HYPOFIBRINOGENEMIA

(A Report of Two Cases)

Maj HARSH KUMAR*, Surg Cdr P SIVADAS+,
Brig C LAKSHMANAN#, Col JR BHARDWAJ**

ABSTRACT

Hypofibrinogenemia is an uncommon bleeding disorder. Two siblings with mild bleeding diathesis were investigated and diagnosed as suffering from this disorder. The cases are discussed keeping in view of their rarity.

MJAFI 1994; 50 : 291-292

KEY WORDS : Hypofibrinogenemia; Bleeding diathesis.

Introduction

Congenital hypofibrinogenemia is a rare disorder which was first described in 1935, and since then about 40 cases have been reported [1]. The patient may show no apparent clinical abnormality or may present with ecchymoses, subcutaneous haematomas or haemarthroses [1-5]. We describe here two cases of congenital hypofibrinogenemia in siblings.

Case Reports

Two male children aged 9 months and 2 years were admitted with complaints of multiple small nodules all over the body, first observed in both siblings at about two months of age. These lesions were bluish red, came in crops and were at times associated with trauma. There was no history of epistaxis, haematuria, melena or joint pains. Parents and close blood relations had no such complaints. There was no history of consanguinity.

On examination, the lesions were bluish-red, 1-2 cm in diameter and slightly nodular. They were firm and slightly tender. There was no lymphadenopathy. Pallor and mild splenomegaly were present. There were no other significant clinical findings.

Investigations revealed a hypochromic microcytic anaemia in both cases. The bleeding and clotting time and platelet counts were normal. Clot retraction test was performed and the clot that formed was small. Factor XIII screening using 5M urea solution showed no abnormality. Fibrinogen was estimated by the method described by Goodwin *et al* [6]. The patient was not on any drugs during the period of investigation. The coagulation profile is depicted below:-

Test done	Sibling 1	Sibling 2
Platelet count	300,000/cmm	250,000/cmm
Prothrombin time	C=13, T=16 sec	C=13, T=16 sec
PTTK	C=35, T=50 sec	C=35, T=44 sec
Thrombin time	C=10, T=15 sec	C=10, T=14 sec
Fibrinogen level	80 mg/dl	96 mg/dl
F & FDP	Normal	Normal

Discussion

Hypofibrinogenemia is a rare clinical disorder. The coagulation tests in hypofibrinogenemia are abnormal depending upon the fibrinogen levels [1,5]. The fibrinogen levels range between 20 - 100 mg/dl [2]. In both siblings, the fibrinogen levels were less than 100 mg/dl, the partial thromboplastin time with kaolin (PTTK) and the thrombin time (TT) were prolonged. The diagnosis was established by fibrinogen assay [6,7].

Cases of hypofibrinogenemia present with mild spontaneous bleed, ecchymoses, subcutaneous haematomas or first trimester abortions. Sometimes there may be no clinical manifestations [1-5]. The two siblings had presented clinically with multiple small bluish red nodules since 2 months of age. Severe bleeding manifestations like haemarthroses, melena and haematuria were not seen. The

*Reader, +Associate Professor, #ADMS, HQ(WC), Chandimandir; **Professor & Head, Dept of Pathology, AFMC, Pune 411 040.

parents of the siblings did not have a consanguinous marriage, as might be expected in 50% of cases [1]. Furthermore, no history of bleeding diathesis in the previous generation or near relatives was present.

Hypofibrinogenemia is rare in clinical practice and can be confused with mild haemophilia because of similar clinical manifestations. Cases with undiagnosed bleeding diathesis should be carefully and systematically investigated so that they are correctly identified and managed.

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