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Case Report Open Access

Anesthetic Management of a Patient with Laurence Moon Biedl Bardet Syndrome

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Summary

Laurence Moon Bardet-Biedl syndrome (LMBBS) is a rare autosomal recessive disorder with clinical and genetic heterogenenity. This syndrome was first described by Laurence and Moon in 1866 and additional cases were described by Bardet and Biedl between 1920 and 1922. The main features are obesity, polydactyly, pigmentary retinopathy, learning disabilities, and various degrees of intellectual impairment, hypogonadism, and renal abnormalities. Bardet-Biedl syndrome is both phenotypically and genetically heterogeneous. Clinical diagnosis is based on the presence of 4 of the 5 cardinal features.

LMBBS needs special attention in anesthetic management due to the syndromic features causing general status instability which should be dealt promptly in an emergency surgery status.

Literature review revealed a few case reports about this rare syndrome. We describe the presenting features of the syndrome and management of a patient who was scheduled for emergency surgery under general anesthesia. We also discuss the relevant points for the busy anesthesiologist.

Introduction

LMBBS is characterized by cone-rod dystrophy, postaxial polydactyly, cognitive impairment, male hypogonadotrophic hypogonadism, complex female genitourinary malformations, truncal obesity, and renal dysfunction [1,2]. A renal disease is a major cause of morbidity and mortality. The secondary features include speech disorder, cardiovascular abnormalities, retinal abnormalities, diabetes mellitus, diabetes insipidus, ataxia, hepatic and dental involvements [1,2].

This syndrome is much common in the Middle East with an incidence of 1:13,500. In the rest of the world, the incidence is 1:160,000 with a male-to-female ratio of approximately 1.3:1. Renal failure in 95-100%, obesity in 90-95%, mental retardation in 80-87%, retinopathy in 91-93%, polydactyly in 70-74% and hypogonadism in 65-69% [3,4].

The diagnosis of LMBBS is established by the clinical findings. Twelve genes are known to be associated with Bardet-Biedl syndrome: BBS1, BBS2, ARL6/BBS3, BBS4, BBS5, MKKS/BBS6, BBS7, TTC8/BBS8, B1/BBS9, BBS10, TRIM32/BBS11, and BBS12. Molecular genetic testing is available on a clinical basis for p.M390R, the common mutation in BBS1 that is present in approximately 18%-32% of individuals with BBS and p.C91LfsX4 (also known as C91fsX95), a common mutation in BBS10, that is present in 10% of individuals with BBS. However, despite the identification of 12 BBS genes, the molecular basis of BBS remains elusive, it is now evident that all of the known BBS proteins are components of the centrosome and/or basal body and have an impact on ciliary transport [3,4].

We describe the presenting features and management of a patient who was scheduled for emergency surgery with general anesthesia and discuss the relevant points for the busy anesthesiologist.

Case Presentation

A 24-year old woman patient with lmbbs was diagnosed with cholecystitis and written informed consent was obtained from her parents for the operation and to publish this case report. She had chronic renal failure and had been undergoing dialysis treatment for the previous five years. In the upper abdomen ultrasound examination, the right kidney was atrophic, and the left kidney was measured 44×23 mm. She was obese (Body-Mass Index [BMI] 32.5kg/m2) but did not have diabetes mellitus. She showed bilateral nystagmus and was mentally retarded. The patient had an active infiltration in her lungs,mild fever, elevated white blood cell, and being treated oral antibiotic. (amoxicillin). In her echocardiographic examination, left ventricular septal hypertrophy and pulmonary hypertension (systolic pulmonary artery pressure 55 mmHg) were reported. Systemic hypertension did not exist. Laboratory examinations of the patient showed: glucose 105 mg dL^{-1}, urea 80 mg dL^{-1}, creatinine 5.6 mg dL^{-1}, AST; 43 U/L, ALT; 70U/L, cholesterol 245 mg dL^{-1}, triglyceride 490 mg dL^{-1}, total protein 8 mg dL^{-1}, hgb 12 gdL^{-1}, htc 34.4%. Hepatic markers were negative.

