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CASE REPORT

Acquired Hemophilia 'A' – An Uncommon Condition Presented with Common Manifestations

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Abstract

Hemophilia A is usually a congenital bleeding disorder, which manifest in males due to its X linked recessive pattern of inheritance. It commonly presents with bruises, uncontrolled bleeding post minor trauma or post circumcision, knee swelling etc. Acquired hemophilia is a rare condition and could be associated with autoimmune diseases. We discuss this rare condition of acquired hemophilia in a female patient who presented with common manifestations but in unlikely host.

Keywords: Hemophilia A, Acquired haemophilia, Factor viii, Autoimmune disorder, AMA antibody

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ABBREVATIONS

AHA – ACQUIRED HEMOPHILIA A
SLE – SYSTEMIC LUPUS ERYTHMATOSUS
RA – RHEMATOID ARTHRITIS
APTT – ACTIVATED PARTIAL THROMBOPLASTIN TIME
PT – PROTHROMBIN TIME
TT – THROMBIN TIME
OPD – OUT PATIENT DEPARTMENT
CHD – CONGENITAL HEART DISEASES
VSD – VENTRICULAR SEPTAL DEFECT
TFT – THYROID FUNCTION TESTS
LFT – LIVER FUNCTION TESTS
KFT – KIDNEY FUNCTION TESTS
FFP – FRESH FROZEN PLASMA
PCV – PACKED RED CELL

Introduction

One of the rare autoimmune diseases, acquired hemophilia A (AHA) is associated with a high morbidity and is often missed by physicians due to its rare presentation. While hemophilia usually is a congenital condition and affects only males due to its X linked recessive pattern of inheritance. Rarely, AHA can also affect females and is usually associated with other auto immune conditions like SLE, RA.

AHA patients present with subcutaneous ecchymoses, joint hematomas, muscle and gum bleeding. A few cases of intracranial bleeds were also reported [1]. The key features for diagnosis include the classical clinical presentation and family history. Investigations show increased APTT, near normal or mildly deranged PT and TT, low levels of factor viii. Some cases even show decreased factor ix and VWF and Platelet count is usually normal [2].

Acquired Hemophilia differs from familial hemophilia with absence of family history of bleeding disorder and no specific age prevalence. High degree of suspicion and investigation into the cause of bleeding tendency is required to diagnose such cases.

Case presentation

A 19-year Female presented to outpatient department with chief complaint of painful swelling of right knee joint for two weeks, with petechial haemorrhages over forearm, abdomen and left lower limb. Patient also complained of fatigue, decreased appetite, and joint pains in the last 2 weeks (Figures 1 to 3).

Figure 1. 7- 10 days old bruise on left fore arm.

