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Case Study

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CASE REPORTS ON SEVERE HEMOPHILIA A PRESENTING IN A NEWBORN WITH GERMINAL MATRIX HEMORRHAGE WITH HEMATOMA IN LIVER AND HEMIPERITONEUM

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ABSTRACT

Hemophilia A is an X-linked hereditary condition that leads to decreased factor VIII activity. Subsequently, leading to increased risk of bleeding events. During neonatal period, diagnosis is made after post-partum complication or unexpected bleeding after medical procedure. Germinal matrix hemorrhage during neonatal period is rare, severe intracranial bleeding with high mortality. We present a neonate with unremarkable family history and uneventful pregnancy with a vaginal delivery with no instrumentation, presenting with progressive pallor and shock at 36hours of life. The unexpected pallor and shock lead to a coagulation study and hemophilia A was diagnosed. This case shows the rare presentation of hemophilia A as intracranial bleeding with liver hematoma and hemiperitoneum.

KEYWORDS: Factor VIII level, Hemophilia A, neonates.

INTRODUCTION

Hemophilia is a hereditary coagulopathy linked to X-chromosome which mainly affects male individuals.[1] It can be either hemophilia A & B depending whether there is a decrease of Factor VIII or IX. It has an incidence of 1 per 5000 male birth.[2] Hemophilia A is the most common type accounting for 85% of the cases.[2]

Hemophilia A is an X-linked recessive blood disorder characterized by decreased level of coagulation factor VIII activity, disrupting the normal propagation of the intrinsic coagulation cascade. FVIII is a cofactor for clotting protein factor IX, and together, they form a complex that activates factor X.[3] Normal plasma levels of FVIII range from 50% to 150% (0.5-1.5U/mL). Hemophilia A is categorized according to the amount of FVIII levels present: mild (5% to 50%), moderate (1% to 5%) and severe (<1%, or <0.01U/mL).[3] Newborns with hemophilia are at increased risk of intracranial hemorrhage and extra cranial hemorrhage and its sequelae.[4-7] The prevention, detection, and treatment of these complications are foremost importance during and after delivery.

When there is known family history, usually diagnosis is made during the neonatal period.[8] Neonatal hemophilia presentation is usually either related to a traumatic labor or to iatrogenic bleeding during heel stab sampling, venous puncture or vitamin k administration.[9] Rarely neonatal hemophilia presents with severe bleeding.[10] Better treatment strategies centered on recombinant factors replacement lead to a considerable decrease in mortality rates for hemophilia in last year's, with recent studies showing a nearly normal life expectancy even in severe hemophilia.[11,12]

In this case report, we describe a newborn who sustain germinal matrix hemorrhage with hematoma in liver and hemiperitoneum. The aim of this report is to illustrate the acute potentially life threatening clinical spectrum of hemophilia A in neonates. Up to 30% of patients with hemophilia A have a negative family history[13], and even those with severe hemophilia have a median age of diagnosis of 5.8months. Therefore, it is important that neonatal care providers are cognizant of the different presentation of hemophilia A to prevent mortality and minimize morbidity.

CASE REPORT

A term male neonate was born vaginally to a 24 year old primigravida with no history of bleeding disorder in family. The pregnancy was uneventful. The infant's APGARs were 8 at 1min and 9 at 5min, with no complication. Birth weight of 2700 grams, length 48cm and head circumference 35cm. The initial physical examination in the delivery room was unremarkable and

he was transferred with his mother to obstetrics ward for regular neonatal care. Vitamin k was administered at birth and breastfeeding was initiated. At 36 hours of life he was urgently assessed for increasing pallor and poor feeding. At presentation the neonate was lethargic, with poor perfusion, heart rate of 145b/min, MAP = 30mmhg.

