Anesthetic Management of Gaucher Disease

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ABSTRACT

Gaucher, the most common form of lysosomal storage diseases, is caused by an inherited deficiency of the beta-glucocerebrosidase enzyme. Gaucher disease may present a challenge to the anesthesiologist because of abnormal coagulation and multiorgan disease in obstetric situations. These factors may affect choice of anesthesia type. We aimed to report the diverse anesthetic management of Gaucher Disease depending on patient's characteristics in two different cases.

Key words: *Gaucher, anesthesia, obstetric, thrombocytopenia*

Gaucher Hastalığında Anestezik Yaklaşım

ÖZET

Gaucher, lizozomal depo hastalıklarının en sık görülen formu olup, kalıtsal beta glukoserebrozidaz enzim eksikliği nedeni ile oluşur. Gaucher hastalığı, obstetrik durumlarda anormal koagülasyon ve çoklu organ tutulumu nedeniyle anestezistler için zorluk oluşturur. Bu faktörler anestezi türünün seçimini etkileyebilir. Bu olgu sunumunda iki olguda Gaucher hastalığında hastaların özelliklerine göre değişebilen farklı anestezik yaklaşımların bildirilmesini amaçladık.

Anahtar kelimeler: *Gaucher, anestezi, obstetrik, trombositopeni*

INTRODUCTION

Gaucher, the most common form of lysosomal storage disease, is caused by an inherited deficiency of the beta-glucocerebrosidase enzyme. Pathology results from the accumulation of glucocerebroside in the reticuloendothelial system. Three variants are usually recognized: type I, non-neuropathic or adult form, type II, acute neuropathic or infantile form, type III, subacute neuropathic or juvenile form (1-3). Type I (adult, chronic, nonneuronopathic) is the most common and mildest form, marked by absence of neurological involvement by virtue of the presence of the common 1226G (N370S) mutation, is especially prevalent among the Ashkenazi Jewish population with a disease frequency of about 1:850 in live births. It is also present in different ethnic groups with a frequency between 1:40 000 to 1:60 000 in the general population. Patients often present with hepatosplenomegaly and a combination of anemia, thrombocytopenia, and leucopenia (4). Type I Gaucher disease may present a challenge to the anesthesiologist because of abnormal coagulation and multiorgan disease in obstetric situations. These factors may affect choice of mode of delivery and the type of anesthesia (5). We aimed to report the diverse anesthetic management of Gaucher Disease depending on patient's characteristics in two different cases.

CASE 1

A 29-year-old pregnant women (gravidia 4, parity 2, abortus 2, (weight 69 kg, BMI 27 kg/m2) with type I Gaucher's disease was admitted to our institution at 37th weeks' gestation with painful contractions. Gaucher's disease had been diagnosed 1 years earlier and her fam-

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ily history revealed that her two sisters had same diagnosis for two years. She had one successful vaginal birth and two abortus in past 5 years. She didn't take her enzyme therapy properly. She had hepatoesplenomegaly, as well as osteopenia and gastroesophageal reflux. Signs of pulmonary hypertension were absent and chest X-ray and ECG prior to pregnancy were normal.Due to fetal distress in non stress test, an emergency caesarean section was planned. Our patient did not provide any history of easy bleeding or bruising. Physical examination, including cardiopulmonary auscultation, was normal. Laboratory results were as follows: white blood cells: 3.871/mm3 hemoglobin (Hgb): 10.12 g/dL, platelet (plt): 82270/mm3, international normalized ratio (INR): 1.23. Blood group analysis was performed and the laboratory prepared 2 units of packed red blood cells to be used in case of need. Since her preoperative fasting period was not enough for general anesthesia we considered that the patient had a higher risk for aspiration (in the presence of pregnancy and gastroesophagic reflux), and since coagulation parameters were within normal limits, we decided that epidural anesthesia would be suitable for her. An epidural catheter was inserted via median approach at the L3/L4 interspace, in the left lateral position with 18G Tuohy needle, using loss of resistance technique. Epidural space was encountered at 3.5 cm and 5 cm of catheter was inserted in cephalic direction. 5mL of 2% lidocaine was administered, followed by 1mL (0.05mg) fentanyl and a total of 9 mL of 0.375% levobupivacaine, carefully titrated to achieve a sensory block at T4 level. Healthy female infant was delivered with an uneventful cesarean section. The patient remained haemodynamically stable. We estimated total blood loss to be 1050 mL. Postoperative analgesia was accomplished with epidural morphine (3 mg every 12 h) and intravenous acetaminophen (8 hourly).The catheter was removed without complications after 48 hours. Mother and child were discharged four days after surgery.

