Case Report

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An Unexpected Detection of Hemophilia in an Infant

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Abstract

Hemophilia A is usually diagnosed once the infant starts cruising. This makes it challenging to diagnose the condition in early infancy. We report a case of 4 months old male child who presented with resolvable swellings on the trunk. Patient had family history of bilateral painful knee swellings in his maternal uncle. Thus patient was suspected to have a bleeding disorder which was confirmed by laboratory testing of coagulation profile and factor VIII levels.

Keywords: Hemophilia A; Infant; Swelling.

Introduction

Hemophilia is a coagulation factor deficiency. Hemophilia A affects 1 in 5000 male birth. It is inherited in an X linked recessive manner, thus seen more commonly in males.[1] It may affect 1/3rd of the patient with no family history suggesting sporadic mutation. Symptoms of Hemophilia depend upon the levels of factor VIII.[2]. Here in, we report a rare case of Hemophilia diagnosed in early infancy period.

Case Report

A 4 months old, male child born out of non-consanguineous marriage was brought to the hospital with chief complaints of pea sized swellings on trunk since 8 days. They were self resolving, not associated with pain, itching. There was no history of trauma, swelling anywhere else in the body. There was no history of bleeding from any other site, no joint pain or bleeding from vaccination sites. He did not have any history of recurrent infection or fever. He was exclusively breastfed. There were no similar complaints in the past. Patient was born full term to a primigravida mother, appropriate for gestational age, via caesarean section in view of non-progression of labour. Neonatal period was uneventful. On general examination child was active. There was no pallor, cyanosis, swellings of joints or muscles. Patient had no other associated complaints. There was no history of bleeding episodes after minor trauma, skin bruises. He was developmentally normal. His weight was 6kgs, length was 65 cm and head circumference was 42 cm. His heart rate was 120/min, respiratory rate was 34/min, peripheral pulses were well felt and spo2 was 96%. On cardiovascular system examination–heart sounds were normal. No murmur was heard. Other systemic examinations were normal. There were two swelling of size 2x2 cm on the right lower chest wall. There were no abnormalities of the overlying skin. It was a round, non tender, smooth swelling which was freely mobile and not pulsating. It was non fluctuant with

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normal temperature and negative transillumination test. On repeatedly asking direct questions mother gave history of swelling of bilateral knees in her brother (maternal uncle of the case).

His complete blood count was suggestive of Haemoglobin- 12gm/dl, platelet 3.2 lakhs and total leukocyte count of 7500. Bleeding time, prothrombin time were normal. Activated Partial Thromboplastin Time (APTT) was prolonged (45 seconds). It was suggestive of coagulation abnormality of the indirect pathway. USG local site showed hypoechoic lesions in subcutaneous plane with central vascularity suggestive of vasculitis or some blood collection. The swelling got disappeared after 2 days without any intervention. Patient's factor VIII levels were 1% which was suggestive of moderate haemophilia. Thus patient was diagnosed to be a case of Hemophilia A. Patient was referred to local haemophilia society for further management.

Discussion

Hemophilia is a coagulopathy involving the deficiency of factor VIII, IX or XI. Hemophilia A is an X linked recessive disorder. Mansouritorghabeh H et al.[2] found out that in patients with Hemophilia A bleeding manifestations were depending upon the levels of factor VIII and they range from fatal spontaneous brain bleed to ecchymosis of the skin. Severe hemophilia cases may have spontaneous bleeding after minor trauma in the form of hemarthrosis and intramuscular bleeding. In moderate cases patients experience bleeding after mild to moderate injuries. In mild cases patient may be undiagnosed for years and may bleed heavily during surgery or major trauma. In paediatric age group 70% cases with positive family history are diagnosed at birth or after first bleeding episode. Patients with negative family history are usually diagnosed in conditions like bleeding post circumcision, post injection or vaccine, easy bruising after minor trauma, ecchymosis in buttocks at the beginning of crawling or walking. Frequent falls or impacts from furniture while learning to ambulate can induce extensive soft tissue contusions and hemorrhage that can mimic the appearance of child abuse in the young hemophilic.[2-3] Similarly in our case the patient started swelling after he achieved rolling over at 4 months of age.

The hallmark of severe hemophilia is hemarthrosis. It presents with symptoms like warm tingling sensation. It further causes inflammation of joint capsule with joint movement restriction. Inflamed joints are warm to touch. In chronic cases damage to cartilage may be seen. Muscle bleeding is one of the features of severe Hemophilia A. It occurs following minor trauma or spontaneously or due to emotional stress. Its severity depends upon severity of hemophilia, type of muscle and fascia involved. Muscle spasm associated with the hematoma may cause joint movement restriction. Hemophilia patients may have life threatening CNS bleeds. Diagnosis of Hemophilia A mainly involves laboratory testing.[2-4] The coagulation profile, specifically APTT may be prolonged if factor VIII levels < 0.3 IU/ml. In some cases APTT may be normal due to different sensitivities of reagents to factor VIII levels. Hence normal APTT time does not rule out Hemophilia A. Thus even if APTT is normal, but Hemophilia A is clinically suspected, factor VIII assay should be done.[3] Severity of Hemophilia A depends upon levels of factor VIII. Severe form is defined as <1 % of factor VIII activity. Moderate form is defined as 1-5 % of factor VIII activity. Mild cases are defined as 5% -30% of factor VIII activity. Treatment depends upon severity of condition. Mild cases may respond to desmopressin which promotes endogenous release of factor VIII.[4] Factor VIII activity and desmopressin induced rise in von Willebrand factor antigen were strong predictors of peak factor VIII levels. Mild to moderate cases can be managed with nasal spray of Desmopressin. Loomans J et al.[4] suggested that moderate to severe cases may require infusion of factor VIII. Recent development of long acting factor concentrates has promised treatment adherence and improvement in quality of life amongst hemophilia patients.[5] In conclusion, we report a case of Hemophilia A diagnosed in early infancy. Early diagnosis and treatment initiation of Hemophilia A may lead to decreased morbidity in the form of arthropathy. It may also help in preventing life threatening bleeds like CNS bleeds. Hence any bleeding tendencies in any age group, specifically early infancy should be evaluated thoroughly.

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