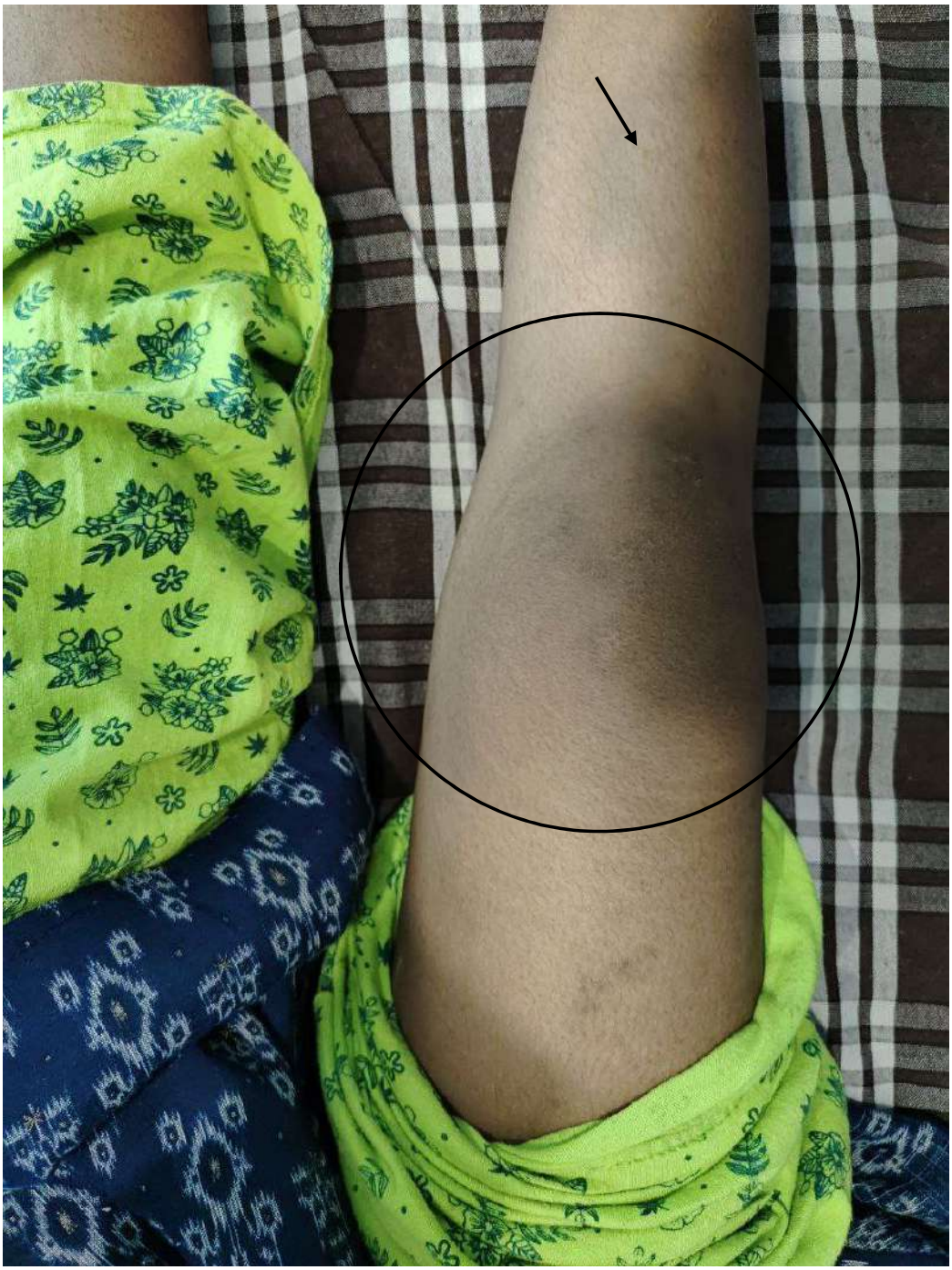

Figure 2. Knee joint bruse and heamarthrosis.

