Spinal Anaesthesia For A Patient With Bloom's Syndrome

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Citation

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Abstract

Bloom's syndrome (BS) is a rare autosomal recessive disorder by proportionate pre-and postnatal growth deficiency; sun-sensitive, telangiectatic, hypo- and hyperpigmented skin; and chromosomal instability. There is no report in the anesthetic literature using regional anesthesia for patients with BS. In a BS patient, it should be paid attention to potential difficult airway and immunodeficiency. In this paper, we reported the anesthetic experience of a male patient with Bloom's syndrome who undergoing minor urologic procedure.

INTRODUCTION

Bloom syndrome is a rare autosomal recessive genetic disease that features an elevated rate of sister chromatic exchange [1,2] which is assumed to be responsible for the phenotype and the cancer predisposition [3,4,5] . Although BS's incidence is unknown, in USA more than 100 case have been made. The male-to-female ratio is 1.3:1. Bloom syndrome is more common in eastern European Ashkenazi Jews.

The disorder is caused by loss of function of a 3' to 5' RecQ DNA helicase, BLM [1]. BLM was mapped to 15q26.1 and its product was found to encode a RecQ DNA helicase. A single predominant mutation of BS was reported in Ashkenazi Jews [4,6] . There the predominant mutation, referred to as “blmAsh,” is a 6-bp deletion and 7-bp insertion at nucleotide position 2281 in the BLM cDNA [7] .

The genome in BS somatic cells in unstable, and hypermutability explains many clinical features [8] . At cellular level is an increased frequency of spontaneous mutation [9] and somatic recombination [10] . Histological changes comprise an increase in dilated vessels in the upper dermis and damage and loss of elastic fibers [11] .

Term birth measurements confirm that the growth deficiency of BS has prenatal onset. It is reported that children with Bloom syndrome have significant growth retardation and wasting.

Growth continues by at least 1 cm/year until age 21 years for both sexes. More than half of children with Bloom syndrome are significantly wasted until age 8 years, which is not related to early death or underlying malignancy. The mean body mass index for adults with Bloom syndrome after age 25 years is low normal (n = 22, mean = 20.2 kg/m ^{2}) [12] .

Affected individuals, who have inherited two copies of the Bloom's syndrome gene mutation, typically have the following features: (a) Growth: Prenatal onset growth deficiency; average adult male height, 151 cm, and adult female height, 144 cm (b) Craniofacial: Mild microcephaly, malar hypoplasia, with or without small nose (c) Skin: Facial telangiectatic erythema involves the butterfly midface region and is exacerbated by sunlight. Small and large areas of hyperpigmentation and hypopigmentation [6,7,8,11,13,14,15] .

Eye findings have rarely been mentioned of this syndrome. Sahn et all. [16] reported a child with Bloom syndrome who had pronounced bulbar conjunctival telangiectasia originally diagnosed as episcleritis.

The 14 Japanese cases reported by German and Takebe [17] differed somewhat from most cases recognized elsewhere in that dolichocephaly was a less constant feature, the facial skin lesions were less prominent, and life-threatening infections were less frequent.

BS predisposes affected individuals to a wide variety of neoplasms including hematological malignancies due to genomic instability that arise than earlier expected in the general population [3,4,5,8,13,18] . Of great importance is the high leukemia morbidity among individuals with this syndrome; chromosomal aberrations and breakages play a significant role [11] . Twenty-seven percent of patients with BS have

malignant neoplasms at a mean age of 20.7 years [18] . Poppe et all present the karyotypic findings in a BS patient diagnosed with acute myeloid leukemia (AML), FAB subtype M1, showing the preferential occurrence of total or partial loss of chromosome 7 in BS patients with AML or myelodysplastic syndromes [19] .

The syndrome is also associated with a facultative lack of antibodies. Patients with Bloom syndrome have decreased immunoglobulin A and immunoglobulin M, with recurrent respiratory and gastrointestinal tract infections. Suspectibity to infection decreases with age. However, chronic lunge disease has been responsible for three deaths, at, age 18, 19 and 24 [6,11] .

In BS, diabetes mellitus unusually frequently develops as a complication. The onset of diabetes in patients with BS is in late adolescence or early adulthood [6,20] reported on a 21-year-old Japanese male patient with BS who exhibited impaired glucose tolerance (IGT) in the initial oral glucose tolerance test (OGTT) and had developed patterns of diabetes mellitus by the second OGTT at the 2-years-and-2-months follow-up. In addition to, when a person with BS reaches late adolescence or early adulthood, an OGTT is necessary to ascertain whether the patient has GT or diabetes mellitus as a complication.

Since the first description of BS in 1954, five cases of primary head and neck cancer have been identified in the first 103 patients, including two tongue carcinomas and three laryngeal carcinomas. The patients ranged in age from 26 to 34 and included smokers and nonsmokers [6,18] . Head and neck cancer represents approximately 6% of all human tumors. This is in contrast with an 18% incidence rate of head and neck cancer among all cancers observed in BS patients. The head and neck surgeon should consider BS in the differential diagnosis of young cancer patients [18] .

