

SUBGALEAL BLEED AND BILATERAL PROPTOSIS AS AN INITIAL MANIFESTATION OF MILD HEMOPHILIA A IN A CHILD.

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ABSTRACT

A nine year old boy presented with bilateral protrusion of eyes and gross decrease in visual acuity, and was evaluated for a systemic etiology. Radioimaging revealed the presence of subgaleal hematoma and bilateral subperiosteal haemorrhage. In the course of his stay in hospital, he had worsening proptosis, and deteriorating vision due to corneal decompensation and optic nerve ischaemia. After a battery of investigations, a diagnosis of mild Haemophilia A was

made. Subgaleal bleed with subperiosteal haemorrhage in a male child should raise the suspicion of mild Hemophilia A. A high index of suspicion, early diagnosis and appropriate therapy would prevent dismal visual prognosis in these children.

Key words: Mild Hemophilia A, proptosis, subgaleal hematoma, subperiosteal haemorrhage.

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INTRODUCTION

Subperiosteal haemorrhage can result from trauma, blood dyscrasias and haematological malignancies. Patients usually present with proptosis of acute onset, visual dysfunction and ophthalmoplegia.^[1] Hemarthrosis is usually the presenting feature in Hemophilia A and there is almost always a history of preceding trauma. While subperiosteal haemorrhage as a presenting sign of Hemophilia A has been rarely reported, a combination of subgaleal bleed and subperiosteal haemorrhage as presenting features of mild Hemophilia A has not been reported, to the best of our knowledge. Subperiosteal haemorrhage follows subgaleal bleed and is a poor prognostic indicator, and when accompanied by ophthalmoplegia, compressive neuropathy and corneal exposure, poses a surgical dilemma in the absence of a definitive diagnosis of a bleeding disorder.

CASE REPORT

A 9 year old boy presented with bilateral proptosis of recent onset, associated with diminution of vision and frontal headache. Child had an episode of trivial injury of slip and fall while playing at school, one month ago, following which he had no complaints related to the trauma. There was no history of pain, fever, similar episodes in the past, swelling of knees or any other bleeding tendencies. He was the elder of two siblings, born of a third degree consanguinous marriage, and had achieved developmental milestones appropriate for age, and maintained a good scholastic performance.

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He had a visual acuity of counting fingers close to face in the right eye, and 2/60 on the left eye. Ocular examination revealed bilateral severe proptosis, periocular ecchymosis, corneal xerosis with peripheral corneal guttering, and total external ophthalmoplegia [Fig.1]. A relative afferent pupillary defect (RAPD) was demonstrated in the right eye. Fundus of the right eye was not visualised, whereas left fundus was normal.



Fig.1: Clinical picture of the male child with bilateral extensive proptosis and severe exposure keratitis

Magnetic Resonance Imaging (MRI) brain showed extensive subgaleal and superior subperiosteal bleed [Fig. 2]. Simultaneous systemic evaluation was done, which revealed a near normal coagulation profile and haematological assay (platelet count -3.40, PT-15.9, aPTT -37.2) initially. Normal bone marrow study and ultrasonogram abdomen, ruled out acute myeloid leukemia and neuroblastoma respectively. Cerebral angiogram showed no evidence of arteriovenous malformations. Frequent instillation of topical lubricants and antibiotics was done to prevent corneal decompensation.