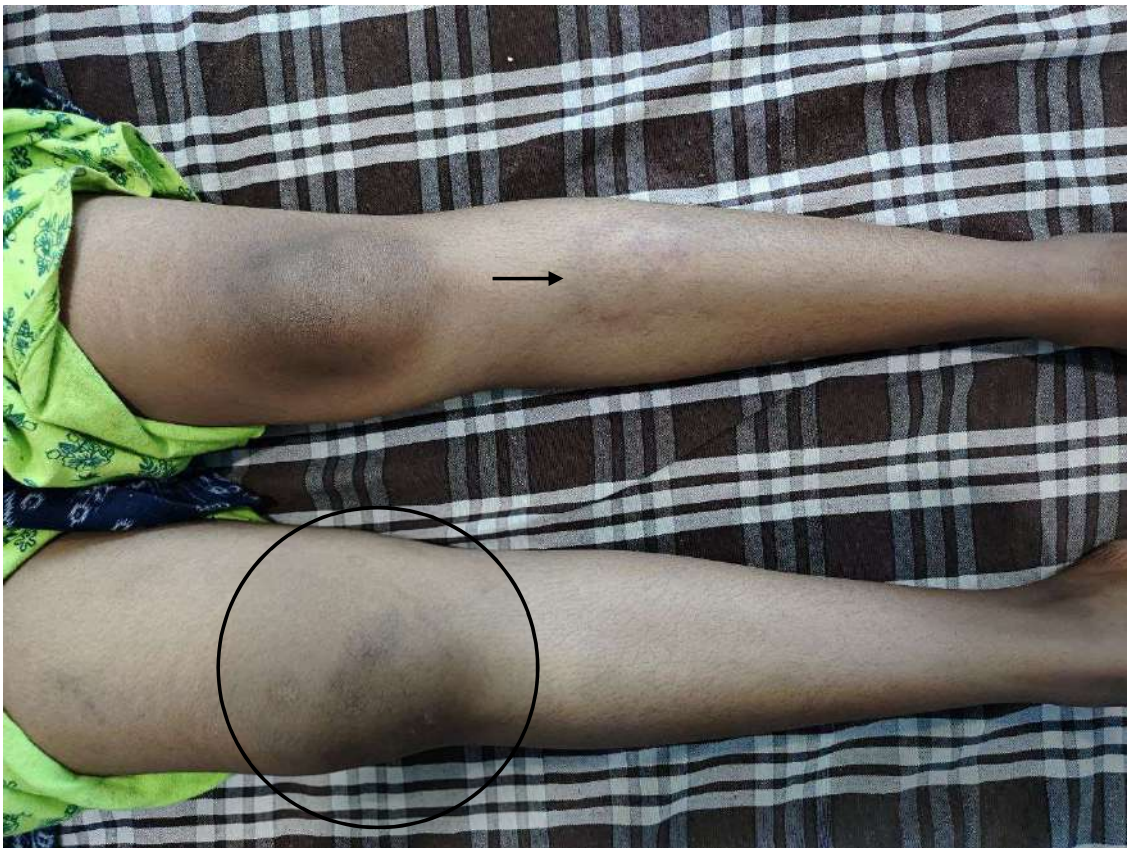

Figure 3. Limbs showing old bruises and knee heamarthrosis.

Patient had repeated bruises all over the body in the last 2 years. Patient had increased bleeding even with minor trauma like brushing teeth or any abrasions while playing outdoor games. Patient had moderate to severe bleeding during menses usually lasting for 5 – 7 days. Patient had visited many physicians and gynaecologists over the last one year for her increased bruises and menorrhagia without any relief.

Patient did not have any comorbidities and was not on any medication. Dietary history was unremarkable. No bowel and bladder abnormality noted. There was no history of any bleeding disorders in the family.

On general examination, patient was pale with multiple bruises over the body. Normal vital signs. Systemic examinations revealed no significant findings.

Lab reports revealed haemoglobin – 4.7g/dl, mcv – 72fl, platelet 1.4 lakh/ul. Prothrombin time – 23.6 sec, prothrombin index – 52.9%, INR – 1.88. APTT – 51.3 seconds.In suspicion of hemophilia, factor viii and xi has been paged which revealed factor viii < 0.70% and factor xi – 48.32%. An autoimmune panel showed AMA-M2 to be strongly positive. Other antibodies like Sm, SS – a/b, dsDNA, Jo1, scl 70 were negative.

TFT, RFT, LFT, serum electrolytes, urine routine were within normal limits. Ultrasonography abdomen was not contributory.

Patient had been treated with FFP and corticosteroids. Patient was given 4 PRC transfusions in view of severe anemia.

Patient was also given, calcium and vit d3 supplements, tranexamic acid 500mg, iv iron and b12 preparations. After the availability of factor VIII, 5 vials had been transfused. Intranasal desmopressin was started. Patient had been discharged on 10th day in stable condition. Decreased bruises and joint swelling seen although patient had pain in knee joint on weight bearing.

Repeated hemoglobin – 8.1 g/dl, prothrombin time is 18.2 seconds, and APTT is 40.3 seconds.

Discussion

Acquired hemophilia A is condition carrying high mortality and morbidity. Disease is characterized by formation of auto antibodies against coagulation factors, mostly against factor VIII. It carries about 8 – 20% mortality if not diagnosed and treated promptly. It might be associated with many autoimmune disorders like RA, SLE, dermatomyositis although some cases found no underlying conditions [3]. Our patient had AMA-M2 positivity which shows possibility of underlying autoimmunity.

Though, common clinical features include skin ecchymoses (post trauma, injection sites), and gastrointestinal bleeds. Surprisingly joint arthrosis is not as common as congenital hemophilia A. Our patient showed classical features of congenital hemophilia including hemarthrosis but turned out to be acquired hemophilia. The absence of positive family history and presenting age well beyond the typical years of hemophilia makes it difficult to suspect.

Severe Anaemia in our patient underlines the importance of early diagnosis and treatment. Minor bleeds can be treated conservatively with avoidance of any invasive procedures, and desmopressin supplements (0.3 – 0.5mg/kg) are helpful if factor viii level > 5%. But major bleeds need to be treated aggressively. If there are low titres of auto antibodies (<5BU), human factor VIII or desmopressin can be considered as first line treatment. But in cases where auto antibody titres are high, bypassing agents (activated prothrombin complex concentrates and recombinant activated factorVII) are preferred [4,5].

Conclusion

Any patient with bleeding tendency should be investigated and treated aggressively. Possibility of acquired hemophilia should be considered when the family history is absent. Association with other autoimmune conditions should be explored.

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Conflict of interest

The authors declare that they have no competing interests.

Ethics approval, Consent to participate, Consent to publish, Availability of data and material, Code availability

Not applicable

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