Laboratory workup revealed hemoglobin of 7.7gm/dl, white blood cell 25.80k/mm^3 with platelet count 153k/mm^3. SGPT was 203u/L and SGOT was 480u/L. The infant blood group was A positive. In view of low hemoglobin coagulation profile was sent which revealed Prothrombin time >60s and APTT time > 120s. Subsequently, due to concerns of bleeding diathesis labs were sent for coagulation studies of FVIII activity, FIX activity, von Willebrand assay. He was transferred to neonatal intensive care unit and was given cryoprecipitate for the presumed diagnosis of hemophilia while reports were awaited.

Lab work showed FVIII level <1%, FIX activity 10%, von willebrand level 77%. Once FVIII deficiency was established, recombinant factor VIII was provided to the neonate.

A bedside Neurosonogram demonstrated a hyper echoic area in the right caudothalamic groove with internal echoes in the lateral ventricle suggestive of grade III germinal matrix hemorrhage. Abdominal ultrasound revealed a hematoma in segment VI and VII of liver and hemiperitoneum.

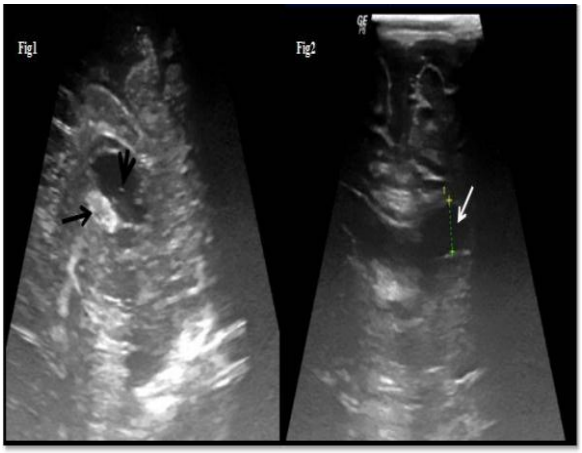

Fig. 1 & Fig. 2 are the parasagittal and coronal sections of cranial sonogram respectively. There is hyper echoic area seen in the right caudothalamic groove (black arrow in fig1) with internal echoes in the lateral ventricle (arrowhead in fig1). The frontal horns of the bilateral lateral ventricles are also dilated (white arrow in fig2). These findings are suggestive of grade 3 germinal matrix hemorrhage.

DISCUSSION

In any child with bleeding symptoms, initial evaluation should include a complete blood count, prothrombin time, and activated partial thromboplastin time. Testing factor levels, platelet function, and fibrinogen can also be considered based on the initial lab testing.

Assessing family history of bleeding disorders is crucial for rapid diagnosis, as 50% of all males born to HA-carrying mothers will inherit the disorder. Infants born to known carriers are usually diagnosed early in the neonatal period; however, up to 30% of newborns with this condition may have a negative family history. Ideally, infants who are known to be at risk for hemophilia should have cord blood sent for diagnosis and should avoid intramuscular vitamin K administration.[35] Prophylactic therapy for children with the severe form of the disease has improved prognosis over the years.

This case shows a germinal matrix hemorrhage with liver hematoma wand hemiperitoneum as a presentation of a severe hemophilia. Despite hemophilia being a known risk factor for cranial bleeding, we found no record of a similar presentation after an uncomplicated vaginal delivery and with no traumatic history or family history of coagulopathy. Our patient presented with pallor, lethargy and shock, the most common presentation in hemophilic patient with cranial bleeding.[14] We focused our approach on aggressive correction of hypovolemic shock with saline bolus and cryoprecipitate, according to the existing literature.[15]

CONCLUSIONS

After reviewing existing literature we concluded that our patient had a rare presentation of hemophilia. He presented a germinal matrix hemorrhage with liver

hematoma after an uncomplicated vaginal delivery. Aggressive supportive treatment was effective in controlling the initial bleeding. Hemophilia A was diagnosed and factor VIII was given. This case report reminds that hemophilia can present in several different ways. When facing a significant bleeding in a neonate with no risk factors, a blood coagulation disorder must be thought along with immediate aggressive supportive measures. An early diagnosis allows to an early focused treatment and better outcome.

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