Inferring from the initial normal coagulation profile,

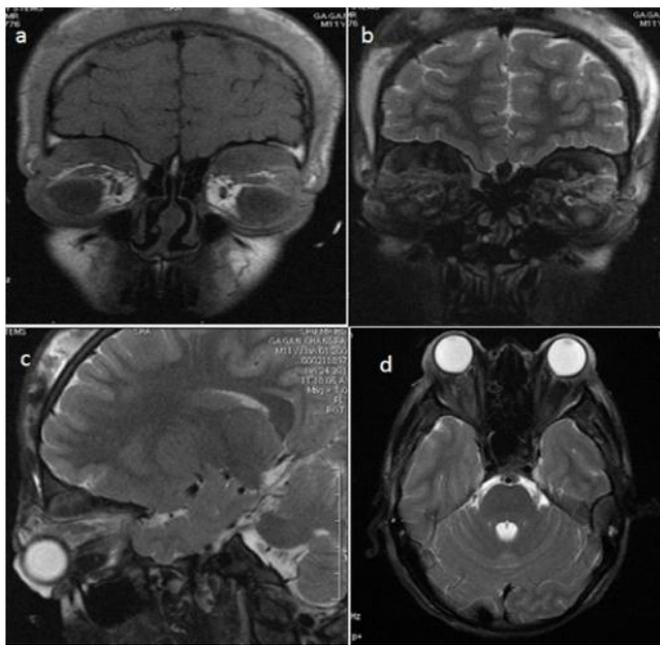


Fig.2 :(a,b) Coronal T1 T2 weighted images reveal subgaleal haemorrhage and bilateral relatively symmetrical areas of subperiosteal haemorrhages involving superior aspect of orbit. (c) Sagittal T2 weighted image showing extensive proptosis with adjacent extraocular muscles, and optic nerve displaced inferiorly. (d) Axial T2 weighted image reveals subperiosteal haemorrhages with diffuse scalp haemorrhage.

the subgaleal bleed and the subperiosteal haemorrhage were thought to result from spontaneous venous bleed. Patient had increased orbital pressure and resultant orbital compartment syndrome as evidenced by bilaterally stretched optic nerves in MRI and RAPD in the right eye, along with constant corneal exposure and progressive corneal decompensation. Hence, after obtaining informed consent from parents, bilateral lateral canthotomy and subperiosteal drainage were performed under general anaesthesia. 40 cc of subperiosteal altered blood from the right side, and 30 cc from the left side were aspirated. Approximately 1cm of the lateral orbital margin was removed on both sides, involving the frontozygomatic suture to decompress the orbit and provide space for the regression of proptosis.

Post orbital decompression, the orbital pressure reduced remarkably and visual acuity improved to 6/60 (right eye) and 2/60 (left eye), pupillary reactions were restored, along with minimal improvement of extraocular movements. Unfortunately, the corneal decompensation progressed and bilateral amniotic membrane graft with cyanoacrylate glue application under general anaesthesia was done to prevent corneal perforation.

Despite aggressive ocular therapy, child developed bilateral exudates in the anterior chamber with peripheral ring infiltrates in the left eye. Microbiological studies of the corneal scrapings, showed no growth of organism. Topical antibiotic therapy with 2 hourly instillation of Besifloxacin 0.6% was initiated. Bilateral Bscan

ultrasonography and Visually evoked Potential (VEP) showed normal study. Informed consent was obtained, and bilateral lateral tarsorrhaphy was done under general anaesthesia, to arrest the deterioration of the corneal status.

In view of persisting subgaleal bleed despite repeated blood transfusions and fresh frozen plasma infusions, child was suspected to have a haematological disorder. Repeat blood investigations revealed an abnormal Activated Partial Thromboplastin Time (aPTT - 47.2 seconds) value and low Factor VIII (11.0%) assay, confirming the diagnosis of Mild Hemophilia A. The delay in diagnosis was attributed to the initial near normal coagulation profile. Despite best efforts by a multidisciplinary team, child developed right corneal perforation and lost perception of light in the right eye. [Fig.3a] Left eye had resultant central corneal opacity with healed peripheral gutter, and maintained at 2/60. [Fig. 3b]

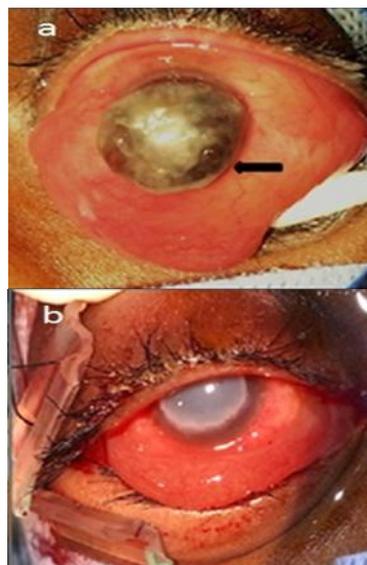


Fig.3: (a) Right eye demonstrating severe exposure keratitis and corneal perforation (arrow). (b) Left eye with central corneal opacity and healed peripheral gutter.

