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Is homocystinuria a real challenge for anesthetist? Are we making a difference?

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Sir,

Homocystinuria is characterized by impaired synthesis of cystathionine, decreased cysteine, and increased methionine and homocysteine due to cystathionine b-synthase deficiency. Weakened collagen in connective tissue due to decrease cysteine explains lax ligaments and ectopia lentis as being the common presentation in children. Vascular injury through oxidative mechanism and platelet aggregation leading to hypercoagulable state is another manifestation but usually in adults. Accumulated methionine causes increase insulin release resulting in hypoglycemia. These patients usually seek medical attention due to wide spectrum of the disease and are therefore at increased risk for venous thrombosis[1] and hypoglycemia.[2]

A 10-year-old school going girl presented with cyanosis of fingertips for a month. Further history and examination was unremarkable, suspecting homocystinuria its levels were advised, and results were much higher (350 μmol/L) than normal (5–12 μmol/L) confirming the diagnosis. She was started on protein-restricted diet along with aspirin, pyridoxine, folic acid, trimethylglycine, and Vitamin C supplements. Fortunately, she responded and homocysteine levels dropped down (99.8 μmol/L) with improvement of symptoms.

Within a month of diagnosis, she started complaining of decreased vision in both eyes. On examination, bilateral ectopia lentis was diagnosed, she was scheduled for lensectomy under general anesthesia. Although lensectomy is a day care procedure, after explaining perioperative risks, patient was electively admitted. Preoperative assessment was done, homocysteine levels were 83.76 μmol/L, and written informed consent was taken. Compression stockings were provided and subcutaneous enoxaparin 0.5 mg/kg/dose every 12 hourly was started. Intravenous line was maintained and ½ strength dextrose started and continued perioperatively. Blood glucose level was measured every 4 hourly and routine medications were continued.

In operating room, monitoring for electrocardiogram, noninvasive blood pressure, and oxygen saturation were applied. Intravenous induction with propofol 2 mg/kg, fentanyl 0.2 μg/kg, and atracurium 0.6 mg/kg was done. Anesthesia was maintained with a mixture of isoflurane in air:oxygen in 50:50 ratio to maintain a mixture of air-oxygen of 0.9. Nitrous oxide was avoided, it increases homocysteine levels by inhibiting methionine synthase and is associated with postoperative morbidity and mortality.[3] Intermittent pneumatic compression stockings were applied over both the legs with pressure of 40–50 mmHg. After completion of 21 min surgery, the patient was extubated and shifted to recovery room.

Patient was discharged on the 2nd postoperative day from hospital. She was doing well with improvement in vision and started going back to school after a week of surgery. Monthly follow-up with homocysteine level was advised. On her last follow-up in February 2017, levels were 4.76 μmol/L which were almost normal.

A perioperative multidisciplinary approach including pediatrician, anesthetist, surgeon, and parents is required for these patients to prevent complication and improves outcome. Preoperatively lower levels of homocystinuria <50 μmol/L should be targeted. Maintaining adequate hydration and providing standard antithrombotic measures perioperatively decreases the risk of thrombosis. Rapid recovery after anesthesia using short-acting agents, minimizing the surgical time, and encouraging for early mobilization postoperatively further reduces the complications.

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