CASE 2

A 36-year-old pregnant women (gravidia 1, parity 1, (weight 66 kg, BMI 29 Kg/m2) with type I Gaucher's disease was admitted to our institution at 35th weeks' gestation with painful contractions. Gaucher's disease had been diagnosed 2 years. She had splenomegaly, as well as trombocytopenia and anemia (Hgb: 6.4 g/dL, plat: 40200/mm3, INR:1.25) We planned general anaesthesia because of trombositopenia and cardiac pathologies on transthoracic cchocardiography (Ejection fraction 45-50 %, moderate to severe aortic and mitral insufficiency).

The patient was positioned supine, with left lateral uterine displacement, to minimize aortocaval compression in addition to routine monitors (non-invasive blood pressure cuff, 5 lead ECG, pulse oximeter). The patient was lightly sedated with remifentanil infusion (0.02–0.08 μg/kg/min) to minimize increases in BP and HR. Modified rapid sequence induction was performed using remifentanil 1 μg/kg iv, propofol 2 mg/kg iv, and rocuronium 0.5 mg/kg. Using a 7.0 mm cuffed endotracheal tube, her trachea was easily intubated. After confirming correct endotracheal tube placement by capnography and auscultation, the obstetrician proceeded with caesarean delivery. Anesthesia was maintained using remifentanil (0.04–0.08μgkg–1min–1), sevoflurane 1.5% end-tidal concentration (in a mixture of 50% N2O), and oxygen. The patient remained hemodynamically stable. Bag-mask assisted ventilation of the newborn was required for one minute following delivery. Operation was lasted in 45 minutes.Residual paralysis was reversed with neostigmine 2.5 mg iv and atropine 0.5mg iv. After tracheal extubation, patient was transferred to ICU. The patient remained in the ICU for 18 hr, and was transferred to obstetric department and she was discharged 4th day of hospital stay.

DISCUSSION

Gaucher disease is still a dilemma for anesthesiologists as both general and regional anesthesia can be used in management of cases. Because GD is a disease with multiorgan involvement, preoperative assessment should be carried out in order to determine the extent of organ involvement, with particular emphasis on central nervous system, liver, spleen, bone, bone marrow and probably the lung (6,7). Hepatosplenomegaly is the most common sign of visceral involvement in GD and may be seen in all three types (8). Hepatic involvement may lead to cirrhosis of the liver, which results in hepatic fibrosis and portal hypertension. However, the latter may be seen in the absence of cirrhosis (8). Splenomegaly may produce hypersplenism, which, in turn, results in a variety of hematological disorders, including thrombocytopenia, hemolytic anemia and leucopenia (9). For patients with Gaucher's disease, the preoperative evaluation

of haematological status is of primary concern. Many have very low platelet counts with or without abnormal platelet function (10). Thrombocytopenia may be sufficiently severe as to preclude regional anesthesia (5). Coagulation factor deficiencies are also common in this population and may occur independently of platelet abnormalities (11).

Therefore, complete blood count and bleeding and clotting time investigation should be carried out. However, increased operative bleeding may be seen in the absence of clotting parameter alterations as a result of a platelet function abnormality, which is not unusual in GD patients (9). The anesthetic management of parturients with Gaucher disease will depend on the clinical manifestations of the disease. There is insufficient information in the literature to allow strong recommendations. We think that small number of patients with regional anesthesia does not provide proof of safety. Experience with large patient series, and the presence of reliable bedside tests of platelet function, may help make better decisions in the future. A review reported that because of a lack of data from a large series of reports, there have been no guidelines for anesthetic management of obstetric patients with this disease. Authors suggested that anesthetic management requires attention to hematological parameters before delivery. They also recommended careful attention to positioning and patient transfer due to the presence of hip prostheses and osteopenia (5).

In conclusion, multiorgan involvement of GD, coexisting systemic illnesses and laboratory abnormalities play key role in determination of the mode of anesthesia for obstetric patients with Gaucher disease. A multidisciplinary approach and extensive communication between the obstetrician, hematologist and anesthesiologist is crucial.

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