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Medicine

Autosomal Recessive Transmission of Hemophilia A: A Rare Case Report

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ABSTRACT A 25 years old female patient was admitted in this hospital for complaints of abdo pain, easy fatiguability and giddiness since 7 days. Patient was a known case of some haematological disorder since childhood and required blood transfusion for the same every 2 to 3 yearly. After detailed history taking and thoroughly investigating, she was found to have Hemophilia A with an Autosomal Recessive Pattern of Inheritance.

KEYWORDS :

INTRODUCTION:

Hemophilia A also known as Factor VIII deficiency is most commonly inherited as a x-linked recessive disease. So, females with this disease are rarely seen. This may happen if both the X chromosomes are affected; but then offspring survives for a very short period of life. However Hemophilia A can be seen in elderly females if it is inherited in a different fashion. Very few cases of females having Hemophilia A have been reported in the literature and due to its rarity we present a case of Hemophiliac female with Autosomal Recessive pattern of inheritance.

CASE DETAIL:

A 25 years female patient was admitted to the ward for complaints of abdominal pain, giddiness and easy fatiguability since 7 days. Patient had Chronic hematologic disease since childhood for which she required blood transfusion every 2-3 yearly. She had the habit of checking her Hemoglobin(Hb) at home which was 11g%, 7 days back. There was no any active bleeding from any site of body. She had no vomiting, loose motions or fever. On enquiry, she gave history of right hemorrhagic cyst 1 year back, which was managed conservatively. She had a very significant family history. Out of the 7 siblings, she was one of the three survivors. It was a 3rd degree consanguineous marriage between her parents. All the male siblings who died, had died while performing circumcision at the age of 2. Amongst the surviving siblings, her brother is affected by the disease and her sister is not. Her sister is married and has a daughter too.

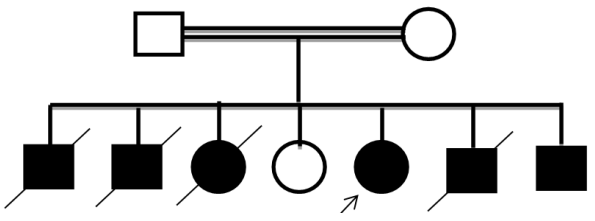

Diagram showing Autosomal Recessive pattern of inheritance in this patient.

Pic-1. Clinical photograph of the patient with Hemophilia A

Pic-2. Family photograph of the patient with her brother and sister (affected) and parents.

Examination revealed fully conscious patient with PR-72/min, RR-16/min, BP-110/70mm Hg , No signs of respiratory distress, No signs of CHF, Patient had severe abdominal tenderness in lower abdomen. Liver, spleen were not palpable. The cvs examination: normal heart sounds without murmur. Other systems were normal.

Her Investigations revealed Hb-4.3gm%, TLC- 8700/mm3, plt-2.4 lacs/mm3; rest blood indices were normal, liver function tests and kidney function tests were normal.

- Her serum LDH was raised- 380 U/L.
- Factor VIII:C Assay revealed very low levels-4.2%(50-150); and so was vonWillebrand Antigen-4.2%(50-200).
- Her Lupus Anticoagulant was negative.
- Serum β-hcg, α-feto protein were normal.
- ECG revealed sinus tachycardia.
- USG abdomen+pelvis revealed a 8.5 x 9cm hyperechoic lesion suggestive of a large pelvic hematoma, with free fluid in the abdomen.

Because of coagulopathy it was managed conservatively and the patient was discharged after giving packed cell transfusion till her Hb raised upto 8g%. With above history and investigations a diagnosis of Hemophilia A with Autosomal Recessive pattern of inheritance was made.

DISCUSSION:

Due to the X-linked recessive mode of inheritance, Hemophilia A, It usually affects males, and females are carriers who may pass the disease on their progeny. Thus females with Hemophilia A are rarely observed. However, there are few reported cases of Hemophilia A in females. In 1992, Matsushita T. et al reported a 2 year old girl having homozygous haemophilia A with hereditary coagulation factor XII deficiency[1]. Later in 1993, RJ Wise et al reported a case of Autosomal recessive transmission of haemophilia A due to a von Willebrand factor

mutation[2]. P. G. Mori et al also reported a female with haemophilia A who had an X isochromosome and it was not able to mask the abnormal gene[3]. One more such case of female hemophilia was published by Emily et al. and Morita et al[4,5]. Preethi S. Nair et al reported a case of homozygous female with haemophilia[6]. Again in 2013, Shukai et al reported a female patient with compound heterozygous Hemophilia A and identified a novel missense mutation, p.Met1092Ile[7].

SUMMARY:

The reported case had shown autosomal recessive pattern of inheritance as the disease was inherited in males as well as females and not all the individuals were affected. Thus we report this middle aged female patient of Hemophilia A with such an unusual pattern of inheritance.

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