# Homocystinuria and Anesthesia

*HOMOSİSTİNÜRİ VE ANESTEZİ*

Ömür ERÇELEN*, Nursel KARAKELLE", Bilge ÇELEBİOĞLU*, Ülkü AYPAR"*, Kemal ERDEM

* Assist.Prof.Anesthesiology and Reanimation, Hacettepe University Faculty of Medicine,
** Resid.Anesthesiology and Reanimation, Hacettepe University Faculty of Medicine,
*** Prof.Anesthesiology and Reanimation, Hacettepe University Faculty of Medicine, Ankara, TURKEY

**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 hypogiicemia.***

Key Words: Homocystinuria, General anesthesia

T Klin J Med Sci 1995, 15: 357-358

**ÖZET**

***Bilateral sublükse lensleri nedeniyle öpere edilen, 7 yaşındaki bir kız çocuğuna genel anestezi uygulaması sunulmuştur. Homosistinüri ile genel anestezi tartışılmış, tromboemboli ve hipoglisemi gibi bazı önemli noktalar hatırlatılmıştır.***

Anahtar Kelimeler: Homosistinüri, Genel anestezi

T Klin Tıp Bilimleri 1995, 15: 357-358

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 ressesive disorder and its incidence is aproximately 1/200000 (2). Clinical manifestations are osteoporozis of the long bones, mental retardation, hypoglycemia, dislocation of the lenses, lax ligaments, kyphoscoliosis, genu valgum, light-coloured hair, flat feet, malar rush 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).

Geliş Tarihi: 06.02.1995

Yazışma Adresi: Ömür ERÇELEN
Hacettepe Üniversitesi Tıp Fakültesi
Anesteziyoloji ve Reanimasyon ABD
ANKARA

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 iridodenesis. She has observed by our hospitals pediatrics 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/mm$^3$, 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 acetylsalicylicacid (100 mg/daily) four days before surgery. The patient was premedicated with atropine 0.25 mg intramuscularly. 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 glucouse levels were measured 3 times during the procedure and were founded between 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 statis. 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 aplications 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

1. Crooke J, Towers J, Taylor W. Management of patients with homocystinuria requiring surgery under general anesthesia. Br J Anaesth 1971; 43:96.
2. Rezvani I, Auerbach V. Metabolic diseases. In: Nelson WE, ed. Textbook ot Pediatrics. 14* ed. Philadelphia: WB Saunders, 1992:312-4.
3. Brown B, Walson P, Taussig L. Congenital metabolic diseases of pediatric patients: Anesthetic implications. Anesthesiology 1975; 43:197.
4. Parris W, Quimby C. Anesthetic considerations for the patient with homocystinuria. Anesth Analg 1982; 61:708.
5. Carson N, Dent C, Field C, Gaull G. Homocystinuria: clinical and pathological review of ten cases. J Pediat 1965; 66:565.
6. Grover V, Malhotra S, Kaushik S. Anesthesia and homocystinuria. Anaesthesia 1979; 34:913.
7. Frost P. Anaesthesia and homocystinuria. Anaesthesia 1980; 35:918.
8. Claeys M, Gepts E, Camu F. Haemodynamic changes during anesthesia induced and maintained with propofol. Br J Anaesth 1988; 60:3.
9. Victory R, Pace M, Anaes F, White P. Propofol: an update. Anesth Cl of North Am 1993; 11:831.
10. Larijani G, Goldberg M. Alfentanil hydrochloride. A new short acting narcotic analgesic for surgical procedures. Clinical Pharmacy 1987; 6:275.