After replacement therapy with Factor VIII, the proptosis regressed after 2 weeks. [Fig.4] With the



Fig.4: Resolving proptosis post bilateral lateral tarsorrhaphy

consent of the parents, patient underwent right corneoscleral graft with Vitrectomy and Ahmed glaucoma valve implantation (due to a high probability of

trabeculectomy failure), followed by left Penetrating Keratoplasty, under general anaesthesia.^[8] Postoperatively, he had a visual acuity of counting fingers at 1 metre right eye, with residual vitreous haemorrhage, and 6/18 left eye respectively. [Fig. 5]

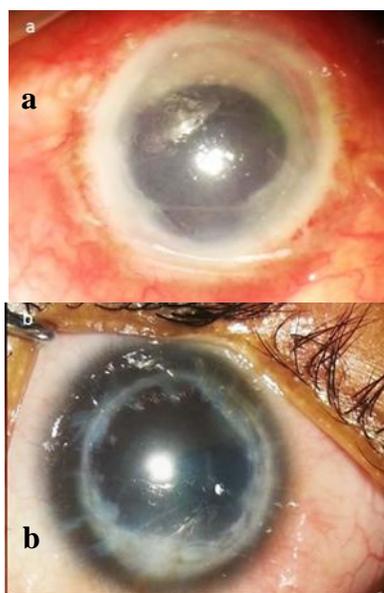


Fig.5: Postoperative picture of right (a) and left eye (b) respectively following right anterior vitrectomy and sclerocorneal graft, and left penetrating keratoplasty.

DISCUSSION

Hemophilia A is an X linked, recessive disorder caused by deficiency of Factor VIII (F VIII), which may be inherited or arise from spontaneous mutation. The coagulation cascade comprises of an intrinsic and extrinsic pathway which aids in the formation of a stable fibrin clot at the site of injury. Factors VIII and IX when activated, cooperate to cleave and activate factor X, which is vital for conversion of fibrinogen to fibrin.^[2] Hemophilia is classified based on the plasma procoagulant level as severe (F VIII < 1% of normal), moderate (1 - 5% of normal) and mild (5 - 40% of normal).^[3] Severe hemophilia presents in children younger than 1 year with spontaneous bleeding whereas mild Hemophilia presents in children older than 2 years with bleeding only after significant trauma or surgery. Activated partial thromboplastin time (aPTT) is usually significantly prolonged in severe cases, but may be near normal in mild to moderate hemophilia.

Proptosis is usually a late occurrence in cases of subgaleal haematomas, the reason being the anatomical configuration of the orbital septum, which acts as a barrier between the facial and orbital structures. The orbital septum extends from the periosteum to the upper eyelid, blending anteriorly with the levator aponeurosis. This continuity is lost at the lateral canthal region, creating a communication between the subgaleal space and the periorbital space.^[4]

Subperiosteal bleed could be spontaneous or could result from trauma, bleeding diathesis, vascular diseases,

lymphangiomas and cavernous hemangiomas.^[5] Mild hemophilia A should be thought to be the causal factor in a male child who presents with subgaleal bleed and proptosis arising from subperiosteal haemorrhage. Factor XIII deficiency^[4] and Christmas disease (Factor IX deficiency)^[6] are known to have produced subgaleal and orbital hematoma. Guirgis et al have reported subperiosteal haematoma as an initial manifestation of Christmas disease.^[6] Poor vision at presentation, RAPD, external ophthalmoplegia and exposure keratopathy at presentation are all poor prognostic indicators for visual recovery. In the absence of subgaleal bleed, prompt replacement therapy with Factor VIII and supportive therapy can cause resolution of proptosis. However factor substitution therapy could be ineffective in patients with autoantibodies to Factor VIII.^[7] Orbital decompression needs to be resorted to when there is an orbital compartment syndrome.^[4]

A high index of suspicion, appropriate haematological assay and prompt replacement of the deficient factors can prevent adverse visual prognosis in children who present with subgaleal and bilateral subperiosteal haemorrhage.

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