Case Report

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Anaesthesia management of a young patient with homocystinuria

Abstract

Homocystinurea is a rare autosomal recessive genetic disease. It is caused by deficiency in cystathionine-beta synthase leading to defect in methionine metabolism. HIGH LEVEL of plasma homocysteine are associated with vascular injury via mechanism of oxidative damage. Vascular smooth muscle proliferation, promotion of platelet activation and aggregation and disruption of normal procoagulant-anticoagulant balance favoring thrombosis. This is a case of seventeen-year-old girl. Known case of homocystinurea who scheduled for cataract surgery for her dislocated lens. The major anesthetic consideration includes the development of thromboembolism, need to avoid nitrous oxide in balance anesthesia and avoid dehydration, long fasting, hypoglycemia.

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Introduction

We are reporting a case of Homocystinuria with megaloblastic anemia which we anaesthetized for cataract surgery.

Case report

A seventeen years female patient with Homocystinuria present for cataract surgery for her dislocated microspherical lens. She was diagnosed as homocystinuria patient since she was 11 years old when she present with recurrent illness, developmental delay, hyper pigmentation and decreased vision. Pediatrician worked up reveal the diagnoses which confirmed by amino acid chromatography. She received Pyridoxine & folic acid orally. She has no history of convulsion or thromboembolic presentation but yet she has marphanoid feature, anemia, decreased vision and mental retardation.

Pre-anesthetic assessment & planning

Assessment 2 weeks earlier reveal that the patient was uncooperative, mentally retarded, has high arched palate, long extremities and osteoporosis on X-ray, indeed, defective vision the concern of the operation. The requested investigations (CBC, RFT, blood glucose, urine analysis, & ECHO) were normal. Continuation of the pyridoxine and folate was advised in order to control the level of homocystine to the maximum reachable and improve the megaloblastic anemia; while advised to withhold aspirin 72 hours preoperatively.

Peri-anesthetic period

Patient was guided cautiously into the O.R. reassured hence she is visually and mentally defective; this help in later fixation of small IV canula gently. Phenytoin 100mg IV given as premedication. Fluid hydration was given generously with D5w to guard against hypoglycemia and dehydration so consequent stasis and thrombosis. Her legs were elevated. Monitoring include pulse oximetry, noninvasive blood pressure and pericardial stethoscope. Induction of general anesthesia and endotracheal intubation achieved with fentanyl, propofol and rocuronium. Anesthesia maintained with halothane 0.5 – 1% and 100% O2/. Recovery went smooth; residual muscle relaxant reversed with atropine 1mg and neostigmine 2.5mg. Postoperatively she was encouraged to walk early. She was discharged with good uneventful postoperative period.

Discussion

Homocystinuria is a rare inborn error of amino acid metabolism with an incidence of 1:200000. It leads to the accumulation of the raised level of homocystine in blood, urine and connective tissue that may present with subluxation of the lens or glaucoma. It has been documented that elevated levels of homocystine is a strong risk for occlusive arterial disease and venous thromboses and impaired cognitive performance; so these patients are susceptible to thromboembolic complications during general anesthesia.[1,2] Mudd et al. found 25 postoperative thromboembolic occasion after 586 surgeries. Skeletal features suggestive of marfan-like syndrome with limitation of joint mobility and osteoporosis raise special concern during airway and position management.[1–3] Anemia of megaloblastic type was reported despite it is not a fixed feature with all types of homocystinuria; folic acid and pyridoxine help in its control. Hyperinsulinemia and hypoglycemic convulsions are also common.[3,4]

Aims and measures for safe Anesthesia

A. Reassurance and building relationship with such mentally retarded patient during preoperative visit well help handiling in the OR.

B. Reduction of thromboembolic incidence by:

a. Maintain good hydration by decrease fasting period and fluid preloud.

b. Vasodilatation achieved by anesthetic drug with vasodilating property (propofol, halothane….)

c. Maintain blood pressure.

d. Lower limbs elevation and stocking.

e. Early ambulation.

C. Aspirin used for reduction of platelets adhesiveness might be needed to be discontinued 72h. preoperatively according to the nature of surgery.

D. Convulsion prophylaxis. (e.g. phenytoin)

E. Prevent hypoglycemia with glucose containing fluids.

F. Anticipate difficult intubation and positioning.

In a summary

Safe anesthesia management requires pretreatment with acetylsalicylic acid, adequate hydration with glucose, and maintenance of good arterial blood pressure and peripheral vasodilatation. In addition, to prevent venous stasis, the patient should wear elastic stockings or pneumoboots during surgery and walk as soon as possible.[3–8]

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Conflicts of interest

Author declare that there is no conflict of interest.

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