Jain D et all. [13] reported sibs with BS. The older, a 15-year-old developed a hepatocellular carcinoma, a neoplasm not yet reported in association with BS. The younger, developed an anaplastic Wilms tumor (WT) associated with nephrogenic rests at the age of 3 1/2 years, and this was followed by a myelodysplastic syndrome. These examples expand the spectrum of malignancies occurring in BS to include liver cell neoplasms, and confirm the association of nephrogenic rests with WT, even in the setting of BS.

In fertility due to lack of spermatogenesis is the rule in males. Subfertility in females may be common [6] . Despite reduced fertility, conception can occur, and women with Bloom syndrome should receive appropriate reproductive counseling to prevent unintended pregnancies and increased surveillance for preterm birth. Chiskolm CA et all. [14] reported a 19-year-old woman with typical clinical features of Bloom syndrome with a successful pregnancy. Because of her small pelvis on clinical examination, the patient underwent computed tomography pelvimetry, which showed adequate pelvic capacity. Preterm labor occurred at 32 weeks' gestation, and the infant was ultimately delivered at 35 weeks' gestation. The infant was less than the tenth percentile for length and weight for gestational age, but was otherwise.

It is reported that difficult intubation can be expected in this patients [21] . We were unaware of any regional technique for surgery with BS patient. In this paper, we reported the anesthetic experience of a male patient with BS who underwent minor urologic surgery.

CASE REPORT

A 23 year-old-man with Bloom syndrome has been diagnosed by pediatric physicians at 1983. Patients chromosomes analyze were done by using Sister Chromatid Exchange (SCE) Analyse at Akdeniz University Hospital. His chromosome analyze results was 61.3% sister chromatic exchange, 12 single and double chromatic breakage, 19% abnormal chromosome, 1 deletion. Operation was planned because of urethral obstruction by urological surgeon. He was premedicated before operation with 0.03 mg/kg midazolam. On arrival in operation room, 0.5 liter 0.9 % isotonic solution was given through an 18-gauge cannula by the left forearm. Monitoring consisted of noninvasive arterial pressure measurement, ECG and pulse oximeter.

Physical examination showed a 40 kg man, height 143 cm. Her face was typical of BS with telangiectatic erythema, prominent nose and ears (Figure 1). He had small jaw, abnormal gape (Mallampati grade 4) (Figure 2).

Figure 1

Figure 1: Telangiectatic erythema, prominent nose and ears.

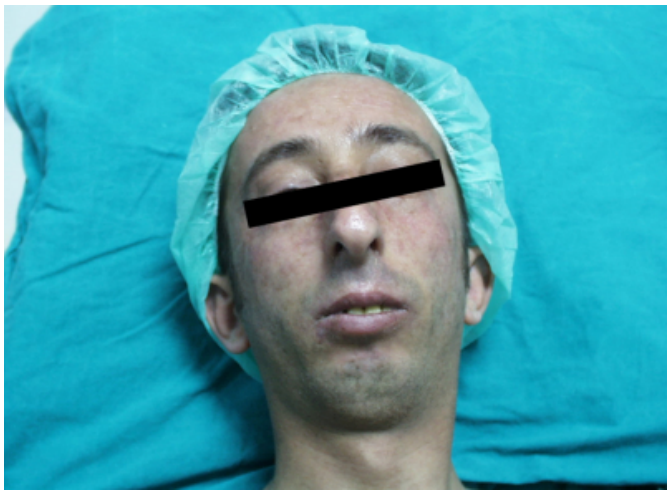
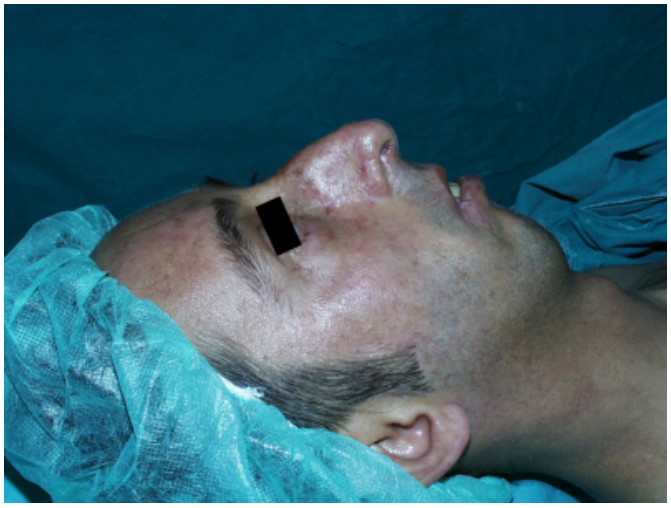


Figure 2

Figure 2: Small jaw, abnormal gape



Investigation revealed a hemoglobin concentration of 12.4 g dl^{-1} , platelet account 208 x 10^{9} lt^{-1} , prothrombin time 11.3 minute, activated partial thromboplastin time 24.7 minute, sodium 143 mmol lt^{-1} , potassium 4.1 mmol lt^{-1} , urea 11 mmol lt^{-1} , creatinine 0.55 µmol lt^{-1} .