One day before operation, the patient underwent hemodialysis. In her airway assessment, a class III Mallampati airway was revealed. She was premedicated with 4mg i.v. midazolam (0.70-0.1 mg kg^{-1}) 45 minute prior to the operation. Monitoring included intra-arterial blood pressure, electrocardiography and pulse oximetry. A difficult airway cart was kept ready. Sodium thiopental (480mg) was used for induction of anaesthesia with fentanly (100mcg) and atracurium besylate (50mg) was administered to facilitate tracheal entubation. At induction, we attempted to mask ventilate the patient and it was easy. An endotracheal tube (number 7.5) was used for entubation of trachea. Anesthesia was maintained with sevoflorane (1-2 % Minimum alveolar concentration) in 35%O_2 in N_2O. Peroperative arterial blood pressure and heart rate were stable. The operation lasted for 1 hour and the intraoperative course was uneventfull. After the operation the patient was extubated and she was transferred to the intensive care unit. This was a planned

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admission. Our concerns were about patient's problems. Postoperative analgesia was achived with 75 mg diclofenak im. She was discharged on the forth day postoperatively.

Discussion

Patients with this syndrome frequently require multiple anaesthetic procedures for both diagnostic and therapeutic measures including MRI, uro-gynaecological procedures, or corrective surgeries for limb deformities [1]. The perioperative period may be hazardous for patients with this disorder. Patients with LMBB syndrome should be assessed carefully in terms of general anesthesia.

Obesitiy is a cardinal feature of the disease. It is the leading feature of this syndrome and 85% of the LMBBS patients are obese. Obesity in these patients forms a further predisposition for endocrine and cardiovascular failures [4]. Low and his colleagues reported several anaesthetic problems associated with this syndrome including obesity with consequent problems with venous access and placement of local anaesthetic blocks [1]. Depending on obesity, in these patients, difficulties can be faced during regional block application and finding veins. Facial dysmorphism, and dental abnormalities can create difficulties in the mask ventilation during the anesthesia, and the abnormalities seen in the epiglottis can also create an intubation difficulty [5]. Our patient was assessed with Mallampati score 3 and a difficult airway cart was kept ready but fortunately we did not have any problem in the intubation.

Besides the risks that could develop under the anesthesia related with obesity, attension should be paid to hypertension, diabetes mellitus and existing heart diseases.Hypertension, diabetes mellitus should be regulated well in the preoperative period. DM develops in 32% of these patients [3]. Our patient did not have DM, therefore there was no need to analize blood glucose values continuesly. Hypertension is seen 50% of patients. A high incidence of congenital and aquired heart disease was reported. Elbedour et al. reported 11 out of 22 patients (50%) had hypertrophy of the interventricular septum and dilated cardiomyopathy [6]. Furthermore these patients have abnormal electrical and autonomic behaviors that represent risk factors for life threatening arrhythmia and fatal events. Because of this we would suggest it is prudent to consider a 12-lead ECG and echocardiogram during the preanaesthetic assessment.

In these patients, nephrotoxic agents should be avoided as much as possible [7]. In our patient, a full renal failure was developed and the patient became dependent on dialysis. Peroperative fluid consumption was recorded and it was carefully followed.

One of the most frequent symptoms of the syndrome, retinal dystrophy occurs in the early childhood and can develop rapidly into blindness. In the patients with this syndrome, nystagmus, myopia, glaucoma are reported in the cataract [1]. Interaction of the medicines used for these ocular diseases with anesthetic medicines should also be considered [1].

Special attension should be given to mentally retarded patients, and they should be premedicated before coming to the operation room. Our patient was also mentally retarded and had premedication 45 minutes before the operation time so no problem occured in the operating theatre.

Anesthesiologists should prefer appropriate anesthetic tecniques in regards of the operation site and the patients status. For example; R.Majahan and his colleagues reported that they had applied combined spinal-epidural block to avoid the risk of general anesthesia for the operation that is done for a patient with LMBB syndrome, dilated cardiomyopathy and fractured right femur and tibia requiring open reduction and internal fixation. They reported that the peroperative course was uneventful [7]. When regional anesthesia could not be performed But general anesthesia with appropriate anestetic drugs should be used. For example Ewa Podwinska and her colleagues reported that their patient who was diabetic, hypertensive,obese and mentally retarded and with laparoscopic cholecystectomy was presented for LMBBS syndrome. They induced anesthesia with thiopentone and fentanyl,and used rocuronium bromide as a muscle relaxation, and later on maintained anesthesia with sevoflurone like us [8]. They reported that peroperative and the rest were uneventful. Our patient was stable throught the operation and experienced no adverse events as well.

Conclusion

General anaesthesia was safely performed on this patient with a rare congenital condition. Although no complication was encountered in our case, this syndrome has the potential for difficulties in managing the airway and the cardiovascular and renal systems. Patients with LMBBS should be prepared carefully in preoperatively and anesthesiologist should be prepared for difficulties that may develop intraoperatively.

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