LETTER TO EDITOR (VIEWER'S CHOICE)

CONGENITAL FACTOR XIII DEFICIENCY

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An eleven year old male child born of consanguineous marriage presented with pain in right lower abdomen and limping of right leg following trauma. On enquiring there was history of umbilical bleeding at birth, prolonged bleeding from cut injury, easy bruisability after trauma. He had received blood transfusion six times for bleeding in the past without any investigation. There was no history of similar complaints in other family members. On examination his general condition and vitals were stable, pallor was present. Ecchymosis over back, gluteal region and thighs was noticed. Movements at hip joint on right side were painful and restricted. All other systems were clinically normal. Complete blood count showed anemia. Coagulation screening tests were normal. Fibrinogen level was normal. Ultrasound abdomen showed right illiopsoas hematoma. Factor VIII and Factor IX levels were normal. Urea clot lyses showed abnormal solubility of clot in 5M urea. Factor XIII deficiency was thus confirmed. The child was given Fresh Frozen Plasma (FFP) and packed cell transfusion to treat anemia, and immunized with Hepatitis-B vaccine. He was advised to avoid NSAIDS, injuries and intramuscular injections. Child is doing well and receiving FFP every four weeks.

Congenital factor XIII deficiency, originally recognized by Duckert in 1960, is a rare autosomal recessive disease usually associated with a severe bleeding diathesis (1). The incidence of this disease is about 1 case per 2-5 million population. The male to female ratio is 1:1. (1) The mortality and morbidity are primarily related to bleeding. Intracranial hemorrhage can be life threatening. The diagnosis is made at an early age, often during infancy because the clinical bleeding is severe in most patients with Factor XIII deficiency. Bleeding from the stump of the umbilical cord within the first days to weeks of life is a characteristic feature that occurs in 80% of affected individuals, bleeding from this specific site is uncommon in other inherited haemostatic diseases except afibrinogenemia. (2,3) Other clinical features include CNS hemorrhage (25-30%), Hemarthroses (20%), soft tissue bleeding and bruising. Recurrent spontaneous abortions are very common in women with FXIII deficiency, who do not receive FXIII replacement. (4,5) Delayed bleeding after trauma or surgery in pathognomic of FXIII deficiency. Wound healing is abnormal in a subset of patients. Although this child had features of Factor XIII deficiency since birth he was not diagnosed due to lack of facilities. Diagnosis is done by screening tests, functional assays, immunologic assays and genotyping. Appropriate imaging studies are required in the evaluation of suspected bleeding for instance CT scan in a patient with suspected CNS hemorrhage. Plasma, cryoprecipitate, and Factor XIII concentrates are used to treat bleeding in Factor XIII deficiency. The treatment of choice is plasma derived FXIII concentrate that is pasteurized to provide virologic safety and is less likely than plasma to cause systemic reactions (2). In our patient due to financial constraints we treated patient with fresh frozen plasma.

Prophylaxis is the management strategy of choice. Factor XIII concentrate 10-20U/kg every 4-6 weeks provides adequate plasma levels in most patients. (6) Genetic counseling and family studies should be part of a complete evaluation.

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