0.9 % NaCl solution (500 ml) was given as a preload. Spinal block was performed at L_{3-4} interspaces by using 25 G spinal needle in midline approach with the patient in the right lateral position. Dural puncture was confirmed by the sight of clear CSF at the hub of the spinal needle, after which 0.75 % isobaric ropivakain 2 ml was injected. Immediately after the injection, the patient was placed supine position. Patient remained in the supine position for 5 min and then placed in the lithotomy position for the operative procedure. Motor block was assessed by the loss of lower limb muscle power a four-point modified Bromage scale. Sensory block was assessed by pinprick. Within 5 min of spinal injection, bilateral sympathetic, sensory (T6) and motor (unable to flex knees or feet) blocks were produced.

The patients remained comfortable during the procedure and required no analgesic supplementation. Haemodynamic stability was maintained throughout the operation. Heart rate averaged 90 beats.min^{-1} and systolic arterial pressure 123/72 mmHg and oxygen saturation 98.-100% (4 lt of oxygen were administered by nasal canula). The patient hadn't postoperative problem and 1 week later the patient went for second operation. All of the laboratory assessments were normal and that time spinal anesthesia was performed with %0.5 hyperbaric bupivacaine 1.5 ml. Within 5 min of spinal injection, bilateral sympathetic, sensory (T4) and motor (unable to flex knees or feet) blocks were produced. The patients remained comfortable during the procedure and required no analgesic supplementation and haemodynamic stability was maintained throughout the operation.

DISCUSSION

Bloom syndrome is a rare autosomal recessive genetic disease which is characterized by microcephaly, malar hypoplasia. General anesthesia can be complicated by increased likelihood of difficult intubation.

Induction of general anesthesia followed by direct laryngoscopy and oral intubation is dangerous, if not possible, in several situations such as congenital anomalies, anatomic variations [22] . To determine the optimal intubation technique, the anesthesiologist must elicit an airway history and carefully examine the patient's head and neck. If a facial deformity is severe enough to preclude a good mask seal, positive pressure ventilation may be impossible. In a BS patient, it should be paid attention to potential difficulties with mask fit and laryngoscopy [21] because of microcephaly, malar hypoplasia and prominent nose.

Among the neoplasms in BS patients, 18 % of them are head and neck's malignant tumor's which will frequently require surgical treatment and anesthesia. The head and neck surgeon should consider BS in the differential diagnosis of young cancer patients [18] . Also in BS should be aware of head and neck cancers which cause difficult intubation.

Aono J. et all. [21] reported the anesthetic experience of a male patient (37-yr-old, 26 kg, 136 cm) with Bloom's syndrome who underwent emergency laparotomy. In this case, they carried out the awake laryngoscopy, but they could not see

the vocal cord directly and the trachea was blindly.

If general anesthesia is suitable, routine oral and nasal intubations can be performed in wakeful patients. The alternatives are awake nasal fiberoptic intubation and awake nasal blind intubation. The final decision depends on the availability of a bronchoscope and personnel experienced in its use. Regardless of which alternative is chosen, an emergency tracheotomy may be necessary. Therefore, an experienced team including a surgeon should be in the operating room, all necessary equipment should be available and unwrapped, and the neck should be prepped and draped [22].

In BS, regional anesthesia may choose because of difficult airway expectation. Spinal block can be desirable in this patient who had underlying lung infections or a potentially difficult airway. If spinal anesthesia ascending into the cervical levels, it causes severe hypotension, bradycardia, and respiratory insufficiency. Treatments of high spinal anesthesia consist of the airway, maintaining adequate ventilation, and supporting the circulation [23]. But in BS this treatments are impossible. Spinal block level should be well controlled. There has been no case report using regional anesthesia in a patient with BS for surgery.

In our case we preferred spinal anesthesia for minor urologic procedure. We thought that spinal catheter is the best choice but we haven't spinal catheter. Two different local anesthetic and concentration were used. At first operation we used isobaric solution and found high spinal block. So, at second time we decided to use hyperbaric solution and found same dermatomal level.

Patients with Bloom syndrome have decreased immunoglobulin A and immunoglobulin M. with recurrent respiratory and gastrointestinal tract infections. This immunodeficiency is associated with recurrent respiratory tract (including ear) and gastrointestinal infections, and increased incidence of coeliac disease and autoimmune disorders. The susceptibility to infections improves with age, even if the deficiency persists. Treatment includes prophylactic antibiotics, especially during winter months, monitoring and treating associated complications. Immunoglobulin replacement therapy is not indicated. Some patients occasionally develop antibodies to Immunoglobulin A in blood products, which potentially may cause severe reactions [24].

Diabetes mellitus unusually frequently develops as a complication in BS. Successful perioperative glucose management depends on careful monitoring. The regimen selected to manage diabetics undergoing surgery depends on the severity of the diabetes and the magnitude of the surgery. But in our case patient haven't diabetes mellitus and its complications [25].

In summary, at anesthesia management of patients with Bloom Syndrome should be careful because of difficult airway risk and infection risk. In this situation, regional low dose anesthesia may have low complication rate than general anesthesia.

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