy in the first attempt.

About 50% of patients with BBS are hypertensive and 32% have diabetes mellitus that needs to be optimized. We were able to provide controlled hypotension and stable hemodynamics throughout the operation with intravenous administration of propofol and remifentanil. Cardiac anomalies are found in 50% of BBS. In addition to congenital heart defects (ASD, VSD, PDA, transposition of great vessels, etc.), interventricular septal hypertrophy and dilated cardiomyopathy can also be seen[7]. Therefore, echocardiographic examinations should be performed together with clinical evaluation. Our patient had no abnormal findings on echocardiography. Subclinical hypothyroidism is accompanying in 19.4% of the patients[8]. Our patient's hypothyroidism was under control with levothyroxine.

Since both pharmacokinetic properties and neuromuscular monitoring will be applied during scoliosis surgery, we preferred total intravenous anesthesia for the maintenance of anesthesia. Remifentanil has an ester structure and is metabolized by ester hydrolysis; therefore, it is characterized by extrarenal excretion and short duration of action. Rocuronium has been preferred as a muscle relaxant due to its rapid onset of action, no histamine release, and containing specific antagonist. Sugammadex was used to rapidly and completely reverse rocuronium-induced neuromuscular blockade. In these patients, ciliary dysfunction may cause thickening of secretions, leading to difficulties in removing secretions after general anesthesia. Therefore, avoiding residual blockage becomes even more important to ensure the safety of the patient's airway. The use of sugammadex in patients with kidney transplantation or renal failure is controversial. Sugammadex is excreted by the kidneys; up to 97% cleared within 24 hours in cases of normal kidney function. In cases of severe renal failure, the clearance of sugammadex is reduced by 17-fold and the elimination half-life increased by 15-fold and is not licensed for severe renal impairment[9]. BBS is reported to cause renal tubular dysfunction, decreased urine concentration, glucosuria and aminoaciduria in 50% of affected patients. . Renal failure is the major contributor to early mortality and morbidity from BBS[10]. Our patient had normal renal functions and sugammadex was preferred for airway safety.

As a result, general anesthesia was safely administered in the patient who was diagnosed with BBS and underwent scoliosis surgery. Although no complications were observed in our case, difficulties may be encountered in hemodynamic stability and airway management due to the multiple system anomalies of this syndrome. We achieved hemodynamic stability by intravenous administration

of propofol and remifentanil. We think that this combination is an appropriate and safe anesthetic approach in scoliosis surgery in children with BBS. In BBS treatment, safe anesthesia management can be achieved by making a good preoperative evaluation, examining the airway, carefully evaluating cardiovascular abnormalities, kidney function and endocrine function.

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