Homocystinuria and Anesthesia

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SUMMARY

A 7 year old girl with homocystinuria is presented in this report she required general anesthesia for bilateral subluxed lenses.

Homocystinuria and general anesthesia was discussed. Some important points were reminded such as thromboembolism and hypoglycemia.

Key Words: Homocystinuria, General anesthesia


ÖZET

Bilateral sublükse lensleri nedeniyle öpere edilen, 7 yaşındaki bir kız çocuğa genel anestezî uygulaması sunulmuştur. Homosistinüri ile genel anestezi tanımlanmış, tromboemboli ve hipoglisemi gibi bazı önemli noktalara hân/autoloadılmıştır.

Anahtar Kelimeler: Homosistinüri, Genel anestezi

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Advances in diagnosis and therapy have resulted in the increased survival of patients affected by rare diseases. An example of such a disease is homocystinuria, first described by Field and associates in 1962 (1).

Homocystinuria (homocystinemia) is an inborn error of metabolism that is due to the failure of transsulfuration of the precursors of cysteine. It is an autosomal recessive disorder and its incidence is approximated 1/200000 (2). Clinical manifestations are osteoporosis of the long bones, mental retardation, hypoglycemia, dislocation of the lenses, lax ligaments, kyphoscoliosis, genu valgum, light-coloured hair, flat feet, malar rash and repeated venous and arterial thrombotic episodes (3,4). The diagnosis is usually confirmed by the presence of homocystine in the urine.

Disease may be prevent early diagnosis, dietary management, pyridoxine and folic acid administration (4).

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Patients with homocystinuria may require general anesthesia on lens extraction for ectopia lentis. Sudden occlusion of cerebral, pulmonary, renal and myocardial vessels in the perioperative period presents the anesthesiologist with a high risk of mortality and a great challenge in management.

Consequently, we present a case report of a patient with homocystinuria and we offer some suggestions that may be helpful in the anesthetic management of similar cases.

CASE REPORT

The patient was an 7-year-old, 25 kg girl and was admitted to the hospital with bilateral subluxed lenses and iridodenedesis. She has observed by our hospitals pediatric clinic because of mental and motor retardation when she was 1.5 years old. Presence of methionine and homocystine in her urine and blood, she was diagnosed homocystinuria. Pyridoxine, folic acid and a special diet was administered till now.

Only, II/VI pansystolic murmur was observed in her physical examination. There were no significant cardiopulmonary or renal problems. Her platelet count was 378000/mm³, Hb 13.5 gr/dl, Htc 41.3%, prothrombin time 13 seconds, blood glucose 111 mg/dl. PA chest x-ray and other lobarotry findings were normal.
She was accepted to operating room for pars plana lens extraction. She was administered acetylsalicylic acid (100 mg/daily) four days before surgery. The patient was premedicated with atropine 0.25 mg intra-muscularly. Anesthesia was induced with propofol (2 mg/kg), vecuronium (0.1 mg/kg) and 6 mm non-cuffed orotracheal tube was used for intubation.

Anesthesia was maintained with 6 mg/kg/hr propofol Infusion and 50% N2O/O2. 0.25 mg bolus doses of alfentanil was repeated in every 20 minutes. Dextran 40 (200 ml) and 5% dextrose (200 ml) was used during 1 hour and 30 minutes procedure.

Her legs were wrapped in elastic bandages. EKG, O2 saturation and blood pressure were monitored. Her blood glucose levels were measured 3 times during the procedure and were found to be within normal values.

At the end of the procedure vecuronium was reversed with atropin 0.25 mg and prostigmin 0.5 mg. Then the patient was extubated. Her anesthetic course was uneventful. She was observed closely for signs of pulmonary emboli on postoperative days.

The patient was operated from her other eye one month later using with the same anesthetic technic and no complications were observed. She was discharged home on the seventh postoperative day.

**DISCUSSION**

Patients with homocystinuria may die at an early age due to the thromboembolic phenomena. Carson et al presented ten cases who between them suffered more than ten thrombo-embolic episodes, there was a fatal outcome in three cases (5). Henkind et al reported two postoperative deaths following ocular surgery because of thromboembolism (1). So anesthesiologist must prevent the patient from thromboembolism. Careful attention must be paid to the intraoperative administration of fluids (6). Although Crooke et al and Graver et al have recommended the use of dextran 70 (1,6). Parris et al have recommended dextran 40 (4). We use dextran 40 because it diminishes platelet adhesiveness.

Prevention from hypoglycemia, dextrose solutions should be administered (4,6). Elastic bandages or stockings must be wrapped to the legs for prevention from blood stasis. McDonald et al have demonstrated increased platelet adhesiveness in affected patients (1). Dipyridamole or acetylsalicylic acid can be used for prevention from thromboembolism (3). Postoperative early ambulation must be provided.

Although general anesthesia can induce thromboembolism, there is no anesthetic drug indicated or contraindicated. Some reports advice that narcotic analgesics should not be administered because of respiratory depressant effects that could delay early postoperative ambulation (4).

Meanwhile we used a new short acting narcotic agent alfentanil, we didn't observe any respiratory depression or delayed recovery period.

Inhalation anesthetics was used for operations in literature (1,4,6,7). We used propofol infusion and alfentanil intravenously which were new Intravenous anesthetic agents. These are short acting drugs so they provide rapid recovery and early ambulation.

Propofol is a potent hypnotic, currently formulated as an aqueous emulsion of soybean oil (8). Its pharmacokinetic profile suggested that anesthesia could be induced and maintained by a continuous infusion of the drug. The advantage is the rapid, smooth postoperative recovery which is free of nausea and vomiting (9).

Alfentanil is a useful synthetic narcotic. It may have many applications as an analgesic in surgical procedures. Important positive points include its rapid onset of action, its moderate effects on hemodynamic stability and its rapid elimination, generally allow in for good recovery (10). Following anesthesia all patients should be observed 48 hours closely.

In summary special attention must be paid to minimize the risks. Maintenance of high cardiac output, reduction of blood viscosity, reduction of vascular resistance, improvement of periferal perfusion, good venous return, avoidance from dehydration, stress and hypoglycemia, rapid recovery from anesthesia, providing early postoperative ambulation are the important points for anesthesia to the patient with homocystinuria.

**REFERENCES**