"Heterozygous MTHFR 677C-t Mutation and Intermediate Hyperhomocysteinemia Associated with Hyphema and Renal Vein Thrombosis"

"Heterozigot MTHFR 677C-t Mutasyonu ve Orta Derecede Hiperhomosisteinemi ile Hifema ve Renal Ven Trombozu Birlikteliği"

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ABSTRACT

Hyphema, an accumulation of red blood cells within the anterior chamber, is a benign condition and expected to disappear within a few days. Hyphema have been reported in the neonatal period mostly after spontaneous vaginal delivery. Thrombosis is the formation of a blood clot inside a blood vessel, obstructing the flow of blood through the circulatory system. Renal vein thrombosis (RVT) is a serious proceses which might lead to either renal failure or to secondary thromboembolic conditions. Herein, a case of a newborn, who has a hyphema and renal vein thrombosis together with heterozygous MTHFR 677C-t mutation and intermediate hyperhomocysteinemia, is presented.

Key Words: Hyphema, thrombosis, newborn.

ÖZ

Hifema gözün ön kamerasında kırmızı kan hücrelerinin birikmesidir ve birkaç gün içinde iyileşmesi beklenen benign bir durumdur. Hifema yenidoğan döneminde çoğunlukla normal spontan vajinal yolla doğum sonrası bildirilmiştir. Tromboz kan akışını engelleyen, kan damarı içinde kan pıhtısının oluşmasıdır. Renal ven trombozu (RVT) böbrek yetmezliğine ya da ikincil tromboembolik olaylara sebep olabilen ciddi bir durumdur. Burada orta derecede hiperhomosisteinemiye eşlik eden hifema ve renal ven trombozu olan bir yenidoğan olgusu sunulmuştur.

Anahtar Sözcükler: Hifema, trombozis, yenidoğan.

INTRODUCTION

Hyphema is the presence of blood in the anterior chamber of the eye and mostly associated with penetrating or blunt ocular injuries.[1] Prothrombotic factors leading to thrombosis and venous stasis may simplify hyphema formation. Few cases of hyphema have been reported in the newborn period mostly after instrumental or spontaneous vaginal delivery.[2] There is no neonatal hyphema case after cesarean delivery in the literature. Renal vein thrombosis is a multifactorial disease that predominantly affects newborn infant. Dehydration, polycythemia, cyanotic congenital heart disease, sepsis, maternal diabetes mellitus, shock, asphyxia, umbilical venous catheterization, and inherited prothrombotic abnormalities may also have a role in the pathogenesis of neonatal RVT.[4]

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CASE REPORT

A 3100-g female infant was delivered by cesarean section after prolonged labor at 40 weeks of gestation. Due to depressed delivery, resuscitation after birth was applied. APGAR scores were 2 and 5 at 1th and 5th minutes, respectively. She was intubated and admitted to neonatal intensive care unit. On admission, she had poor perfusion, cyanosis, respiratory depression. pH obtained at postnatal 15th minute was pH 7.21. On physical examination she had caput succedaneum but no evidence of subconjunctival hemorrhage or ecchymosis on the face and eye lids. Blood was noted in the anterior chamber of the left eye and ophthalmology consultation was requested. Initial examination of the ophthalmologist revealed a normal anterior segment of the right eye but a total hyphema of the left eye. Fundus details of the left eye were obscured because of anterior chamber hemorrhage (Fig. 1). Intraocular pressure (IOP) measured via Tonopen-Avia was 10 mmHg and 22 mmHg in the right and left eye, respectively. Eye drops including betoxolol 0.25 mg twice a day, prednisolone acetate 1% 4 times a day, cyclopentolate 0.5% twice a day were initiated. Ophthalmologic examination on the second day of life revealed a further increase in hyphema and IOP (30 mmHg) in the left eye. The examination of the right eye was unchanged from baseline. The B-mod ultrasonography of the left eye was normal without evidence of vitreous hemorrhage and retinal detachment. An anterior chamber wash-out was not considered by the ophthalmologist because of poor general health of the neonate. Coagulation profile including thrombocyte count, prothrombin, thrombin, and partial thromboplastin and, bleeding times, and fibrinogen level were all normal. Hematuria was detected with urinalysis on the second day. Renal ultrasonography revealed left RVT. Renal functions were normal and she had no systemic symptom associated with RVT. She was treated with conservative treatment for RVT.

