MENORRHAGIA;

INHERITED FACTOR-VII DEFICIENCY IN A 12 YEARS OLD GIRL FROM KARACHI

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ABSTRACT….. Factor VII deficiency is a rare, autosomal recessive coagulopathy that becomes symptomatic in the form of a hemorrhagic syndrome characterized by severe life threatening bleeding. This may present in young women as severe anemia due to bleeding per vaginum. We report one such case of a 12 year old girl who presented at the gynecology outpatient department with complaints of severe menorrhagia at menarche. Her past history was consistent with episodes of profuse epistaxis and bleeding from gums. Her complete blood count showed severe anemia. Upon further investigation her prothrombin time was prolonged but her APTT was normal which was indicative of Factor VII deficiency and was confirmed by serum assays of Factor VII. It is important to diagnose this disorder earlier in order to avoid long term complications especially in women who may suffer from severe life threatening hemorrhage during menses or recurrent miscarriages during pregnancy. Therefore, our patient was transfused with packed cells and fresh frozen plasma immediately and started on low dose oestrogen and progesterone pill along with tranexemic acid to control her symptoms.

Key words: Blood Coagulation Tests, Female, Hypoprothrombinemias/complications.

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INTRODUCTION

Factor-VII (FVII) deficiency or hypoproconvertinemia is a rare cause of hereditary hemorrhagic disease. FVII is a vitamin K dependent factor with a short half life of 3 to 4 hours[1]. Clinical presentation of the disease varies in individuals and does not correlate with FVII levels which make it difficult to anticipate the severity of the disease and manage it accordingly (2). The disease is symptomatic in about 1 in 500,000 people but the prevalence is about 1 in 1,000,000[2,3].

We report one such case of a young girl with FVII deficiency who presented at our emergency department with hemorrhage at menarche and a review of management of such cases.

CASE

A 12 year old female, resident of Thatta was admitted in emergency room with complaint of Heavy menstrual cycles since 15 days. According to her mother, she had menarche 2 months back; the first cycle after menarche was normal, lasted for 5-7 days, the second cycle that started 15 days back was heavy with passage of clots. She was admitted in hospital, as her Haemoglobin dropped from 12 gm/dl to 5 gm/dl. She was transfused four units of packed cells and one unit of FFP. She was started on a Progesterone tablet but did not prove to be effective and she continued to bleed. She had one episode of syncope prior to her admission to the hospital.

Family history revealed that her parents had a consanguineous marriage. Among her seven siblings, out of which five are daughters, the patient is the second youngest in the family. There has been no menstrual or bleeding problem in the family .Patient was delivered at 36 weeks of gestation. Her mother had history of fall, followed by preterm premature rupture of membranes. At the time of delivery, there was difficulty in controlling bleeding from umbilical stump. The bleeding stopped spontaneously after 24 hours.

Past Medical history revealed that the patient had a heavy episode of epistaxis at the age of four years. She has difficulty in blood clotting after cuts and wounds and was advised to avoid falls, trips and lifting heavy weights. At the age of seven years, she had on and off

complains of bleeding from gums which resolved spontaneously.

During the course of Admission, the patient was investigated. Her prothrombin time (PT) was prolonged to 112 seconds and INR was also increased although her APTT was normal. Her thyroid profile was normal though clinically thyroid was mildly enlarged on palpation. Ultrasound of the pelvis showed slightly increased endometrial thickness.

Her case was discussed with physicians and haematologists and according to their advice, further investigations were carried out to find out the cause. Levels of VonWillebrand factor, Anti tissue transglutaminase IgG and IgA were done which came out to be normal. Mixing studies report showed correction with normal plasma indicated some factor deficiency and aged plasma but not with adsorbed plasma suggested factor VII deficiency.

Acquired causes of factor-VII deficiency include mainly liver disorders and vitamin K deficiency[2]. Patients LFT's were normal and she did not respond to injectible Vitamin-K. In addition her normal APTT levels, also confirmed the diagnosis of inherited factor VII deficiency.

As mixing studies are only a screening test, a Factor VII assay was done which showed decreased levels. The patient was labeled as a case of inherited factor VII deficiency.

Patient was transfused - PCV and FFP over all. She was discharged on low dose oestrogen and progesterone pill, but she did not respond effectively and continued to have on and off bleeding per vagina and was later on shifted to high dose oestrogen/progesterone pill ,along with tranexemic acid during the course of active bleeding.

DISCUSSION

Factor VII deficiency is classified into four clinical forms of progressively increasing severity ranging from an asymptomatic to a late onset: mild form, a severe hemorrhagic form to a severe life threatening form[2]. The disease is classified clinically since factor VII levels do not correlate with the severity of disease. As seen in our patient who was clinically classified to be of late onset mild form but had a factor VII level of 2.3% (normal 50 to 150%).

Since the bleeding tendency does not manifest clinically until levels fall < 10 IU/dl, because of higher levels of factor VII (upto 20 to 60 iu/dl)[3]. This was seen in our patient who had self resolving episodes of bleeding diathesis which became more severe at menarche.

To date, 36 cases of factor VII have been reported from Pakistan in a tricentre study conducted by the Aga Khan university[4,5,6]. Sajid et.al reported the prevalence of F VII in one study to be 0.4%[6]. The reason for a higher incidence of factor VII deficiency and other bleeding disorder in Pakistan seems to be due to the number of consanguious marriages as seen in our case.

As there is suitable way to predict the susceptibilty of hemorrhage, in asymptomatic individual management is mostly expectant. FFPs may be used but are not very effective due to the large volume that is required to replenish factor VII levels[1]. However as they readily available and is less thrombogenic, they are usually given at the time of presentation as was done in our case[7]. Also fatal thromboses have been reported due to hypercoagulability and needs serial monitoring[8]. It is for this reason our patient's coagulation screen was done during the course of administration of FFPs[9].

Lately, the treatment of choice for acute hemorrhage is replacement of factor VII due to the specificity of its action, it has significant benefit over FFPs and a low

thrombogenic risk[10,11]. However our patient did not receive.

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