Anesthetic Management Of Parturient With Gaucher's Disease

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Citation

J Pinto, S Ribeiro, R Amaral, E Dionísio. *Anesthetic Management Of Parturient With Gaucher's Disease*. The Internet Journal of Anesthesiology. 2007 Volume 18 Number 2.

Abstract

Gaucher's disease is a genetic deficiency in the activity of the lysossomal enzyme ß-glucocerebrosidase, causing monocytes and macrophages to store excessive amounts of glucocerebroside in lysossomes. Pregnancy, labour and delivery in patients with type I Gaucher disease may present a challenge to the anesthesiologist because of abnormal coagulation and multiorgan disease. These factors may affect choice of mode of delivery and consequently the type of anesthesia used during delivery. We describe a case of anesthetic management of a pregnant woman with type I Gaucher's disease, presenting for caesarean-section.

INTRODUCTION

Gaucher's disease, the most common sphingolipidosis, is caused by an inherited defect of the lysossomal enzyme -glucocerebrosidase, with consequent accumulation of undegraded glucocerebroside in monocyte-macrophage cells, the Gaucher cells. [1,4] The diagnostic criteria for Gaucher's disease include a decrease in -glucocerebrosidase activity to less than 15% in peripheral blood lymphocytes or in cultured skin fibroblasts [1] , mutation analysis by polymerase chain reaction and chorionic villus sampling or amniocentesis testing for prenatal diagnosis.

Despite the heterogeneity of Gaucher's disease, three basic clinical forms have been distinguished based on the degree of neurological involvement. Most patients with Gaucher's disease have the non-neuronopathic form (type I). The remainder of patients with Gaucher's disease have either the acute neuronopathic form (type II) or the subacute neuronopathic form (type III) [4] . In the acute neuronopathic form, the neurological symptoms may include cranial nerve and extrapyramidal tract involvement. Neurological deterioration progresses quickly and death from apnoea or aspiration usually occurs in early childhood [4] . The neurological symptoms in the subacute neuronopathic form can include myoclonic seizures, dementia, ocular muscle apraxia, mental retardation, and myoclonus [1,4] .

Most patients with type I disease present with hepatosplenomegaly, anemia, thrombocytopenia and platelet aggregation defects.In some cases splenectomy is required to reduce the severity of thrombocytopenia. Liver function tests are usually normal [1] .

Skeletal involvement includes osteopenia, osteonecrosis, osteosclerosis, avascular necrosis, lytic lesions, bone pain and, in 20%, impaired mobility. Skeletal deformities affecting the pelvis and hips can lead to a higher incidence of C-section, although vaginal delivery can be possible with carefully positioning [3] .

Pulmonary involvement may include parenchymal lung disease, abnormal pulmonary function and pulmonary hypertension in severe cases.

CASE REPORT

A 25-year-old primigravida (weight 59kg, BMI 25Kg/m ^{2}) with type I Gaucher's disease was admitted to our unit at 38 weeks' gestation with painful contractions. Gaucher's disease had been diagnosed 4 years earlier and she was medicated with imiglucerase 15U/kg twice a month. According to the Haematologist's most recent information, she had hepatoesplenomegaly and thrombocytopenia, as well as osteopenia and gastroesophagic reflux. Signs of pulmonary hypertension were absent and chest X-ray and ECG prior to pregnancy were normal.

Due to fetal breech presentation, caesarean section was scheduled for that day. Our patient did not provide any history of easy bleeding or bruising. Physical examination,

including cardiopulmonary auscultation, was normal. Even though complete blood count showed 100 x 10^9 platelets, with normal haemoglobin levels and coagulation tests, we had our patient's blood typed and had the laboratory prepare 2 units of packed red blood cells in case of need. Because these values were within normal limits and considering that the patient had a higher risk for aspiration (pregnancy plus gastroesophagic reflux), we decided to perform an epidural anesthesia.

After informed consent, an epidural catheter was inserted via median approach at the L3/L4 interspace, in the sitting position with 18G Tuohy needle, using loss of resistance technique. Epidural space was encountered at 4cm and 4cm of catheter was inserted in cephalic direction. 5mL of 2% Lidocaine was administered, followed by 1mL (0,05mg) fentanyl and a total of 9mL of 0,75% ropivacaine, carefully titrated to achieve a sensory block (tested with thermal stimuli) at T4 level. Surgery was started and a healthy male infant was delivered. The patient remained haemodynamically stable (1000Ml of Ringer Lactact plus 1000mL of Normal saline were administrated). We estimated total blood loss to be 1050mL.

Postoperative analgesia was accomplished with epidural morphine (3mg every 12h) and intravenous acetaminophen (8-hourly).The catheter was removed 48 hours after, without complications. Mother and child were discharged four days after surgery.

DISCUSSION

The anesthetic management of parturients with Gaucher's disease can be challenging and depends on the clinical manifestations of the disease.

Most patients with type I disease present with hepatosplenomegaly, anemia, thrombocytopenia and platelet aggregation defects. Post partum haemorrhage occurred in 5 of 16 pregnancies in one case series [3] . For these reasons, obstetric patients with Gaucher's disease require a full hematologic work-up prenatally including: complete history of bleeding disorders (easy bruising, gum/nose bleeds, heavy/prolonged menses); complete blood count before pregnancy and at the beginning of the third trimester.

Patients who have low platelet counts should have these confirmed by manual counts. Coagulation factor assays should be done if the prothrombin or partial thromboplastin times are abnormal, such as fibrinogen levels and D-dimers, ristocetine, adenosine 5?-diphosphate, collagen and Von Willebrand factor.
In patients who have history of excessive or spontaneous bleeding, the risks associated with regional anesthesia /analgesia are unlikely to outweigh the benefits, and alternative methods should be used. Furthermore, the use of thromboelastography, a bedside test of coagulation, has not been evaluated in this setting. Whether it is safe to provide spinal anesthesia in the parturient with Gaucher´s disease, as in any parturient with a low platelet count, is uncertain.

Enzyme replacement therapy with recombinant imiglucerase does not seem to relieve pulmonary symptoms, thus some authors recommend performing an echocardiogram in patients on enzyme replacement therapy. [3] Although our patient was receiving imiglucerase, she didn´t present any signs of pulmonary hypertension and once echocardiogram isn´t readily available in our hospital, we decided not to perform it.

Haematological parameters need particular attention prior to delivery. Ideally, and in contrast to what happened in this case (we only had contact with the patient at 38 weeks pregnancy) the correct anesthetic management of these patients requires an early multidisciplinary approach including obstetrician, haematologist and anaesthesiologist in order to anticipate the possibility of cardiopulmonary complications associated with possible pulmonary hypertension, post-partum haemorrhage and to preclude skeletal damage.

References

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