Feeding with orogastric catheter was started on the second day. Nasal CPAP therapy was initiated on the fourth day. On the sixth day, full enteral feeding was used to feed and she tolerated. She did not need oxygen after the 12th day.

The infant was heterozygous for MTHFR 677C-t mutation. Serum levels of Vitamin B12 and folate were normal. Serum homocystein level was 32 μmol/L (N: 5.5–17 μmol/L). The mother was also heterozygous for MTHFR 677C-t mutation.

On follow up, the hyphema on the left eye cleared gradually starting from the first week and totally disappeared at the third week. The dilated fundus examination was normal at that time. She was discharged on 22th day. Examination performed one month after birth revealed a normal anterior segment and fundus examination bilaterally (Fig. 2). The hyphema cleared completely with the conservative ophthalmological management without any complication.

DISCUSSION

The majority of hyphemas are traumatic and mostly result from ocular trauma secondary to tears in the ciliary body and/or iris root.[1] Spontaneous hyphema without a history of trauma may occur in vascular and neoplastic lesions of the iris, and clotting abnormalities.[3] In early infancy spontaneous hyphema have been reported in association with iris hemangioma in diffuse neonatal hemangiomatosis, juvenile xanthogranuloma, retinoblastoma, and various hematological disorders including hemophilias, disseminated intravascular coagulation and acute lymphoblastic leukemia.[5,8] In the present case, routine blood tests and detailed ophthalmological examination including anterior segment, and dilated fundus examination, and ocular ultrasonography were performed and none of those causes was responsible for the hyphema in our case.

Hyphema presenting at birth is very rare and suggests birth trauma as the underlying cause. Hyphema is an uncommon type of eye trauma in neonates and the mechanism by which anterior chamber hemorrhage occurs is mostly unknown. Forceps use during vaginal delivery may cause ocular trauma. Paciuc et al. Reported hyphema together with vitreous hemorrhage in full-term newborn girl with face presentation delivered vaginally after prolonged labor. They stated that

Figure 1. *Total hyphema of the left eye just after birth.*

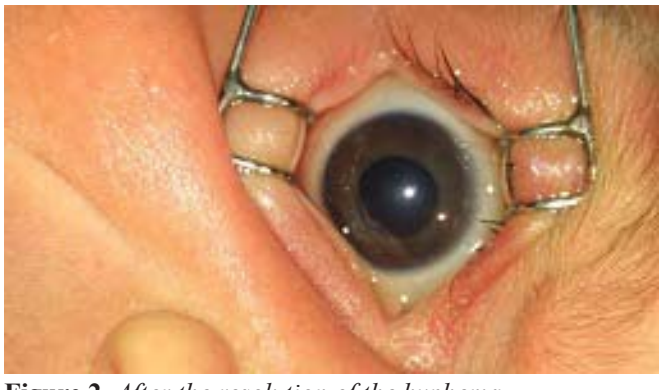

Figure 2. *After the resolution of the hyphema.*

increased venous pressure and stasis during prolonged labor, plus the trauma of face presentation could be causative.[9] Misra et al. reported a case of neonatal hyphema associated with precipitous vaginal delivery induced with dinoprostone and postulated that modified venous pressure may predispose to hyphema.[10] Infants delivered by cesarean section reportedly do not have hyphema. This is the first report of neonatal hyphema presenting at birth after cesarean delivery. Prolonged labor seems to be a predisposing factor in our case. Increased venous pressure in iris vessels during prolonged engagement of the head in the pelvis may be the underlying mechanism.

Renal vein thrombosis may also be associated with prolonged labour, low APGAR scores and resuscitation. Our patient had heterozygous MTHFR 677C-t mutation and intermediate hyperhomocysteine level. We consider that the coexistence of this mutation in addition to prolonged labor may contribute to formation of thrombosis leading to both RVT and hyphema. Thrombosis of iris vessels and venous stasis may be the mechanisms of hyphema in our patient.

Hyphema is usually a benign condition that resolves quickly without serious sequelae. However in some cases increased intraocular pressure or recurrent bleeding can be observed, which can lead to loss of vision. Close follow-up with proper medical and surgical therapies is essential to prevent visual loss.[11] Diagnosis can be missed when there is no external sign of trauma around the face and eye lids.

CONCLUSION

Careful inspection of the eyes should be a part of initial examination of the newborn infant especially when predisposing factors for ocular trauma is present. Prolonged labor even followed by cesarean section can be one of the risk factors for neonatal hyphema. Prothrombotic factors should be kept in mind in patients with hyphema.

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